

Patient Name \_\_\_\_\_

Date      

## CACT - Intake Demographics

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NBSTRN ID \_\_\_\_\_

Version 2.1.2

Center name

 A  B  C  D  E  F  G  H  I  J  K  L M  N  O  P  Q  R  S  T  U  V  OtherIntake date      

### Consent

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Consent obtained  Yes  No  IRB ExemptAssent obtained  Yes  NoType of assent  Written  VerbalPermission to recontact  Unknown  Yes  NoProtocol ID  A

### Demographics Information

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Patient last name \_\_\_\_\_

Patient first name \_\_\_\_\_

Date of birth      

Age \_\_\_\_\_

Gestational age in weeks \_\_\_\_\_

Societal sex  Unknown  Male  Female

Biological sex

- Not tested  Unknown  XX genotype/Female
- XY genotype/Male  XXX Triple X syndrome  XXY Klinefelter's syndrome
- XO Turner's syndrome  XXXY syndrome  XXYY syndrome
- Mosaic including XXXXY  Penta X syndrome  Other

Biological sex-other, specify \_\_\_\_\_

Biological mother's maiden name \_\_\_\_\_

Zip code \_\_\_\_\_

### Condition

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Patient condition category  Fatty acid oxidation disorders

Specify fatty acid oxidation disorder diagnosis for the patient

 Carnitine acylcarnitine translocase deficiency (CACT)

Patient disorder identification method

Patient Name \_\_\_\_\_

Date       Unknown  Abnormal newborn screen  Abnormal labs Clinical presentation  Family member with this condition

Family member with this condition

 Biological mother  Biological father  Full sibling  Half sibling  Other

Family member with this condition \_\_\_\_\_

**Care and Other Studies**

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Miles from home to primary care \_\_\_\_\_

Miles from home to specialty care \_\_\_\_\_

Specify type of primary care provider  Unknown  Family practice  Internal medicine  Pediatrics

Name of primary care provider \_\_\_\_\_

Specify medical home  Unknown  None  Primary care center  Speciality care center  Other

Specify medical home-other, specify \_\_\_\_\_

Patient is in other research studies  Unknown  Yes  NoOther research studies are clinical trials  Unknown  Yes  No

Research study-other, specify \_\_\_\_\_

Clinicaltrials.gov identifier \_\_\_\_\_

**Education**

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Maternal education

 Unknown 8th grade/less 9th-12th grade, no diploma High school graduate or GED completed Some college credit but no degree Associate degree (e.g., AA, AS) Bachelor's degree (e.g., BA, AB, BS) Master's degree (e.g., MA, MS, MEng, MEd, MSW, MBA) Doctorate (e.g., PhD, EdD) or Professional degree (e.g., MD, DDS, DVM, LLB, JD)

Paternal education

 Unknown 8th grade/less 9th-12th grade, no diploma High school graduate or GED completed Some college credit but no degree Associate degree (e.g., AA, AS) Bachelor's degree (e.g., BA, AB, BS)

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- Master's degree (e.g., MA, MS, MEng, MEd, MSW, MBA)
- Doctorate (e.g., PhD, EdD) or Professional degree (e.g., MD, DDS, DVM, LLB, JD)

## Patient education

- Unknown
- 8th grade/less
- 9th-12th grade, no diploma
- High school graduate or GED completed
- Some college credit but no degree
- Associate degree (e.g., AA, AS)
- Bachelor's degree (e.g., BA, AB, BS)
- Master's degree (e.g., MA, MS, MEng, MEd, MSW, MBA)
- Doctorate (e.g., PhD, EdD) or Professional degree (e.g., MD, DDS, DVM, LLB, JD)

Special education services received prior to intake  Unknown  Yes  No

## Age patient qualified for special education services

- Unknown  <1  1 year  2 years  3 years  4 years  5 years  6 years
- 7 years  8 years  9 years  10 years  11 years  12 years  13 years  14 years
- 15 years  16 years  17 years  18 years

**Ancestral Origin, Race and Ethnicity**Ancestral Origin  Unknown  Africa  Asia  Europe  North America  South America  Oceania  Other

## Ancestral Origin-Africa

- Unknown  Egypt  Eritrea  Ethiopia  Liberia  Somalia  South Africa
- Other

Ancestral Origin-Africa-Other, specify \_\_\_\_\_

## Ancestral Origin-Asia

- Unknown  Bhutan  China  Hmong
- India  Israel  Japan  Jordan
- Korea-North  Korea-South  Laos  Lebanon
- Palestinian territories  Pakistan  Philippines  Russian Federation
- Syria  Thailand  Vietnam  Other

Ancestral Origin-Asia-Other, specify \_\_\_\_\_

## Ancestral Origin-Europe

- Unknown  Austria  Belgium  Bulgaria  Croatia
- Czech Republic  Denmark  Finland  France  Germany
- Greece  Hungary  Iceland  Ireland  Italy
- Lithuania  Malta  Netherlands  Norway  Poland

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- Romania       Serbia       Slovakia       Slovenia       Spain  
 Sweden       Switzerland       Ukraine       United Kingdom       Other

Ancestral Origin-Europe-Italy     Unknown     SicilyAncestral Origin-Europe-Romania     Unknown     TransylvaniaAncestral Origin-Europe-United Kingdom     Unknown     England     Northern Ireland     Scotland     Wales

Ancestral Origin-Europe-Other, specify \_\_\_\_\_

Ancestral Origin-North America

- Unknown       Aleutian Islands       Canada       Dominican Republic     Honduras  
 Mexico       Puerto Rico       United States       Other

Ancestral Origin-North America-Canada     Unknown     French Canadian

Ancestral Origin-North America-Other, specify \_\_\_\_\_

Ancestral Origin-South America     Unknown     Colombia     Venezuela     Other

Ancestral Origin-South America-Other, specify \_\_\_\_\_

Ancestral Origin-Oceania     Unknown     Australia     Other

Ancestral Origin-Oceania-Other, specify \_\_\_\_\_

Ancestral Origin-Other     Unknown     Amish     Arabic     Hutterite     Mennonite     Jewish     OtherAncestral Origin-Other-Jewish     Unknown     Ashkenazic     Sephardic

Ancestral Origin-Other, specify \_\_\_\_\_

Race

- Not reported       American Indian/Alaskan Native  
 Asian       Black or African American  
 Native Hawaiian or Other Pacific Islander     White

Race-American Indian/Alaskan Native     Aleutian     Cherokee     Other

Race-American Indian/Alaskan Native-Other, specify \_\_\_\_\_

Patient is Hispanic or Latino     Not reported     Yes     No**Socioeconomics**

Maternal age (in years) at patient's birth \_\_\_\_\_

Paternal age (in years) at patient's birth \_\_\_\_\_

Mother's marital status at patient's birth

- Unknown       Married       Widowed       Divorced       Separated  
 Never married       Living with partner

County mother resides in at patient's birth \_\_\_\_\_

State mother resides in at patient's birth

- Unknown       Not Applicable     AL       AK       AZ       AR  
 CA       CO       CT       DE       DC       FL

Patient Name _____
Date <input type="text" value="M"/> <input type="text" value="M"/> <input type="text" value="D"/> <input type="text" value="D"/> <input type="text" value="Y"/> <input type="text" value="Y"/>

- |                          |                          |                          |                          |                          |                          |
|--------------------------|--------------------------|--------------------------|--------------------------|--------------------------|--------------------------|
| <input type="radio"/> GA | <input type="radio"/> HI | <input type="radio"/> ID | <input type="radio"/> IL | <input type="radio"/> IN | <input type="radio"/> IA |
| <input type="radio"/> KS | <input type="radio"/> KY | <input type="radio"/> LA | <input type="radio"/> ME | <input type="radio"/> MD | <input type="radio"/> MA |
| <input type="radio"/> MI | <input type="radio"/> MN | <input type="radio"/> MS | <input type="radio"/> MO | <input type="radio"/> MT | <input type="radio"/> NE |
| <input type="radio"/> NV | <input type="radio"/> NH | <input type="radio"/> NJ | <input type="radio"/> NM | <input type="radio"/> NY | <input type="radio"/> NC |
| <input type="radio"/> ND | <input type="radio"/> OH | <input type="radio"/> OK | <input type="radio"/> OR | <input type="radio"/> PA | <input type="radio"/> PR |
| <input type="radio"/> RI | <input type="radio"/> SC | <input type="radio"/> SD | <input type="radio"/> TN | <input type="radio"/> TX | <input type="radio"/> UT |
| <input type="radio"/> VT | <input type="radio"/> VA | <input type="radio"/> WA | <input type="radio"/> WV | <input type="radio"/> WI | <input type="radio"/> WY |

### Medical Coverage

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Medical coverage at time of intake

- Unknown
- None
- Commercial/private
- Medicaid
- Medicare
- Military
- Newborn screening funds
- Patient assistance program
- State Children's Health Insurance Program (SCHIP)
- State Children with Special Health Needs (CSHN) Program
- Other

Medical coverage at intake-Patient assistance program, specify \_\_\_\_\_

Medical coverage at intake-Other, specify \_\_\_\_\_

### Language

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Primary language spoken at home  Unknown  English  Non-English  Both

Identify Non-English language(s) spoken at home  Arabic  Hmong  Polish  Somalian  Spanish  Other

Identify Non-English language(s) spoken at home-other, specify \_\_\_\_\_

Written/web-based information on this condition provided to the patient/primary caregiver in his/her primary language

- Unknown  Yes  No

### Comments

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Intake demographics comments

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## CACT - Intake Family History

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### Family History

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Consanguinity Unknown Yes NoPatient was adopted Unknown Yes No

Number of pregnancies for patient's mother \_\_\_\_\_

Number of live births for patient's mother \_\_\_\_\_

### Siblings

Number of biological siblings in the patient's family \_\_\_\_\_

Sibling 1: Sibling type Full HalfSibling 1: Half Maternal PaternalSibling 1: Gender Unknown Male Female

Sibling 1: Year of birth \_\_\_\_\_

Sibling 1: Affected with this condition Unknown Yes NoSibling 1: Enrolled in this study Unknown Yes No

Sibling 1: NBSTRN ID for this study \_\_\_\_\_

Sibling 1: Method of diagnosis

Unknown Clinical exam Diagnostic test(s) Newborn screenPatient/parent report Prenatal testing Other

Sibling 1: Method of diagnosis-other, specify \_\_\_\_\_

Sibling 1: Newborn screen performed for this condition Unknown Yes No

Sibling 1: Results of newborn screening for this condition

Unavailable Presumptive positive Negative/Normal BorderlineSibling 1: Diagnostic tests performed for this condition Unknown Yes No

Sibling 1: Specify the type of diagnostic tests performed

Unknown Biochemical Molecular Imaging Other

Sibling 1: Diagnostic tests performed-other, specify \_\_\_\_\_

Sibling 1: Deceased Unknown Yes No

Sibling 1: Age of death (in years) \_\_\_\_\_

Sibling 1: Timing of diagnosis Unknown Pre-mortem Post-mortemSibling 2: Sibling type Full HalfSibling 2: Half Maternal PaternalSibling 2: Gender Unknown Male Female

Sibling 2: Year of birth \_\_\_\_\_

Sibling 2: Affected with this condition Unknown Yes No

Patient Name \_\_\_\_\_

Date      Sibling 2: Enrolled in this study  Unknown  Yes  No

Sibling 2: NBSTRN ID for this study \_\_\_\_\_

Sibling 2: Method of diagnosis

Unknown  Clinical exam  Diagnostic test(s)  Newborn screen  
 Patient/parent report  Prenatal testing  Other

Sibling 2: Method of diagnosis-other, specify \_\_\_\_\_

Sibling 2: Newborn screen performed for this condition  Unknown  Yes  No

Sibling 2: Results of newborn screening for this condition

Unavailable  Presumptive positive  Negative/Normal  Borderline

Sibling 2: Diagnostic tests performed for this condition  Unknown  Yes  No

Sibling 2: Specify the type of diagnostic tests performed

Unknown  Biochemical  Molecular  Imaging  Other

Sibling 2: Diagnostic tests performed-other, specify \_\_\_\_\_

Sibling 2: Deceased  Unknown  Yes  No

Sibling 2: Age of death (in years) \_\_\_\_\_

Sibling 2: Timing of diagnosis  Unknown  Pre-mortem  Post-mortemSibling 3: Sibling type  Full  HalfSibling 3: Half  Maternal  PaternalSibling 3: Gender  Unknown  Male  Female

Sibling 3: Year of birth \_\_\_\_\_

Sibling 3: Affected with this condition  Unknown  Yes  NoSibling 3: Enrolled in this study  Unknown  Yes  No

Sibling 3: NBSTRN ID for this study \_\_\_\_\_

Sibling 3: Method of diagnosis

Unknown  Clinical exam  Diagnostic test(s)  Newborn screen  
 Patient/parent report  Prenatal testing  Other

Sibling 3: Method of diagnosis-other, specify \_\_\_\_\_

Sibling 3: Newborn screen performed for this condition  Unknown  Yes  No

Sibling 3: Results of newborn screening for this condition

Unavailable  Presumptive positive  Negative/Normal  Borderline

Sibling 3: Diagnostic tests performed for this condition  Unknown  Yes  No

Sibling 3: Specify the type of diagnostic tests performed

Unknown  Biochemical  Molecular  Imaging  Other

Sibling 3: Diagnostic tests performed-other, specify \_\_\_\_\_

Sibling 3: Deceased  Unknown  Yes  No

Patient Name \_\_\_\_\_

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Sibling 3: Age of death (in years) \_\_\_\_\_

Sibling 3: Timing of diagnosis  Unknown  Pre-mortem  Post-mortemSibling 4: Sibling type  Full  HalfSibling 4: Half  Maternal  PaternalSibling 4: Gender  Unknown  Male  Female

Sibling 4: Year of birth \_\_\_\_\_

Sibling 4: Affected with this condition  Unknown  Yes  NoSibling 4: Enrolled in this study  Unknown  Yes  No

Sibling 4: NBSTRN ID for this study \_\_\_\_\_

Sibling 4: Method of diagnosis

 Unknown  Clinical exam  Diagnostic test(s)  Newborn screen Patient/parent report  Prenatal testing  Other

Sibling 4: Method of diagnosis-other, specify \_\_\_\_\_

Sibling 4: Newborn screen performed for this condition  Unknown  Yes  No

Sibling 4: Results of newborn screening for this condition

 Unavailable  Presumptive positive  Negative/Normal  BorderlineSibling 4: Diagnostic tests performed for this condition  Unknown  Yes  No

Sibling 4: Specify the type of diagnostic tests performed

 Unknown  Biochemical  Molecular  Imaging  Other

Sibling 4: Diagnostic tests performed-other, specify \_\_\_\_\_

Sibling 4: Deceased  Unknown  Yes  No

Sibling 4: Age of death (in years) \_\_\_\_\_

Sibling 4: Timing of diagnosis  Unknown  Pre-mortem  Post-mortemSibling 5: Sibling type  Full  HalfSibling 5: Half  Maternal  PaternalSibling 5: Gender  Unknown  Male  Female

Sibling 5: Year of birth \_\_\_\_\_

Sibling 5: Affected with this condition  Unknown  Yes  NoSibling 5: Enrolled in this study  Unknown  Yes  No

Sibling 5: NBSTRN ID for this study \_\_\_\_\_

Sibling 5: Method of diagnosis

 Unknown  Clinical exam  Diagnostic test(s)  Newborn screen Patient/parent report  Prenatal testing  Other

Sibling 5: Method of diagnosis-other, specify \_\_\_\_\_

Sibling 5: Newborn screen performed for this condition  Unknown  Yes  No



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Sibling 5: Results of newborn screening for this condition

 Unavailable       Presumptive positive     Negative/Normal       BorderlineSibling 5: Diagnostic tests performed for this condition     Unknown     Yes     No

Sibling 5: Specify the type of diagnostic tests performed

 Unknown       Biochemical       Molecular       Imaging       Other

Sibling 5: Diagnostic tests performed-other, specify \_\_\_\_\_

Sibling 5: Deceased     Unknown     Yes     No

Sibling 5: Age of death (in years) \_\_\_\_\_

Sibling 5: Timing of diagnosis     Unknown     Pre-mortem     Post-mortemSibling 6: Sibling type     Full     HalfSibling 6: Half     Maternal     PaternalSibling 6: Gender     Unknown     Male     Female

Sibling 6: Year of birth \_\_\_\_\_

Sibling 6: Affected with this condition     Unknown     Yes     NoSibling 6: Enrolled in this study     Unknown     Yes     No

Sibling 6: NBSTRN ID for this study \_\_\_\_\_

Sibling 6: Method of diagnosis

 Unknown       Clinical exam       Diagnostic test(s)       Newborn screen  
 Patient/parent report     Prenatal testing       Other

Sibling 6: Method of diagnosis-other, specify \_\_\_\_\_

Sibling 6: Newborn screen performed for this condition     Unknown     Yes     No

Sibling 6: Results of newborn screening for this condition

 Unavailable       Presumptive positive     Negative/Normal       BorderlineSibling 6: Diagnostic tests performed for this condition     Unknown     Yes     No

Sibling 6: Specify the type of diagnostic tests performed

 Unknown       Biochemical       Molecular       Imaging       Other

Sibling 6: Diagnostic tests performed-other, specify \_\_\_\_\_

Sibling 6: Deceased     Unknown     Yes     No

Sibling 6: Age of death (in years) \_\_\_\_\_

Sibling 6: Timing of diagnosis     Unknown     Pre-mortem     Post-mortemSibling 7: Sibling type     Full     HalfSibling 7: Half     Maternal     PaternalSibling 7: Gender     Unknown     Male     Female

Sibling 7: Year of birth \_\_\_\_\_

Sibling 7: Affected with this condition     Unknown     Yes     No

Patient Name \_\_\_\_\_

Date      Sibling 7: Enrolled in this study  Unknown  Yes  No

Sibling 7: NBSTRN ID for this study \_\_\_\_\_

Sibling 7: Method of diagnosis

Unknown  Clinical exam  Diagnostic test(s)  Newborn screen  
 Patient/parent report  Prenatal testing  Other

Sibling 7: Method of diagnosis-other, specify \_\_\_\_\_

Sibling 7: Newborn screen performed for this condition  Unknown  Yes  No

Sibling 7: Results of newborn screening for this condition

Unavailable  Presumptive positive  Negative/Normal  Borderline

Sibling 7: Diagnostic tests performed for this condition  Unknown  Yes  No

Sibling 7: Specify the type of diagnostic tests performed

Unknown  Biochemical  Molecular  Imaging  Other

Sibling 7: Diagnostic tests performed-other, specify \_\_\_\_\_

Sibling 7: Deceased  Unknown  Yes  No

Sibling 7: Age of death (in years) \_\_\_\_\_

Sibling 7: Timing of diagnosis  Unknown  Pre-mortem  Post-mortemSibling 8: Sibling type  Full  HalfSibling 8: Half  Maternal  PaternalSibling 8: Gender  Unknown  Male  Female

Sibling 8: Year of birth \_\_\_\_\_

Sibling 8: Affected with this condition  Unknown  Yes  NoSibling 8: Enrolled in this study  Unknown  Yes  No

Sibling 8: NBSTRN ID for this study \_\_\_\_\_

Sibling 8: Method of diagnosis

Unknown  Clinical exam  Diagnostic test(s)  Newborn screen  
 Patient/parent report  Prenatal testing  Other

Sibling 8: Method of diagnosis-other, specify \_\_\_\_\_

Sibling 8: Newborn screen performed for this condition  Unknown  Yes  No

Sibling 8: Results of newborn screening for this condition

Unavailable  Presumptive positive  Negative/Normal  Borderline

Sibling 8: Diagnostic tests performed for this condition  Unknown  Yes  No

Sibling 8: Specify the type of diagnostic tests performed

Unknown  Biochemical  Molecular  Imaging  Other

Sibling 8: Diagnostic tests performed-other, specify \_\_\_\_\_

Sibling 8: Deceased  Unknown  Yes  No

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Sibling 8: Age of death (in years) \_\_\_\_\_

Sibling 8: Timing of diagnosis  Unknown  Pre-mortem  Post-mortem**Parents**Biological mother: Affected with this condition  Unknown  Yes  NoBiological mother: Enrolled in this study  Unknown  Yes  No

Biological mother: NBSTRN ID for this study \_\_\_\_\_

Biological mother: Method of diagnosis

 Unknown  Clinical exam  Diagnostic test(s)  Newborn screen Patient/parent report  Prenatal testing  Other

Biological mother: Method of diagnosis-other, specify \_\_\_\_\_

Biological mother: Newborn screen performed for this condition  Unknown  Yes  No

Biological mother: Results of newborn screening for this condition

 Unavailable  Presumptive positive  Negative/Normal  BorderlineBiological mother: Diagnostic tests performed for this condition  Unknown  Yes  No

Biological mother: Specify the type of diagnostic tests performed

 Unknown  Biochemical  Molecular  Imaging  Other

Biological mother: Diagnostic tests performed-other, specify \_\_\_\_\_

Biological mother: Deceased  Unknown  Yes  No

Biological mother: Age of death (in years) \_\_\_\_\_

Biological mother: Timing of diagnosis  Unknown  Pre-mortem  Post-mortemBiological father: Affected with this condition  Unknown  Yes  NoBiological father: Enrolled in this study  Unknown  Yes  No

Biological father: NBSTRN ID for this study \_\_\_\_\_

Biological father: Method of diagnosis

 Unknown  Clinical exam  Diagnostic test(s)  Newborn screen Patient/parent report  Prenatal testing  Other

Biological father: Method of diagnosis-other, specify \_\_\_\_\_

Biological father: Newborn screen performed for this condition  Unknown  Yes  No

Biological father: Results of newborn screening for this condition

 Unavailable  Presumptive positive  Negative/Normal  BorderlineBiological father: Diagnostic tests performed for this condition  Unknown  Yes  No

Biological father: Specify the type of diagnostic tests performed

 Unknown  Biochemical  Molecular  Imaging  Other

Biological father: Diagnostic tests performed-other, specify \_\_\_\_\_

Biological father: Deceased  Unknown  Yes  No

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Biological father: Age of death (in years) \_\_\_\_\_

Biological father: Timing of diagnosis    Unknown    Pre-mortem    Post-mortem**Grandparents**Maternal grandmother: Affected with this condition    Unknown    Yes    NoMaternal grandmother: Enrolled in this study    Unknown    Yes    No

Maternal grandmother: NBSTRN ID for this study \_\_\_\_\_

Maternal grandmother: Method of diagnosis

 Unknown                       Clinical exam                       Diagnostic test(s)                       Newborn screen Patient/parent report    Prenatal testing                       Other

Maternal grandmother: Method of diagnosis-other, specify \_\_\_\_\_

Maternal grandmother: Diagnostic tests performed for this condition    Unknown    Yes    No

Maternal grandmother: Specify the type of diagnostic tests performed

 Unknown                       Biochemical                       Molecular                       Imaging                       Other

Maternal grandmother: Diagnostic tests performed-other, specify \_\_\_\_\_

Maternal grandmother: Deceased    Unknown    Yes    No

Maternal grandmother: Age of death (in years) \_\_\_\_\_

Maternal grandmother: Timing of diagnosis    Unknown    Pre-mortem    Post-mortemMaternal grandfather: Affected with this condition    Unknown    Yes    NoMaternal grandfather: Enrolled in this study    Unknown    Yes    No

Maternal grandfather: NBSTRN ID for this study \_\_\_\_\_

Maternal grandfather: Method of diagnosis

 Unknown                       Clinical exam                       Diagnostic test(s)                       Newborn screen Patient/parent report    Prenatal testing                       Other

Maternal grandfather: Method of diagnosis-other, specify \_\_\_\_\_

Maternal grandfather: Diagnostic tests performed for this condition    Unknown    Yes    No

Maternal grandfather: Specify the type of diagnostic tests performed

 Unknown                       Biochemical                       Molecular                       Imaging                       Other

Maternal grandfather: Diagnostic tests performed-other, specify \_\_\_\_\_

Maternal grandfather: Deceased    Unknown    Yes    No

Maternal grandfather: Age of death (in years) \_\_\_\_\_

Maternal grandfather: Timing of diagnosis    Unknown    Pre-mortem    Post-mortemPaternal grandmother: Affected with this condition    Unknown    Yes    NoPaternal grandmother: Enrolled in this study    Unknown    Yes    No

Paternal grandmother: NBSTRN ID for this study \_\_\_\_\_

Paternal grandmother: Method of diagnosis

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/>   <input type="text"/> <input type="text"/> <input type="text"/>   <input type="text"/> <input type="text"/> <input type="text"/>
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- Unknown             Clinical exam             Diagnostic test(s)             Newborn screen  
 Patient/parent report    Prenatal testing             Other

Paternal grandmother: Method of diagnosis-other, specify \_\_\_\_\_

Paternal grandmother: Diagnostic tests performed for this condition    Unknown    Yes    No

Paternal grandmother: Specify the type of diagnostic tests performed

- Unknown             Biochemical             Molecular             Imaging             Other

Paternal grandmother: Diagnostic tests performed-other, specify \_\_\_\_\_

Paternal grandmother: Deceased    Unknown    Yes    No

Paternal grandmother: Age of death (in years) \_\_\_\_\_

Paternal grandmother: Timing of diagnosis    Unknown    Pre-mortem    Post-mortem

Paternal grandfather: Affected with this condition    Unknown    Yes    No

Paternal grandfather: Enrolled in this study    Unknown    Yes    No

Paternal grandfather: NBSTRN ID for this study \_\_\_\_\_

Paternal grandfather: Method of diagnosis

- Unknown             Clinical exam             Diagnostic test(s)             Newborn screen  
 Patient/parent report    Prenatal testing             Other

Paternal grandfather: Method of diagnosis-other, specify \_\_\_\_\_

Paternal grandfather: Diagnostic tests performed for this condition    Unknown    Yes    No

Paternal grandfather: Specify the type of diagnostic tests performed

- Unknown             Biochemical             Molecular             Imaging             Other

Paternal grandfather: Diagnostic tests performed-other, specify \_\_\_\_\_

Paternal grandfather: Deceased    Unknown    Yes    No

Paternal grandfather: Age of death (in years) \_\_\_\_\_

Paternal grandfather: Timing of diagnosis    Unknown    Pre-mortem    Post-mortem

**Other Family**

Other affected family members NOT listed above    Unknown    Yes    No

Number of other affected family members that are NOT listed above \_\_\_\_\_

Relationship to affected family member 1 \_\_\_\_\_

Relationship to affected family member 2 \_\_\_\_\_

Relationship to affected family member 3 \_\_\_\_\_

Relationship to affected family member 4 \_\_\_\_\_

Relationship to affected family member 5 \_\_\_\_\_

Relationship to affected family member 6 \_\_\_\_\_

Relationship to affected family member 7 \_\_\_\_\_

Relationship to affected family member 8 \_\_\_\_\_

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Relationship to affected family member 9 \_\_\_\_\_

Relationship to affected family member 10 \_\_\_\_\_

**Comments**

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Intake family history comments

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## CACT - Intake Past Health History

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### Prenatal History

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Prenatal diagnosis done for this condition    Unknown    Yes    No

Form of prenatal diagnosis

- Unknown                       Amniocentesis                       Chorionic villus sampling (CVS)  
 Fetal blood

Amniocentesis diagnosis    Biochemical/enzyme    DNA

Chorionic villus sampling (CVS) diagnosis    Biochemical/enzyme    DNA

Issues concerning mother's pregnancy with this patient

- Unknown                       None                       Pregnancy complications  
 Assisted reproduction

Pregnancy complications/risk factors

- Unknown  
 Acute fatty liver of pregnancy (AFLP)  
 Advanced maternal age (35+ years of age)  
 Ectopic pregnancy  
 Gestational diabetes  
 Group B strep  
 Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)  
 Intrauterine growth restriction (AUGR)  
 Inadequate prenatal care  
 Maternal prenatal substance exposure  
 Mother affected with this condition  
 Preeclampsia  
 Rh isoimmunization  
 Toxemia  
 Young maternal age (15 years of age + under)  
 Preterm labor  
 Other

Type of prenatal substance exposure

- Unknown                       Alcohol                       Tobacco                       Illicit drugs  
 Harmful chemicals    Known teratogens

Maternal treatment for affected fetus    Unknown    None    Yes    No

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Type of maternal treatment for affected fetus

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Pregnancy complication/risk factor-other, specify \_\_\_\_\_

Type(s) of assisted reproductive technology used by the patient's mother

- |  |  |
|--|--|
| <input type="checkbox"/> In vitro fertilization (IVF)    | <input type="checkbox"/> Preimplantation genetic diagnosis (PGD) |
| <input type="checkbox"/> Intrauterine insemination (IUI) | <input type="checkbox"/> Surrogate                               |
| <input type="checkbox"/> Donor sperm                     | <input type="checkbox"/> Donor egg                               |
| <input type="checkbox"/> Donor embryo                    | <input type="checkbox"/> Other                                   |

Type(s) of assisted reproductive technology-other, specify \_\_\_\_\_

## Pregnancy

---

Patient has biological children  Unknown  Yes  No

Please complete the pregnancy form.

Patient is pregnant  Unknown  Yes  No

Please complete the pregnancy form.

## Neonatal History

---

Patient's birth was a result of multiple gestation pregnancy

- Unknown     
  No-single birth     
  Yes-twins (identical)   
  Yes-twins (fraternal)
- Yes-triplets     
  Yes-Other, specify

Specify other number of multiples \_\_\_\_\_

Congenital anomalies  Unknown  Yes  No

Type of congenital anomalies

- |   |  |
|---|--|
| <input type="checkbox"/> Aortic valve stenosis                        | <input type="checkbox"/> Atrial septal defect (A.S.D.) |
| <input type="checkbox"/> Atrioventricular septal defect               | <input type="checkbox"/> Biliary atresia               |
| <input type="checkbox"/> Bladder exstrophy                            | <input type="checkbox"/> Blind                         |
| <input type="checkbox"/> Bronchopulmonary dysplasia                   | <input type="checkbox"/> Choanal atresia               |
| <input type="checkbox"/> Cleft lip and/or palate                      | <input type="checkbox"/> Club foot                     |
| <input type="checkbox"/> Coarctation of aorta                         | <input type="checkbox"/> Common truncus                |
| <input type="checkbox"/> Congenital adrenal hyperplasia               | <input type="checkbox"/> Congenital cataract           |
| <input type="checkbox"/> Congenital heart disease                     | <input type="checkbox"/> Congenital hip dislocation    |
| <input type="checkbox"/> Congenital hypothyroidism                    | <input type="checkbox"/> Diaphragmatic hernia          |
| <input type="checkbox"/> Down syndrome                                | <input type="checkbox"/> Ebstein's anomaly             |
| <input type="checkbox"/> Endocardial cushion defect                   | <input type="checkbox"/> Epilepsy                      |
| <input type="checkbox"/> Esophageal atresia/Tracheoesophageal fistula | <input type="checkbox"/> Fetal alcohol syndrome        |



Patient Name \_\_\_\_\_

Date      

- |   |   |
|---|---|
| <input type="checkbox"/> Gastroschisis                                | <input type="checkbox"/> Hearing loss                         |
| <input type="checkbox"/> Hemoglobinopathies                           | <input type="checkbox"/> Hemophilia                           |
| <input type="checkbox"/> Hirshsprung's disease                        | <input type="checkbox"/> Hydrocephalus                        |
| <input type="checkbox"/> Hypoplastic left heart syndrome              | <input type="checkbox"/> Hypospadias and epispadias           |
| <input type="checkbox"/> Immune deficiency                            | <input type="checkbox"/> Microcephaly                         |
| <input type="checkbox"/> Missing or reduction of limb                 | <input type="checkbox"/> Obstructive genitourinary defect     |
| <input type="checkbox"/> Omphalocele                                  | <input type="checkbox"/> Patent ductus arteriosus (P.D.A.)    |
| <input type="checkbox"/> Pulmonary valve atresia and stenosis         | <input type="checkbox"/> Pyloric stenosis                     |
| <input type="checkbox"/> Rectal and large intestinal atresia/stenosis | <input type="checkbox"/> Renal agenesis/hypoplasia            |
| <input type="checkbox"/> Retinopathy of prematurity                   | <input type="checkbox"/> Severe combined immunodeficiency     |
| <input type="checkbox"/> Spina bifida                                 | <input type="checkbox"/> Tetralogy of fallot                  |
| <input type="checkbox"/> Transposition of the great arteries          | <input type="checkbox"/> Tricuspid valve atresia and stenosis |
| <input type="checkbox"/> Trisomy 13 (Patau syndrome)                  | <input type="checkbox"/> Trisomy 18                           |
| <input type="checkbox"/> Truncus arteriosus                           | <input type="checkbox"/> Ventricular septal defect (V.S.D.)   |
| <input type="checkbox"/> Other  |   |

Type of congenital anomalies-other, specify \_\_\_\_\_

Neonatal complications    Unknown    Yes    No

Type of neonatal complications

- |  |   |
|--|---|
| <input type="checkbox"/> Antibiotics                       | <input type="checkbox"/> APGAR < 5            |
| <input type="checkbox"/> Apnea/Bradycardia spells          | <input type="checkbox"/> Hypoglycemia         |
| <input type="checkbox"/> Intubation/mechanical ventilation | <input type="checkbox"/> Infection/sepsis     |
| <input type="checkbox"/> IV fluids                         | <input type="checkbox"/> Jaundice             |
| <input type="checkbox"/> Premature (< 37 weeks gestation)  | <input type="checkbox"/> Respiratory distress |
| <input type="checkbox"/> Transfused                        | <input type="checkbox"/> Seizures             |
| <input type="checkbox"/> Other                             |   |

Type of neonatal complications-other, specify \_\_\_\_\_

Type of neonatal nutrition

- |   |                                     |  |  |
|---|-------------------------------------|--|--|
| <input type="checkbox"/> Unknown              | <input type="checkbox"/> TPN        | <input type="checkbox"/> Breast milk     | <input type="checkbox"/> Elemental formula   |
| <input type="checkbox"/> Human milk fortifier | <input type="checkbox"/> Intralipid | <input type="checkbox"/> Regular formula | <input type="checkbox"/> Non-Lactose formula |
| <input type="checkbox"/> Metabolic formula    | <input type="checkbox"/> Other      |  |  |

Type of neonatal nutrition-other, specify \_\_\_\_\_

**Birth Measurements**Birth measurements    Unknown    Head circumference    Length    Weight

Birth head circumference \_\_\_\_\_

Birth head circumference units    cm    in

Patient Name \_\_\_\_\_

Date      

Birth length \_\_\_\_\_

Birth length units  cm  in

Birth weight \_\_\_\_\_

Birth weight units  lbs  kg  gm  oz**Health History**

---

Patient has had an outpatient speciality visit  Unknown  Yes  NoDate of last outpatient specialty visit      

Days of age from birth until intervention for this condition \_\_\_\_\_

Days of age from birth until first seen by subspecialist \_\_\_\_\_

**Dialysis**

---

Dialysis (any type) prior to intake  Unknown  Yes  No

Please complete the dialysis form.

**Transplants**

---

Transplant prior to intake

 Unknown Yes-transplant received No-patient was evaluated for transplant but did not receive No

Please complete the transplant form.

**Other History**

---

Cardiomyopathy prior to intake  Unknown  No  No imaging done  Dilated  HypertrophicCardiomyopathy-dilated  Unknown  Left  Right  BothCardiomyopathy-hypertrophic  Unknown  Left  Right  BothHospitalizations prior to intake  Unknown  Yes  No

Number of hospitalizations prior to intake related to this condition \_\_\_\_\_

Number of hospitalizations prior to intake not related to this condition \_\_\_\_\_

Genetic counseling provided  Unknown  Yes  No

Provider of genetic counseling

 Unknown Dietitian Genetic counselor Neuropsychologist Nurse Nurse practitioner Physician Physician assistant Other

Provider of genetic counseling, other- specify \_\_\_\_\_

Patient Name \_\_\_\_\_

Date

Comorbidities at time of intake

**Prior testing**

---

Echocardiogram prior to intake Unknown Yes No

Echocardiogram date

Echocardiogram results Unknown Normal Abnormal

Echocardiogram comments

Electrocardiogram prior to intake Unknown Yes No

Electrocardiogram date

Electrocardiogram results Unknown Normal Abnormal

Electrocardiogram comments

Neurological imaging prior to intake Unknown Yes No

Neurological imaging date

Neurological imaging results Unknown Normal Abnormal

Neurological imaging comments

History of a seizure disorder Unknown Yes No

Patient has biospecimen stored Unknown Yes No

Type of patient biospecimen Blood Urine Other tissue

Type of patient biospecimen-other tissue, specify \_\_\_\_\_

Specify biospecimen location \_\_\_\_\_

**Eye Exam**

---

Eye exam performed prior to intake Unknown Yes No

Eye exam findings Unknown Within normal limits Evidence of nystagmus Other

Patient Name_____
Date <input type="text" value="M"/> <input type="text" value="M"/> <input type="text" value="D"/> <input type="text" value="D"/> <input type="text" value="Y"/> <input type="text" value="Y"/>

Eye exam findings-other, specify\_\_\_\_\_

### Emergency Management

---

Patient was enrolled in web-based emergency alert program     Unknown     Yes     No

Name of web-based emergency alert program\_\_\_\_\_

Patient/primary caregiver was given a written emergency letter     Unknown     Yes     No

Patient/primary caregiver was given a sick day plan specific to this condition     Unknown     Yes     No

Patient/primary caregiver was given the 24 hour on-call contact information for a specialty provider  
 Unknown                               Yes     No

### Comments

---

Intake past health history comments

Patient Name \_\_\_\_\_

Date      

## CACT - Intake Newborn Screening

### Newborn Screening

Newborn screening performed  Unknown  Yes  NoNumber of newborn screen results available  Unknown  0  1  2  3Date first newborn screen collected      First newborn screen take in neonatal intensive care (NICU)  Unknown  Yes  No

Days of age from birth primary or subspecialist first notified about abnormal NBS screen \_\_\_\_\_

Reason for first newborn screen  Routine  Research

Carnitine free (C0)/Palmitoylcarnitine (C16)+Stearoylcarnitine (C18) on first newborn screen \_\_\_\_\_

Tetradecanoylcarnitine (C14) on first newborn screen \_\_\_\_\_

Tetradecanoylcarnitine (C14) on first newborn screen units  umol/L

Palmitoleylcarnitine (C16:1) on first newborn screen \_\_\_\_\_

Palmitoleylcarnitine (C16:1) on first newborn screen units  umol/L

Palmitoylcarnitine (C16) on first newborn screen \_\_\_\_\_

Palmitoylcarnitine (C16) on first newborn screen units  umol/L

Linoleoylcarnitine (C18:2) on first newborn screen \_\_\_\_\_

Linoleoylcarnitine (C18:2) on first newborn screen units  umol/L

Oleoylcarnitine (C18:1) on first newborn screen \_\_\_\_\_

Oleoylcarnitine (C18:1) on first newborn screen units  umol/L

Stearoylcarnitine (C18) on first newborn screen \_\_\_\_\_

Stearoylcarnitine (C18) on first newborn screen units  umol/L

Other result/report on first newborn screen

Other result/report on first newborn screen units \_\_\_\_\_

Date second newborn screen collected      

Reason for second newborn screen

 Unsatisfactory Borderline Result NICU Protocol TPN Transfused State Mandate NBS collected < 24 hours of age  Other

Reason for second newborn screen-other, specify \_\_\_\_\_

Carnitine free (C0)/Palmitoylcarnitine (C16)+Stearoylcarnitine (C18) on second newborn screen

Patient Name \_\_\_\_\_

Date      

Tetradecanoylcarnitine (C14) on second newborn screen \_\_\_\_\_

Tetradecanoylcarnitine (C14) on second newborn screen units  umol/L

Palmitoleylcarnitine (C16:1) on second newborn screen \_\_\_\_\_

Palmitoleylcarnitine (C16:1) on second newborn screen units  umol/L

Palmitoylcarnitine (C16) on second newborn screen \_\_\_\_\_

Palmitoylcarnitine (C16) on second newborn screen units  umol/L

Linoleoylcarnitine (C18:2) on second newborn screen \_\_\_\_\_

Linoleoylcarnitine (C18:2) on second newborn screen units  umol/L

Oleoylcarnitine (C18:1) on second newborn screen \_\_\_\_\_

Oleoylcarnitine (C18:1) on second newborn screen units  umol/L

Stearoylcarnitine (C18) on second newborn screen \_\_\_\_\_

Stearoylcarnitine (C18) on second newborn screen units  umol/L

Other result/report on second newborn screen

Other result/report on second newborn screen units \_\_\_\_\_

Date third newborn screen collected      

Reason for third newborn screen

- Unsatisfactory   
 Borderline Result   
 NICU Protocol   
 TPN   
 Transfused  
 State Mandate   
 Other

Reason for third newborn screen-other, specify \_\_\_\_\_

Carnitine free (C0)/Palmitoylcarnitine (C16)+Stearoylcarnitine (C18) on third newborn screen \_\_\_\_\_

Tetradecanoylcarnitine (C14) on third newborn screen \_\_\_\_\_

Tetradecanoylcarnitine (C14) on third newborn screen units  umol/L

Palmitoleylcarnitine (C16:1) on third newborn screen \_\_\_\_\_

Palmitoleylcarnitine (C16:1) on third newborn screen units  umol/L

Palmitoylcarnitine (C16) on third newborn screen \_\_\_\_\_

Palmitoylcarnitine (C16) on third newborn screen units  umol/L

Linoleoylcarnitine (C18:2) on third newborn screen \_\_\_\_\_

Linoleoylcarnitine (C18:2) on third newborn screen units  umol/L

Oleoylcarnitine (C18:1) on third newborn screen \_\_\_\_\_

Oleoylcarnitine (C18:1) on third newborn screen units  umol/L

Stearoylcarnitine (C18) on third newborn screen \_\_\_\_\_

Patient Name \_\_\_\_\_

Date      Stearoylcarnitine (C18) on third newborn screen units  umol/L

Other result/report on third newborn screen

--

Other result/report on third newborn screen units \_\_\_\_\_

## Newborn Hearing Screen

---

Newborn hearing screen performed  Unknown  Yes  NoR Ear: Equipment Type  Unknown  DPOAE  TEOAE  ABR  AABR  ALGO  OtherRight ear: Screening test results  Pass  Refer  Not TestedL Ear: Equipment Type  Unknown  DPOAE  TEOAE  ABR  AABR  ALGO  OtherLeft ear: Screening test results  Pass  Refer  Not Tested

### Recommendation

 Repeat hearing screen  Referral for diagnostic testing  Risk factor monitoring only Refused further action  No further action requiredDate of Audiological Diagnostic Evaluation      Right ear: Diagnosis: Hearing loss?  Yes  No

Right ear: Diagnosis: Degree of hearing loss

 Mild (21-40db)  Moderate (41-70db)  Severe (71-90db)  Profound (91db +)

Right ear: Diagnosis: Type of hearing loss

 Sensorineural Conductive Mixed Auditory neuropathy/Auditory dys-synchrony OtherLeft ear: Diagnosis: Hearing loss?  Yes  No

Left ear: Diagnosis: Degree of hearing loss

 Mild (21-40db)  Moderate (41-70db)  Severe (71-90db)  Profound (91db +)

Left ear: Diagnosis: Type of hearing loss

 Sensorineural Conductive Mixed Auditory neuropathy/Auditory dys-synchrony Other

## Comments

---

Intake newborn screening comments

--

Patient Name \_\_\_\_\_

Date      

## CACT - Intake Initial Testing

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---

Patient status at time of NBS reporting to specialty center

 Unknown Well Symptomatic Deceased

Patient symptoms at time of initial contact

 Unknown None Alopecia Apnea Arrhythmia Ataxia Athetosis Autistic-like features Body odor Brain abnormalities Brain malformations Candidiasis Cardiomyopathy Cataract(s) Cerebral edema Chorea Cirrhosis Clonus Cognitive impairment Coma Confusion Conjunctivitis Contracture(s)-musculoskeletal Corneal erosion Dehydration Dermatitis Developmental delay(s) Disorientation Drooling/hypersalivation Dysarthria Dysmetria Dysmorphism Dysphagia Dystonia Eczema Edema Failure to thrive Fatigue Flapping tremor Fluctuating level of alertness Gait abnormality (other than ataxia) Genital abnormalities Headache Hearing loss Hepatic encephalopathy Hepatomegaly Hyperreflexia Hypertension Hypertonia Hyporeflexia Hypothermia Hypotonia Increased intracranial pressure Infection/sepsis Irritability Jaundice Keratosis Learning disability Lethargy Liver failure-acute



Patient Name \_\_\_\_\_

Date      

- |  |   |
|--|---|
| <input type="checkbox"/> Loss of consciousness | <input type="checkbox"/> Loss of developmental milestone(s) |
| <input type="checkbox"/> Macrocephaly          | <input type="checkbox"/> Malignant hyperthermia             |
| <input type="checkbox"/> Microcephaly          | <input type="checkbox"/> Multiorgan failure                 |
| <input type="checkbox"/> Myopathy              | <input type="checkbox"/> Nystagmus                          |
| <input type="checkbox"/> Opisthotonos          | <input type="checkbox"/> Optic nerve atrophy                |
| <input type="checkbox"/> Pancreatitis          | <input type="checkbox"/> Peripheral neuropathy              |
| <input type="checkbox"/> Photophobia           | <input type="checkbox"/> Polycystic kidney(s)               |
| <input type="checkbox"/> Poor feeding          | <input type="checkbox"/> Poor growth                        |
| <input type="checkbox"/> Profuse sweating      | <input type="checkbox"/> Renal dysplasia                    |
| <input type="checkbox"/> Renal failure-acute   | <input type="checkbox"/> Retinal hemorrhage                 |
| <input type="checkbox"/> Rickets               | <input type="checkbox"/> Rigidity                           |
| <input type="checkbox"/> Scotomas              | <input type="checkbox"/> Seizure                            |
| <input type="checkbox"/> Slurred speech        | <input type="checkbox"/> Spasticity                         |
| <input type="checkbox"/> Splenomegaly          | <input type="checkbox"/> Stereotyped movements              |
| <input type="checkbox"/> Stomatitis            | <input type="checkbox"/> Stridor                            |
| <input type="checkbox"/> Stroke                | <input type="checkbox"/> Subdural hemorrhage                |
| <input type="checkbox"/> Sudden death          | <input type="checkbox"/> Syncope                            |
| <input type="checkbox"/> Tachycardia           | <input type="checkbox"/> Tachypnea                          |
| <input type="checkbox"/> Tremors               | <input type="checkbox"/> Trichorrhesis nodosa               |
| <input type="checkbox"/> Vision loss           | <input type="checkbox"/> Vomiting                           |
| <input type="checkbox"/> Other                 |   |

Patient symptoms at time of initial contact-other, specify \_\_\_\_\_

Patient lab abnormalities reported or recorded at time patient or caregiver first contacts disease specialist

- |  |  |   |
|--|--|---|
| <input type="checkbox"/> Unknown                   | <input type="checkbox"/> No abnormal labs              | <input type="checkbox"/> No labs done                     |
| <input type="checkbox"/> Aminoaciduria             | <input type="checkbox"/> Anemia                        | <input type="checkbox"/> Bone marrow suppression          |
| <input type="checkbox"/> Coagulopathy              | <input type="checkbox"/> Elevated amylase              | <input type="checkbox"/> Elevated CK                      |
| <input type="checkbox"/> Elevated lipase           | <input type="checkbox"/> Elevated liver function tests | <input type="checkbox"/> Hematuria                        |
| <input type="checkbox"/> Hemolytic anemia          | <input type="checkbox"/> Hyperammonemia                | <input type="checkbox"/> Hyperglycemia                    |
| <input type="checkbox"/> Hyperglycinemia           | <input type="checkbox"/> Hyperinsulinism               | <input type="checkbox"/> Hypertriglyceridemia             |
| <input type="checkbox"/> Hyperuricemia             | <input type="checkbox"/> Hypoglycemia                  | <input type="checkbox"/> Hypokalemia                      |
| <input type="checkbox"/> Hypoproteinemia           | <input type="checkbox"/> Immunological abnormalities   | <input type="checkbox"/> Ketonuria                        |
| <input type="checkbox"/> Ketosis                   | <input type="checkbox"/> Lactic acidosis               | <input type="checkbox"/> Low/absent ketones               |
| <input type="checkbox"/> Metabolic acidosis        | <input type="checkbox"/> Myoglobinuria                 | <input type="checkbox"/> Plasma total carnitine elevation |
| <input type="checkbox"/> Plasma free carnitine low | <input type="checkbox"/> Plasma total carnitine low    | <input type="checkbox"/> Proteinuria                      |
| <input type="checkbox"/> Renal tubular acidosis    | <input type="checkbox"/> Respiratory alkalosis         | <input type="checkbox"/> Other                            |

Patient Name \_\_\_\_\_

Date      

Patient lab abnormalities reported or recorded at time patient or caregiver first contacts disease specialist-other, specify \_\_\_\_\_

## Diagnostic Testing

---

Diagnostic labs performed

- |   |   |
|---|---|
| <input type="checkbox"/> Unknown                          | <input type="checkbox"/> None   |
| <input type="checkbox"/> Blood spot acylcarnitine profile | <input type="checkbox"/> Carnitine:acylcarnitine translocase enzyme assay |
| <input type="checkbox"/> Creatinine kinase                | <input type="checkbox"/> Fatty acid oxidation probe assay                 |
| <input type="checkbox"/> Plasma acylcarnitine profile     | <input type="checkbox"/> Plasma ammonia                                   |
| <input type="checkbox"/> Plasma carnitine                 | <input type="checkbox"/> Urine organic acids                              |
| <input type="checkbox"/> Western blot                     | <input type="checkbox"/> Other  |

Blood spot acylcarnitine profile     Unknown     Within normal limits     Abnormal

Blood spot acylcarnitine profile significance     Diagnostic     Non-diagnostic

Blood spot acylcarnitine profile comments

Blood spot acylcarnitine profile reference range \_\_\_\_\_

Number of times carnitine:acylcarnitine translocase enzyme assay done     1     2

Carnitine:acylcarnitine translocase enzyme assay     Unknown     Within normal limits     Abnormal

Carnitine:acylcarnitine translocase enzyme assay significance     Diagnostic     Non-diagnostic

Carnitine:acylcarnitine translocase enzyme assay value \_\_\_\_\_

Carnitine:acylcarnitine translocase enzyme assay units     ng/mL

Carnitine:acylcarnitine translocase enzyme assay reference range \_\_\_\_\_

Carnitine:acylcarnitine translocase enzyme assay medium

- Unknown     Fibroblasts     Liver     Lymphocytes     Red blood cells
- Other

Carnitine:acylcarnitine translocase enzyme assay medium, other- specify \_\_\_\_\_

Second carnitine:acylcarnitine translocase enzyme assay

Unknown     Within normal limits     Abnormal

Second carnitine:acylcarnitine translocase enzyme assay significance

Diagnostic     Non-diagnostic

Second carnitine:acylcarnitine translocase enzyme assay value \_\_\_\_\_

Second carnitine:acylcarnitine translocase enzyme assay units     ng/mL

Second carnitine:acylcarnitine translocase enzyme assay reference range \_\_\_\_\_

Second carnitine:acylcarnitine translocase enzyme assay medium

Patient Name \_\_\_\_\_

Date      

- Unknown     Fibroblasts     Liver     Lymphocytes     Red blood cells  
 Other

Second carnitine:acylcarnitine translocase enzyme assay medium, other- specify

\_\_\_\_\_

Creatinine kinase     Unknown     Within normal limits     Abnormal

Creatinine kinase significance     Diagnostic     Non-diagnostic

Creatinine kinase value\_\_\_\_\_

Creatinine kinase units     U/L     umol/L     mg/dL

Creatinine kinase reference range\_\_\_\_\_

Fatty acid oxidation probe assay     Unknown     Within normal limits     Abnormal

Fatty acid oxidation probe assay significance     Diagnostic     Non-diagnostic

Fatty acid oxidation probe value\_\_\_\_\_

Fatty acid oxidation probe units     nmol/mg protein     mU/mg

Fatty acid oxidation probe reference range\_\_\_\_\_

Plasma acylcarnitine profile     Unknown     Within normal limits     Abnormal

Plasma acylcarnitine profile significance     Diagnostic     Non-diagnostic

Plasma acylcarnitine profile comments

Plasma ammonia     Unknown     Within normal limits     Abnormal

Plasma ammonia significance     Diagnostic     Non-diagnostic

Plasma ammonia value\_\_\_\_\_

Plasma ammonia units     umol/L     mg/dL

Plasma ammonia reference range\_\_\_\_\_

Plasma carnitine levels

Unknown                                     Free carnitine                                     Total carnitine

Esterified carnitine                                     Esterified:Free carnitine ratio

Free plasma carnitine     Unknown     Within normal limits     Abnormal

Free plasma carnitine significance     Diagnostic     Non-diagnostic

Free plasma carnitine value\_\_\_\_\_

Free plasma carnitine units     umol/L     mg/dL

Free plasma carnitine reference range\_\_\_\_\_

Total plasma carnitine     Unknown     Within normal limits     Abnormal

Total plasma carnitine significance     Diagnostic     Non-diagnostic

Patient Name \_\_\_\_\_

Date      

Total plasma carnitine value \_\_\_\_\_

Total plasma carnitine units  umol/L  mg/dL

Total plasma carnitine reference range \_\_\_\_\_

Esterified plasma carnitine  Unknown  Within normal limits  AbnormalEsterified plasma carnitine significance  Diagnostic  Non-diagnostic

Esterified plasma carnitine value \_\_\_\_\_

Esterified plasma carnitine units  umol/L  mg/dL

Esterified plasma carnitine reference range \_\_\_\_\_

Esterified:Free plasma carnitine ratio  Unknown  Within normal limits  AbnormalEsterified:Free plasma carnitine ratio significance  Diagnostic  Non-diagnostic

Esterified:Free plasma carnitine ratio value \_\_\_\_\_

Esterified:Free plasma carnitine ratio reference range \_\_\_\_\_

Urine organic acids  Unknown  Within normal limits  AbnormalUrine organic acids significance  Diagnostic  Non-diagnostic

Urine organic acid comments

Western blot  < 20%  >=20% < 50%  >=50%  Results unavailable

Diagnostic lab-other, specify \_\_\_\_\_

Other diagnostic lab  Unknown  Within normal limits  AbnormalOther diagnostic lab significance  Diagnostic  Non-diagnostic

Other diagnostic lab comments

Other diagnostic lab value \_\_\_\_\_

Other diagnostic lab units \_\_\_\_\_

Other diagnostic lab reference range \_\_\_\_\_

## Genetic Testing

---

Type of genetic/genomic testing

- |   |  |   |
|---|--|---|
| <input type="checkbox"/> Unknown                | <input type="checkbox"/> Not done            | <input type="checkbox"/> Done, not available          |
| <input type="checkbox"/> Single gene            | <input type="checkbox"/> Mutation panel      | <input type="checkbox"/> Exome sequencing             |
| <input type="checkbox"/> Full genome sequencing | <input type="checkbox"/> Copy number variant | <input type="checkbox"/> Deletion/duplication testing |
| <input type="checkbox"/> Other                  |  |   |

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

Reason genotyping was done

- Unknown                     
  Diagnosis confirmation  
  Modifier genes                     
  Pharmacogenetics  
 Other

Reason genotyping was done-other, specify\_\_\_\_\_

Reason genotyping was not done

- Unknown                     
  Clinician deemed unnecessary  
  Not covered by insurance  
 Parent refusal                     
  Test not available                     
  Other

Reason genotyping was not done-other, specify\_\_\_\_\_

Type of genetic/genomic testing-other, specify\_\_\_\_\_

Gene(s) associated with CACT     SLC25A20     Other

Gene(s) associated with CACT-other, specify\_\_\_\_\_

SLC25A20: Specify allele 1\_\_\_\_\_

SLC25A20: Specify allele 2\_\_\_\_\_

Other: Specify allele 1\_\_\_\_\_

Other: Specify allele 2\_\_\_\_\_

Maternal genetic testing done     Unknown     Yes     No     Genotyping in progress

Mother: Allele 1\_\_\_\_\_

Mother: Allele 2\_\_\_\_\_

Paternal genetic testing done     Unknown     Yes     No     Genotyping in progress

Father: Allele 1\_\_\_\_\_

Father: Allele 2\_\_\_\_\_

**Comments**

---

Initial testing comments

Patient Name \_\_\_\_\_

Date        

## CACT - Visit Demographics And History

---

Visit Date      Date of last outpatient speciality visit      

### Consent

---

Patient consent valid  Unknown  Yes  No

Obtain new consent prior to completing data entry.

### Care and Other Studies

---

Providers seen at this visit

- Unknown       Child and family life     Dietitian                       Genetic counselor  
 Neuropsychologist     Nurse                               Nurse practitioner       Pharmacist  
 Physician                       Physician assistant       Psychologist               Social Worker  
 Other

Providers seen at this visit, other- specify \_\_\_\_\_

Location of visit  In office  Not in person-by telephone  Telemedicine  Other

Location of visit-other, specify \_\_\_\_\_

Patient has moved to a new residence since the last visit  Unknown  Yes  No

Miles from home to primary care \_\_\_\_\_

Miles from home to specialty care \_\_\_\_\_

Name of primary care center \_\_\_\_\_

Patient has enrolled in a research study since the last visit  Unknown  Yes  NoOther research studies are clinical trials  Unknown  Yes  No

Identify the research study \_\_\_\_\_

Clinicaltrials.gov identifier \_\_\_\_\_

### Education

---

Education status has changed since the last visit  Unknown  Yes  No

Complete education questions on the Demographics form.

### Medical Coverage

---

Medical coverage at visit

- Unknown  
 None  
 Commercial/private  
 Medicaid

Patient Name _____
Date <input type="text" value="M"/> <input type="text" value="M"/> <input type="text" value="D"/> <input type="text" value="D"/> <input type="text" value="Y"/> <input type="text" value="Y"/>

- Medicare
- Military
- Newborn screening funds
- Patient assistance program
- State Children's Health Insurance Program (SCHIP)
- State Children with Special Health Needs (CSHN) Program
- Other

Medical coverage at visit-Patient assistance program, specify \_\_\_\_\_

Medical coverage at visit-Other, specify \_\_\_\_\_

**Family History**

---

Patient has new biological sibling since last visit     Unknown     Yes     No

Complete sibling history questions on the Family History form.

**Comments**

---

Visit demographics and history comments

Patient Name \_\_\_\_\_

Date      

## CACT - Visit Health History

### Health Status

#### Immunization status

- Unknown  Not up to date  
 Up to date via report  Up to date via clinical confirmation  
 Immunizations declined

Reason immunization status not up to date

#### Current comorbidities

Patient has had seizures since last visit  Unknown  Yes  No

Highest level of CK since last visit \_\_\_\_\_

Highest level of CK since last visit units  U/L  umol/L  mg/dLDocumented hyperammonemia since last visit  Unknown  Yes  No

Highest level of ammonia since last visit \_\_\_\_\_

Highest level of ammonia since last visit units  umol/L  ug/dL

Documented hypoglycemia (blood glucose &lt; 60 mg/dL) since the last outpatient metabolic visit

 Unknown  Yes  No

Lowest blood glucose value reported since last outpatient metabolic visit \_\_\_\_\_

Lowest blood glucose value reported since last outpatient metabolic visit units  mmol/L  mg/dL

Lowest blood glucose range reported since last outpatient metabolic visit \_\_\_\_\_

### Sick Visits

Sick visits since last outpatient visit  Unknown  Yes  No

Number of sick visits \_\_\_\_\_

Date of sick visit 1      Reason for sick visit 1  Unknown  Condition related  Condition unrelatedSick visit 1 was a condition exacerbation  Unknown  Yes  No

Location for sick visit 1

 Unknown  Emergency department  Retail clinic



Patient Name \_\_\_\_\_

Date |M|M| |D|D| |Y|Y|

Primary care                       Specialty center                       Urgent care

Direct hospital admission  Other

Patient was admitted to the hospital as a result of sick visit 1     Unknown     Yes     No

Name of hospital for sick visit 1 \_\_\_\_\_

ICD-9 codes for sick visit 1 known     Yes     No

ICD-9 codes for sick visit 1 \_\_\_\_\_

Diagnosis for sick visit 1 \_\_\_\_\_

Number of inpatient days for sick visit 1 \_\_\_\_\_

Number of ICU days for sick visit 1 \_\_\_\_\_

Date of sick visit 2 |M|M| |D|D| |Y|Y|

Reason for sick visit 2     Unknown     Condition related     Condition unrelated

Sick visit 2 was a condition exacerbation     Unknown     Yes     No

Location for sick visit 2

Unknown                       Emergency department     Retail clinic

Primary care                       Specialty center                       Urgent care

Direct hospital admission  Other

Patient was admitted to the hospital as a result of sick visit 2     Unknown     Yes     No

Name of hospital for sick visit 2 \_\_\_\_\_

ICD-9 codes for sick visit 2 known     Yes     No

ICD-9 codes for sick visit 2 \_\_\_\_\_

Diagnosis for sick visit 2 \_\_\_\_\_

Number of inpatient days for sick visit 2 \_\_\_\_\_

Number of ICU days for sick visit 2 \_\_\_\_\_

Date of sick visit 3 |M|M| |D|D| |Y|Y|

Reason for sick visit 3     Unknown     Condition related     Condition unrelated

Sick visit 3 was a condition exacerbation     Unknown     Yes     No

Location for sick visit 3

Unknown                       Emergency department     Retail clinic

Primary care                       Specialty center                       Urgent care

Direct hospital admission  Other

Patient was admitted to the hospital as a result of sick visit 3     Unknown     Yes     No

Name of hospital for sick visit 3 \_\_\_\_\_

ICD-9 codes for sick visit 3 known     Yes     No

ICD-9 codes for sick visit 3 \_\_\_\_\_

Diagnosis for sick visit 3 \_\_\_\_\_

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

Number of inpatient days for sick visit 3 \_\_\_\_\_

Number of ICU days for sick visit 3 \_\_\_\_\_

Date of sick visit 4

Reason for sick visit 4    Unknown    Condition related    Condition unrelated

Sick visit 4 was a condition exacerbation    Unknown    Yes    No

Location for sick visit 4

Unknown                       Emergency department    Retail clinic

Primary care                       Specialty center                       Urgent care

Direct hospital admission    Other

Patient was admitted to the hospital as a result of sick visit 4    Unknown    Yes    No

Name of hospital for sick visit 4 \_\_\_\_\_

ICD-9 codes for sick visit 4 known    Yes    No

ICD-9 codes for sick visit 4 \_\_\_\_\_

Diagnosis for sick visit 4 \_\_\_\_\_

Number of inpatient days for sick visit 4 \_\_\_\_\_

Number of ICU days for sick visit 4 \_\_\_\_\_

Date of sick visit 5

Reason for sick visit 5    Unknown    Condition related    Condition unrelated

Sick visit 5 was a condition exacerbation    Unknown    Yes    No

Location for sick visit 5

Unknown                       Emergency department    Retail clinic

Primary care                       Specialty center                       Urgent care

Direct hospital admission    Other

Patient was admitted to the hospital as a result of sick visit 5    Unknown    Yes    No

Name of hospital for sick visit 5 \_\_\_\_\_

ICD-9 codes for sick visit 5 known    Yes    No

ICD-9 codes for sick visit 5 \_\_\_\_\_

Diagnosis for sick visit 5 \_\_\_\_\_

Number of inpatient days for sick visit 5 \_\_\_\_\_

Number of ICU days for sick visit 5 \_\_\_\_\_

Date of sick visit 6

Reason for sick visit 6    Unknown    Condition related    Condition unrelated

Sick visit 6 was a condition exacerbation    Unknown    Yes    No

Location for sick visit 6

Unknown                       Emergency department    Retail clinic

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

- Primary care                       Specialty center                       Urgent care  
 Direct hospital admission    Other

Patient was admitted to the hospital as a result of sick visit 6    Unknown    Yes    No

Name of hospital for sick visit 6 \_\_\_\_\_

ICD-9 codes for sick visit 6 known    Yes    No

ICD-9 codes for sick visit 6 \_\_\_\_\_

Diagnosis for sick visit 6 \_\_\_\_\_

Number of inpatient days for sick visit 6 \_\_\_\_\_

Number of ICU days for sick visit 6 \_\_\_\_\_

Date of sick visit 7

Reason for sick visit 7    Unknown    Condition related    Condition unrelated

Sick visit 7 was a condition exacerbation    Unknown    Yes    No

Location for sick visit 7

- Unknown                               Emergency department    Retail clinic  
 Primary care                               Specialty center                       Urgent care  
 Direct hospital admission    Other

Patient was admitted to the hospital as a result of sick visit 7    Unknown    Yes    No

Name of hospital for sick visit 7 \_\_\_\_\_

ICD-9 codes for sick visit 7 known    Yes    No

ICD-9 codes for sick visit 7 \_\_\_\_\_

Diagnosis for sick visit 7 \_\_\_\_\_

Number of inpatient days for sick visit 7 \_\_\_\_\_

Number of ICU days for sick visit 7 \_\_\_\_\_

Date of sick visit 8

Reason for sick visit 8    Unknown    Condition related    Condition unrelated

Sick visit 8 was a condition exacerbation    Unknown    Yes    No

Location for sick visit 8

- Unknown                               Emergency department    Primary care  
 Specialty center                               Urgent care                               Direct hospital admission  
 Other

Patient was admitted to the hospital as a result of sick visit 8    Unknown    Yes    No

Name of hospital for sick visit 8 \_\_\_\_\_

ICD-9 codes for sick visit 8 known    Yes    No

ICD-9 codes for sick visit 8 \_\_\_\_\_

Diagnosis for sick visit 8 \_\_\_\_\_

Patient Name \_\_\_\_\_

Date      

Number of inpatient days for sick visit 8 \_\_\_\_\_

Number of ICU days for sick visit 8 \_\_\_\_\_

Date of sick visit 9      Reason for sick visit 9  Unknown  Condition related  Condition unrelatedSick visit 9 was a condition exacerbation  Unknown  Yes  No

Location for sick visit 9

 Unknown  Emergency department  Retail clinic Primary care  Specialty center  Urgent care Direct hospital admission  OtherPatient was admitted to the hospital as a result of sick visit 9  Unknown  Yes  No

Name of hospital for sick visit 9 \_\_\_\_\_

ICD-9 codes for sick visit 9 known  Yes  No

ICD-9 codes for sick visit 9 \_\_\_\_\_

Diagnosis for sick visit 9 \_\_\_\_\_

Number of inpatient days for sick visit 9 \_\_\_\_\_

Number of ICU days for sick visit 9 \_\_\_\_\_

Date of sick visit 10      Reason for sick visit 10  Unknown  Condition related  Condition unrelatedSick visit 10 was a condition exacerbation  Unknown  Yes  No

Location for sick visit 10

 Unknown  Emergency department  Retail clinic Primary care  Specialty center  Urgent care Direct hospital admission  OtherPatient was admitted to the hospital as a result of sick visit 10  Unknown  Yes  No

Name of hospital for sick visit 10 \_\_\_\_\_

ICD-9 codes for sick visit 10 known  Yes  No

ICD-9 codes for sick visit 10 \_\_\_\_\_

Diagnosis for sick visit 10 \_\_\_\_\_

Number of inpatient days for sick visit 10 \_\_\_\_\_

Number of ICU days for sick visit 10 \_\_\_\_\_

**Procedures**Anesthesia since last visit  Unknown  Yes  NoSpecific anesthesia precautions recommended due to metabolic condition  Unknown  Yes  No

Patient Name \_\_\_\_\_

Date

Surgical precautions taken

Anesthesia complications Unknown Yes No

Anesthesia complications-specify

Surgeries since last visit Unknown Yes No

Surgical procedure(s) \_\_\_\_\_

Surgery complications Unknown Yes No

Surgery complications-specify

**Pregnancy**

---

Patient has had a biological child since the last visit Unknown Yes No

Please complete the pregnancy form.

Patient has become pregnant since the last visit Unknown Yes No

Please complete the pregnancy form.

**Dialysis**

---

Dialysis (any type) since the last outpatient metabolic visit Unknown Yes No

Please complete the dialysis form.

**Transplants**

---

Transplant since last visit

- Unknown
- Yes-transplant received
- No-patient was evaluated for transplant but did not receive
- No

Please complete the transplant form.

**Other Procedures**

---

Major medical procedure since last visit Unknown Yes No

Description of major medical procedure \_\_\_\_\_

Patient Name \_\_\_\_\_

Date

## Comments

---

Visit health history comments

Patient Name _____
Date <input type="text" value="M"/> <input type="text" value="M"/> <input type="text" value="D"/> <input type="text" value="D"/> <input type="text" value="Y"/> <input type="text" value="Y"/>

# CACT - Visit Findings

## Visit Measurements

Measurements taken at visit

Unknown       None       Blood pressure       Head circumference

Height/length       Weight

Blood pressure-systolic \_\_\_\_\_

Blood pressure-diastolic \_\_\_\_\_

Head circumference \_\_\_\_\_

Head circumference units     cm     in

Height/length \_\_\_\_\_

Height/length units     cm     in

How height/length measured     Unknown     Supine     Standing

Weight \_\_\_\_\_

Weight units     lbs     kg     gm     oz

Body mass index \_\_\_\_\_

Visit findings     Unknown     None     Hypotonia     Nystagmus     Other

Visit findings-other, specify \_\_\_\_\_

## Comments

Visit findings comments

Patient Name \_\_\_\_\_

Date      

## CACT - Visit Ancillary Care

### Care Coordination

Missed subspecialty visits since last visit  Unknown  Yes  No

Number of missed subspecialty visits \_\_\_\_\_

Other health services currently received  Unknown  Yes  No

Specify other current health services

- |  |   |
|--|---|
| <input type="checkbox"/> Unknown                                     | <input type="checkbox"/> Allergy                  |
| <input type="checkbox"/> Audiology                                   | <input type="checkbox"/> Behavioral/Developmental |
| <input type="checkbox"/> Cardiology                                  | <input type="checkbox"/> Dentistry                |
| <input type="checkbox"/> Dermatology                                 | <input type="checkbox"/> Dietitian                |
| <input type="checkbox"/> Endocrinology                               | <input type="checkbox"/> Gastroenterology         |
| <input type="checkbox"/> Genetic Counseling                          | <input type="checkbox"/> Hematology               |
| <input type="checkbox"/> Home health care                            | <input type="checkbox"/> Nephrology               |
| <input type="checkbox"/> Neurology                                   | <input type="checkbox"/> Neuropsychology          |
| <input type="checkbox"/> Neurosurgery                                | <input type="checkbox"/> Occupational therapy     |
| <input type="checkbox"/> Oncology                                    | <input type="checkbox"/> Ophthalmology            |
| <input type="checkbox"/> Orthopedics                                 | <input type="checkbox"/> Otolaryngology           |
| <input type="checkbox"/> Physical medicine and rehabilitation (PM&R) | <input type="checkbox"/> Physical therapy         |
| <input type="checkbox"/> Primary care provider                       | <input type="checkbox"/> Psychiatry               |
| <input type="checkbox"/> Psychology                                  | <input type="checkbox"/> Public health nursing    |
| <input type="checkbox"/> Pulmonology                                 | <input type="checkbox"/> Respiratory therapy      |
| <input type="checkbox"/> Speech-Language therapy                     | <input type="checkbox"/> Surgery                  |
| <input type="checkbox"/> Transplant                                  | <input type="checkbox"/> Other                    |

Specify other current health services-other, specify \_\_\_\_\_

Specify type of primary care provider  Unknown  Family practice  Internal medicine  Pediatrics

Name of primary care provider \_\_\_\_\_

Preventive care status

- Unknown  None
- On schedule for preventative care services  Behind schedule for preventative care services

Type of transplant service  Unknown  Evaluated for transplant  Received transplantTransplant organ  Unknown  Heart  Kidney  Liver  Lung  Stem cell  Other

Transplant organ-other, specify \_\_\_\_\_

Community resources currently received  Unknown  Yes  No



Patient Name \_\_\_\_\_

Date      

Specify current community resources

- Unknown  
 Daycare  
 Early childhood family education  
 Family support  
 Head Start  
 Medical Home  
 Nutritional services (WIC/MAC)  
 Personal care attendant (PCA)  
 Preschool  
 Respite care  
 Social media  
 Social services  
 Waivered services (CAC/CADI waiver/other waivers)  
 Other

Specify current community resources-other, specify \_\_\_\_\_

Specify current family support    Unknown    Family support related to this condition    Other

Specify current family support-other, specify \_\_\_\_\_

Specify medical home    Unknown    Primary care center    Speciality care center    Other

Specify medical home-other, specify \_\_\_\_\_

Specify current social services

- Unknown                       County                       Developmental disability  
 Medical                       Mental health                       Other

Specify current social services-other, specify \_\_\_\_\_

## Emergency Management

---

Patient currently has emergency specialty contact information

 Unknown                       Yes                       No

 Not needed for this condition

Type of emergency contact information

- Web-based                       Letter                       Sick day plan                       Alert accessory  
 Contact information    Other

Type of emergency contact information-other, specify \_\_\_\_\_

## Developmental Assessment

---

Developmental assessment done at this visit    Unknown    Yes    NoStandardized developmental screening tool(s) used    Unknown    Yes    No

Patient Name \_\_\_\_\_

Date      Developmental status  Typical  AtypicalSeverity of atypical development  Unknown  Mild delay  Moderate delay  Severe delay

Developmental milestones that were achieved in a typical order and timeframe

 Unknown  None  Cognitive  Fine motor  Gross motor Social-emotional  Speech-language  OtherDevelopmental milestones that were achieved in typical order and timeframe-other, specify  
\_\_\_\_\_

Referred for further developmental assessment

- Unknown  Yes
- No  Previously referred
- Family declined further assessments

Type of provider/service to whom patient was referred for developmental assessment

- Unknown  Developmental/behavioral pediatrician
- Neuropsychologist  Psychiatric APRN/CNP/CNS
- Psychiatrist  Psychologist
- School psychologist  Other

Type of provider/service to whom patient was referred for developmental assessment-other, specify  
\_\_\_\_\_Neuropsychometric evaluation performed since last visit  Unknown  Yes  NoOverall neuropsychometric impression  Above average  Average  Below average  In progressPatient has mental health concerns  Unknown  Yes  No

Referred for further mental health assessment

- Unknown  Yes
- No  Previously referred
- Family declined further assessments

Type of provider/service to whom patient was referred for mental health assessment

- Unknown  Developmental/behavioral pediatrician
- Marriage and family therapist  Mental health counselor
- Neuropsychologist  Pastoral counselor
- Psychiatric APRN/CNP/CNS  Psychiatrist
- Psychologist  School psychologist
- Social worker  Other

Type of provider/service to whom patient was referred for mental health assessment-other, specify  
\_\_\_\_\_Behavioral concerns  Unknown  Yes  No

Patient Name \_\_\_\_\_

Date |M|M| |D|D| |Y|Y|

Referred for further behavioral assessment  Unknown  Yes  No

Referred for further behavioral assessment-Explain

 Previously referred Family declined further assessments

Type of provider/service to whom patient was referred for behavioral assessment

 Unknown Developmental/behavioral pediatrician Marriage and family therapist Mental health counselor Neuropsychologist Psychiatric APRN/CNP/CNS Psychiatrist Psychologist School psychologist Social worker Other

Type of provider/service to whom patient was referred for behavioral assessment-other, specify \_\_\_\_\_

**Education**

Special education assessment recommended

 Unknown Yes No Special education services already received

Reason special education services received

 Unknown Cognitive disability Developmental delay Fine motor disability Gross motor disability Learning disability Social-emotional disability Speech/Language disability Other health impairment (OHI) Other

Reason special education services received-other, specify \_\_\_\_\_

Special education category

 Unknown Autism spectrum disorders Blind-visually impaired Deaf and hard of hearing Deaf-Blind Developmental cognitive disabilities: mild-moderate Developmental cognitive disabilities: severe- profound Developmental delay Emotional/Behavioral disorders Physically impaired Severely multiply impaired Specific learning disabilities

Patient Name \_\_\_\_\_

Date

Speech or language impairments

Traumatic brain injury

Other health disabilities

Special education, other- specify \_\_\_\_\_

**Comments**

---

Visit ancillary care comments

Patient Name \_\_\_\_\_

Date      

## CACT - Visit Lab Studies

### Biochemical Labs

Biochemical labs associated with this visit

- |  |   |
|--|---|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> None   |
| <input type="checkbox"/> Blood spot acylcarnitine profile    | <input type="checkbox"/> Carnitine:acylcarnitine translocase enzyme assay |
| <input type="checkbox"/> Creatinine kinase                   | <input type="checkbox"/> Fatty acid oxidation probe assay                 |
| <input type="checkbox"/> Plasma acylcarnitine profile        | <input type="checkbox"/> Plasma carnitine                                 |
| <input type="checkbox"/> Plasma essential fatty acid profile | <input type="checkbox"/> Triglycerides                                    |
| <input type="checkbox"/> Urine myoglobin                     | <input type="checkbox"/> Urine organic acids                              |
| <input type="checkbox"/> Western blot                        | <input type="checkbox"/> Other  |

Blood spot acylcarnitine profile

- Within normal limits    Abnormal    In progress    Results unavailable

Blood spot acylcarnitine profile comments

Please complete information about the carnitine:acylcarnitine translocase enzyme assay on the Intake Initial Testing form.

Creatinine kinase    Within normal limits    Abnormal    In progress    Results unavailable

Creatinine kinase value \_\_\_\_\_

Creatinine kinase units    U/L    umol/L    mg/dL

Creatinine kinase reference range \_\_\_\_\_

Please complete information about the fatty acid oxidation probe assay on the Intake Initial Testing form.

Plasma acylcarnitine profile    Within normal limits    Abnormal    In progress    Results unavailable

Plasma acylcarnitine profile comments

Plasma carnitine levels

- |   |  |  |
|---|--|--|
| <input type="checkbox"/> Unknown              | <input type="checkbox"/> Free carnitine                  | <input type="checkbox"/> Total carnitine |
| <input type="checkbox"/> Esterified carnitine | <input type="checkbox"/> Esterified:Free carnitine ratio |  |

Free plasma carnitine    Within normal limits    Abnormal    In progress    Results unavailable

Free plasma carnitine value \_\_\_\_\_

Patient Name \_\_\_\_\_

Date      Free plasma carnitine units  umol/L  mg/dL

Free plasma carnitine reference range \_\_\_\_\_

Total plasma carnitine  Within normal limits  Abnormal  In progress  Results unavailable

Total plasma carnitine value \_\_\_\_\_

Total plasma carnitine units  umol/L  mg/dL

Total plasma carnitine reference range \_\_\_\_\_

Esterified plasma carnitine  Within normal limits  Abnormal  In progress  Results unavailable

Esterified plasma carnitine value \_\_\_\_\_

Esterified plasma carnitine units  umol/L  mg/dL

Esterified plasma carnitine reference range \_\_\_\_\_

Esterified:Free plasma carnitine ratio

 Within normal limits  Abnormal  In progress  Results unavailable

Esterified:Free plasma carnitine ratio value \_\_\_\_\_

Esterified:Free plasma carnitine ratio reference range \_\_\_\_\_

Plasma essential fatty acid profile

- Unknown  Alpha linolenic acid (C18:3w3)  Docosahexaenoic acid (DHA)  
 EPA (C20:5w3)  Linoleic acid (C18:2w6)  Triene/tetraene ratio  
 Total fatty acids  Other

Alpha linolenic acid  Within normal limits  Abnormal  In progress  Results unavailable

Alpha linolenic acid value \_\_\_\_\_

Alpha linolenic acid units  nmol/L  ng/mL  nmol/mL

Alpha linoleic acid reference range \_\_\_\_\_

Docosahexaenoic acid (DHA) acid

 Within normal limits  Abnormal  In progress  Results unavailable

Docosahexaenoic acid (DHA) acid value \_\_\_\_\_

Docosahexaenoic acid (DHA) acid units  nmol/L  ng/mL  nmol/mL

Docosahexaenoic acid (DHA) acid reference range \_\_\_\_\_

EPA  Within normal limits  Abnormal  In progress  Results unavailable

EPA value \_\_\_\_\_

EPA units  nmol/L  ng/mL  nmol/mL

EPA reference range \_\_\_\_\_

Linoleic acid  Within normal limits  Abnormal  In progress  Results unavailable

Linoleic acid value \_\_\_\_\_

Linoleic acid units  nmol/L  ng/mL  nmol/mL

Linoleic acid reference range \_\_\_\_\_

Patient Name \_\_\_\_\_

Date

Triene/tetraene ratio  Within normal limits  Abnormal  In progress  Results unavailable

Triene/tetraene ratio value \_\_\_\_\_

Triene/tetraene ratio reference range \_\_\_\_\_

Total fatty acids  Within normal limits  Abnormal  In progress  Results unavailable

Total fatty acids comments \_\_\_\_\_

Other plasma essential fatty acid \_\_\_\_\_

Other plasma essential fatty acid result

Within normal limits  Abnormal  In progress  Results unavailable

Other plasma essential fatty acid value \_\_\_\_\_

Other plasma essential fatty acid units \_\_\_\_\_

Other plasma essential fatty acid reference range \_\_\_\_\_

Other plasma essential fatty acid comments

Triglyceride condition  Fasting  Non-fasting

Triglycerides  Within normal limits  Abnormal  In progress  Results unavailable

Triglyceride value \_\_\_\_\_

Triglyceride units  mmol/L  mg/dL

Triglyceride reference range \_\_\_\_\_

Urine myoglobin  Within normal limits  Abnormal  In progress  Results unavailable

Urine myoglobin value \_\_\_\_\_

Urine myoglobin units  nmol/L  ug/L

Urine myoglobin reference range \_\_\_\_\_

Urine organic acids  Within normal limits  Abnormal  In progress  Results unavailable

Urine organic acid comments

Western blot  Unknown  < 20%  >=20% < 50%  >=50%

Biochemical lab associated with this visit-other, specify \_\_\_\_\_

Other visit lab  Within normal limits  Abnormal  In progress  Results unavailable

Patient Name \_\_\_\_\_

Date

Other visit lab comments

Other visit lab value \_\_\_\_\_

Other visit lab units \_\_\_\_\_

Other visit lab reference range \_\_\_\_\_

Genetic testing performed for patient, sibling(s), or parent(s) since last visit    Unknown    Yes    No

Genetic testing information updated on the Intake Initial Testing form.    Unknown    Yes    No

### Chemistry Labs

---

Chemistry labs associated with this visit

- |   |  |  |
|---|--|--|
| <input type="checkbox"/> Unknown                    | <input type="checkbox"/> None                  | <input type="checkbox"/> Arterial blood gas          |
| <input type="checkbox"/> Anion gap                  | <input type="checkbox"/> Ammonia               | <input type="checkbox"/> Calcium                     |
| <input type="checkbox"/> Chloride                   | <input type="checkbox"/> Carbon dioxide        | <input type="checkbox"/> Glucose                     |
| <input type="checkbox"/> Potassium                  | <input type="checkbox"/> Lactate dehydrogenase | <input type="checkbox"/> Magnesium                   |
| <input type="checkbox"/> Sodium                     | <input type="checkbox"/> Phosphorous           | <input type="checkbox"/> Total cholesterol (fasting) |
| <input type="checkbox"/> Total cholesterol (random) | <input type="checkbox"/> Uric acid             | <input type="checkbox"/> Other                       |

Arterial blood gas    Within normal limits    Abnormal    In progress    Results unavailable

Arterial blood gas comments

Arterial blood gas value \_\_\_\_\_

Arterial blood gas units    mmHg    mEq/L

Arterial blood gas reference range \_\_\_\_\_

Anion gap    Within normal limits    Abnormal    In progress    Results unavailable

Anion gap value \_\_\_\_\_

Anion gap units    mEq/L    mmol/L    umol/L    nmol/L

Anion gap reference range \_\_\_\_\_

Ammonia    Within normal limits    Abnormal    In progress    Results unavailable

Ammonia value \_\_\_\_\_

Ammonia units    umol/L    ug/dL

Ammonia reference range \_\_\_\_\_

Calcium    Within normal limits    Abnormal    In progress    Results unavailable

Calcium value \_\_\_\_\_



Patient Name \_\_\_\_\_

Date      Calcium units  mmol/L  mg/dL

Calcium reference range \_\_\_\_\_

Chloride  Within normal limits  Abnormal  In progress  Results unavailable

Chloride value \_\_\_\_\_

Chloride units  mmol/L  mg/L

Chloride reference range \_\_\_\_\_

Carbon dioxide  Within normal limits  Abnormal  In progress  Results unavailable

Carbon dioxide value \_\_\_\_\_

Carbon dioxide units  mEq/L  mmol/L

Carbon dioxide reference range \_\_\_\_\_

Glucose  Within normal limits  Abnormal  In progress  Results unavailable

Glucose value \_\_\_\_\_

Glucose units  mmol/L  mg/dL

Glucose reference range \_\_\_\_\_

Potassium  Within normal limits  Abnormal  In progress  Results unavailable

Potassium value \_\_\_\_\_

Potassium units  mEq/L  mmol/L

Potassium reference range \_\_\_\_\_

Lactate dehydrogenase  Within normal limits  Abnormal  In progress  Results unavailable

Lactate dehydrogenase value \_\_\_\_\_

Lactate dehydrogenase units  U/L

Lactate dehydrogenase reference range \_\_\_\_\_

Magnesium  Within normal limits  Abnormal  In progress  Results unavailable

Magnesium value \_\_\_\_\_

Magnesium units  mmol/L  mg/dL

Magnesium reference range \_\_\_\_\_

Sodium  Within normal limits  Abnormal  In progress  Results unavailable

Sodium value \_\_\_\_\_

Sodium units  mEq/L  mmol/L

Sodium reference range \_\_\_\_\_

Phosphorous  Within normal limits  Abnormal  In progress  Results unavailable

Phosphorous value \_\_\_\_\_

Phosphorous units  mmol/L  mg/dL

Phosphorous reference range \_\_\_\_\_

Total Cholesterol (fasting)  Within normal limits  Abnormal  In progress  Results unavailable

Patient Name \_\_\_\_\_

Date

Total Cholesterol (fasting) value \_\_\_\_\_

Total Cholesterol (fasting) units  mmol/L  mg/dL

Total Cholesterol (fasting) reference range \_\_\_\_\_

Total Cholesterol (random)  Within normal limits  Abnormal  In progress  Results unavailable

Total Cholesterol (random) value \_\_\_\_\_

Total Cholesterol (random) units  mmol/L  mg/dL

Total Cholesterol (random) reference range \_\_\_\_\_

Uric acid  Within normal limits  Abnormal  In progress  Results unavailable

Uric acid value \_\_\_\_\_

Uric acid units  umol/L  mg/dL

Uric acid reference range \_\_\_\_\_

Chemistry labs-other, specify \_\_\_\_\_

Other chemistry labs  Within normal limits  Abnormal  In progress  Results unavailable

Other chemistry lab comments

Other chemistry lab value \_\_\_\_\_

Other chemistry lab units \_\_\_\_\_

Other chemistry lab reference range \_\_\_\_\_

### Hematology Labs

---

Hematology labs associated with this visit

- |  |  |   |
|--|--|---|
| <input type="checkbox"/> Unknown                     | <input type="checkbox"/> None                  | <input type="checkbox"/> Fibrinogen                     |
| <input type="checkbox"/> Hematocrit                  | <input type="checkbox"/> Hemoglobin            | <input type="checkbox"/> International Normalized Ratio |
| <input type="checkbox"/> Partial Thromboplastin Time | <input type="checkbox"/> Prothrombin time test | <input type="checkbox"/> Peripheral blood smear         |
| <input type="checkbox"/> Platelet count              | <input type="checkbox"/> Red blood cell count  | <input type="checkbox"/> White blood cell count         |
| <input type="checkbox"/> Other                       |  |   |

Fibrinogen  Within normal limits  Abnormal  In progress  Results unavailable

Fibrinogen value \_\_\_\_\_

Fibrinogen units  umol/L  mg/dL

Fibrinogen reference range \_\_\_\_\_

Hematocrit  Within normal limits  Abnormal  In progress  Results unavailable

Hematocrit value \_\_\_\_\_

Hematocrit units  %  Proportion of total hemoglobin

Hematocrit reference range \_\_\_\_\_

Patient Name \_\_\_\_\_

Date      Hemoglobin  Within normal limits  Abnormal  In progress  Results unavailable

Hemoglobin value \_\_\_\_\_

Hemoglobin units  g/dL  g/L

Hemoglobin reference range \_\_\_\_\_

International Normalized Ratio (INR)

 Within normal limits  Abnormal  In progress  Results unavailable

International Normalized Ratio (INR) value \_\_\_\_\_

International Normalized Ratio (INR) reference range \_\_\_\_\_

Partial Thromboplastin Time (PTT)

 Within normal limits  Abnormal  In progress  Results unavailable

Partial Thromboplastin Time (PTT) value \_\_\_\_\_

Partial Thromboplastin Time (PTT) units  seconds

Partial Thromboplastin Time (PTT) reference range \_\_\_\_\_

Prothrombin time test  Within normal limits  Abnormal  In progress  Results unavailable

Prothrombin time test value \_\_\_\_\_

Prothrombin time test units  seconds

Prothrombin time test reference range \_\_\_\_\_

Peripheral blood smear  Within normal limits  Abnormal  In progress  Results unavailable

Peripheral blood smear value \_\_\_\_\_

Peripheral blood smear units  platelet/RBC/WBC count

Peripheral blood smear reference range \_\_\_\_\_

Platelet count  Within normal limits  Abnormal  In progress  Results unavailable

Platelet count value \_\_\_\_\_

Platelet count units  THOU/uL   $10^9/L$    $10^3/ul$   k/uL

Platelet count reference range \_\_\_\_\_

Red blood cell count  Within normal limits  Abnormal  In progress  Results unavailable

Red blood cell count value \_\_\_\_\_

Red blood cell count units   $10^6/uL$    $10^{12}/uL$ 

Red blood cell count reference range \_\_\_\_\_

White blood cell count  Within normal limits  Abnormal  In progress  Results unavailable

White blood cell count value \_\_\_\_\_

White blood cell count units   $10^3/uL$    $10^9/uL$ 

White blood cell count reference range \_\_\_\_\_

Hematology labs-other, specify \_\_\_\_\_

Other hematology labs  Within normal limits  Abnormal  In progress  Results unavailable

Patient Name \_\_\_\_\_

Date      

Other hematology lab comments

Other hematology lab value \_\_\_\_\_

Other hematology lab units \_\_\_\_\_

Other hematology lab reference range \_\_\_\_\_

## Liver Labs

---

Liver labs associated with this visit

- |   |  |   |
|---|--|---|
| <input type="checkbox"/> Unknown              | <input type="checkbox"/> None                          | <input type="checkbox"/> Albumin                    |
| <input type="checkbox"/> Alkaline phosphatase | <input type="checkbox"/> Alanine aminotransferase      | <input type="checkbox"/> Aspartate aminotransferase |
| <input type="checkbox"/> Direct bilirubin     | <input type="checkbox"/> Gamma-glutamyl transpeptidase | <input type="checkbox"/> Globulin                   |
| <input type="checkbox"/> Prealbumin           | <input type="checkbox"/> Total bilirubin               | <input type="checkbox"/> Total protein              |
| <input type="checkbox"/> Other                |  |   |

Albumin     Within normal limits     Abnormal     In progress     Results unavailable

Albumin value \_\_\_\_\_

Albumin units     g/dL     g/L     mg/L     mg/dL     g/mL     mg/mL     ug/L     ug/mL     ug/dL

Albumin reference range \_\_\_\_\_

Alkaline phosphatase     Within normal limits     Abnormal     In progress     Results unavailable

Alkaline phosphatase value \_\_\_\_\_

Alkaline phosphatase units     U/L     ukat/L

Alkaline phosphatase reference range \_\_\_\_\_

Alanine aminotransferase     Within normal limits     Abnormal     In progress     Results unavailable

Alanine aminotransferase value \_\_\_\_\_

Alanine aminotransferase units     U/L     ukat/L

Alanine aminotransferase reference range \_\_\_\_\_

Aspartate aminotransferase     Within normal limits     Abnormal     In progress     Results unavailable

Aspartate aminotransferase value \_\_\_\_\_

Aspartate aminotransferase units     U/L     ukat/L

Aspartate aminotransferase reference range \_\_\_\_\_

Direct bilirubin     Within normal limits     Abnormal     In progress     Results unavailable

Direct bilirubin value \_\_\_\_\_

Direct bilirubin units

- umol/L     mg/dL     nmol/L     mmol/L     umol/dL     umol/mL     nmol/dL  
 nmol/mL     mmol/mL     mg/L     g/dL     g/L

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

Direct bilirubin reference range \_\_\_\_\_

Gamma-glutamyl transpeptidase

Within normal limits    Abnormal                       In progress                       Results unavailable

Gamma-glutamyl transpeptidase value \_\_\_\_\_

Gamma-glutamyl transpeptidase units    IU/L    U/L

Gamma-glutamyl transpeptidase reference range \_\_\_\_\_

Globulin    Within normal limits    Abnormal    In progress    Results unavailable

Globulin value \_\_\_\_\_

Globulin units    g/dL    g/L

Globulin reference range \_\_\_\_\_

Prealbumin    Within normal limits    Abnormal    In progress    Results unavailable

Prealbumin value \_\_\_\_\_

Prealbumin units    mg/dL    mg/L

Prealbumin reference range \_\_\_\_\_

Total bilirubin    Within normal limits    Abnormal    In progress    Results unavailable

Total bilirubin value \_\_\_\_\_

Total bilirubin units    umol/L    mg/dL

Total bilirubin reference range \_\_\_\_\_

Total protein    Within normal limits    Abnormal    In progress    Results unavailable

Total protein value \_\_\_\_\_

Total protein units    g/dL    g/L

Total protein reference range \_\_\_\_\_

Liver labs-other, specify \_\_\_\_\_

Other liver labs    Within normal limits    Abnormal    In progress    Results unavailable

Other liver lab comments

Other liver lab value \_\_\_\_\_

Other liver lab units \_\_\_\_\_

Other liver lab reference range \_\_\_\_\_

**Renal Labs**

Renal labs associated with this visit

- |  |   |
|--|---|
| <input type="checkbox"/> Unknown             | <input type="checkbox"/> None             |
| <input type="checkbox"/> Blood urea nitrogen | <input type="checkbox"/> Serum creatinine |

Patient Name \_\_\_\_\_

Date       24-hour creatinine clearance Nuclear medicine glomerular filtration rate OtherBlood urea nitrogen  Within normal limits  Abnormal  In progress  Results unavailable

Blood urea nitrogen value \_\_\_\_\_

Blood urea nitrogen units  mmol/L  mg/dL

Blood urea nitrogen reference range \_\_\_\_\_

Serum creatinine  Within normal limits  Abnormal  In progress  Results unavailable

Serum creatinine value \_\_\_\_\_

Serum creatinine units  umol/L  mg/dL

Serum creatinine reference range \_\_\_\_\_

24-hour creatinine clearance  Within normal limits  Abnormal  In progress  Results unavailable

24-hour creatinine clearance value \_\_\_\_\_

24-hour creatinine clearance units  mL/min  mL/s

24-hour creatinine clearance reference range \_\_\_\_\_

Nuclear medicine glomerular filtration rate

 Within normal limits  Abnormal  In progress  Results unavailable

Nuclear medicine glomerular filtration rate value \_\_\_\_\_

Nuclear medicine glomerular filtration rate units  mL/min/1.73m<sup>2</sup>  mL/min/1.73m<sup>3</sup>

Nuclear medicine glomerular filtration rate reference range \_\_\_\_\_

Renal labs-other, specify \_\_\_\_\_

Other renal labs  Within normal limits  Abnormal  In progress  Results unavailable

Other renal lab comments

Other renal lab value \_\_\_\_\_

Other renal lab units \_\_\_\_\_

Other renal lab reference range \_\_\_\_\_

## Miscellaneous Labs

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Miscellaneous labs associated with this visit

 Unknown None B-type natriuretic peptide Creatine phosphokinase C-reactive protein Erythrocyte sedimentation rate Ferritin Thyroid stimulating hormone Thyroxine (free) Thyroxine (total) Transferrin Troponin Urinalysis Zinc Other

Patient Name \_\_\_\_\_

Date      B-type natriuretic peptide  Within normal limits  Abnormal  In progress  Results unavailable

B-type natriuretic peptide value \_\_\_\_\_

B-type natriuretic peptide units  pg/mL  ng/L

B-type natriuretic peptide reference range \_\_\_\_\_

Creatine phosphokinase  Within normal limits  Abnormal  In progress  Results unavailable

Creatine phosphokinase value \_\_\_\_\_

Creatine phosphokinase units  U/L  umol/L  mg/dL

Creatine phosphokinase reference range \_\_\_\_\_

C-reactive protein  Within normal limits  Abnormal  In progress  Results unavailable

C-reactive protein value \_\_\_\_\_

C-reactive protein units  nmol/L  mg/dL

C-reactive protein reference range \_\_\_\_\_

Erythrocyte sedimentation rate  Within normal limits  Abnormal  In progress  Results unavailable

Erythrocyte sedimentation rate value \_\_\_\_\_

Erythrocyte sedimentation rate units  mm/h

Erythrocyte sedimentation rate reference range \_\_\_\_\_

Ferritin  Within normal limits  Abnormal  In progress  Results unavailable

Ferritin value \_\_\_\_\_

Ferritin units  pmol/L  ng/mL

Ferritin reference range \_\_\_\_\_

Thyroid-stimulating hormone  Within normal limits  Abnormal  In progress  Results unavailable

Thyroid-stimulating hormone value \_\_\_\_\_

Thyroid-stimulating hormone units  mIU/L

Thyroid-stimulating hormone reference range \_\_\_\_\_

Thyroxine (free)  Within normal limits  Abnormal  In progress  Results unavailable

Thyroxine (free) value \_\_\_\_\_

Thyroxine (free) units  pmol/L  ng/mL

Thyroxine (free) reference range \_\_\_\_\_

Thyroxine (total)  Within normal limits  Abnormal  In progress  Results unavailable

Thyroxine (total) value \_\_\_\_\_

Thyroxine (total) units  pmol/L  ng/mL

Thyroxine (total) reference range \_\_\_\_\_

Transferrin  Within normal limits  Abnormal  In progress  Results unavailable

Transferrin value \_\_\_\_\_

Transferrin units  mg/dL  g/L

Patient Name \_\_\_\_\_

Date

Transferrin reference range \_\_\_\_\_

Troponin  Within normal limits  Abnormal  In progress  Results unavailable

Troponin value \_\_\_\_\_

Troponin units  ng/mL  ug/L

Troponin reference range \_\_\_\_\_

Urinalysis  Within normal limits  Abnormal  In progress  Results unavailable

Urinalysis comments

Zinc  Within normal limits  Abnormal  In progress  Results unavailable

Zinc value \_\_\_\_\_

Zinc units  umol/L  ug/dL

Zinc reference range \_\_\_\_\_

Miscellaneous labs-other, specify \_\_\_\_\_

Other miscellaneous labs  Within normal limits  Abnormal  In progress  Results unavailable

Other miscellaneous lab comments

Other miscellaneous lab value \_\_\_\_\_

Other miscellaneous lab units \_\_\_\_\_

Other miscellaneous lab reference range \_\_\_\_\_

**Comments**

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Visit lab studies comments



Patient Name \_\_\_\_\_

Date |||||

## CACT - Visit Studies Other

### Home Monitoring

Home monitoring recommended Unknown Yes NoHome monitoring done since the last outpatient visit Unknown Yes No

Type of home monitoring

- Glucose by glucometer Branched chain amino acids (BCAA)  
Phenylalanine Tyrosine  
Urine dinitrophenylhydrazine (DNPH) Urine dipstick for ketones  
Urine dipstick for myoglobin/blood Other

#### Glucose

Frequency of glucose home monitoring Unknown Only when symptomatic Routinely

Specify routine frequency\_\_\_\_\_

Number of glucose home monitoring samples reported since last outpatient metabolic visit\_\_\_\_\_

Lowest reported glucose value on home monitoring samples since last outpatient metabolic visit

\_\_\_\_\_

Lowest reported glucose value on home monitoring samples since last outpatient metabolic visit units

mmol/L mg/dL

#### Branched Chain Amino Acids

Type of branched chain amino acid home monitoring

- Unknown Alloisoleucine Isoleucine Leucine Valine

Sample type of branched chain amino acid home monitoring

- Unknown Filter paper blood Plasma

Frequency of branched chain amino acid home monitoring

- Unknown Only when symptomatic Routinely

Specify routine frequency\_\_\_\_\_

Number of branched chain amino acid home monitoring samples reported since last outpatient metabolic visit\_\_\_\_\_

#### Alloisoleucine

Recommended therapeutic alloisoleucine goal\_\_\_\_\_

Recommended therapeutic alloisoleucine goal units umol/dL umol/L mg/dL

Average alloisoleucine on home monitoring samples\_\_\_\_\_

Average alloisoleucine on home monitoring samples units umol/dL umol/L mg/dL

Average percent above recommended alloisoleucine therapeutic goal\_\_\_\_\_

Patient Name \_\_\_\_\_

Date      

Average percent below recommended alloisoleucine therapeutic goal \_\_\_\_\_

**Isoleucine**

Recommended therapeutic isoleucine goal \_\_\_\_\_

Recommended therapeutic isoleucine goal units umol/dL umol/L mg/dL

Average isoleucine on home monitoring samples \_\_\_\_\_

Average isoleucine on home monitoring samples units umol/dL umol/L mg/dL

Average percent above recommended isoleucine therapeutic goal \_\_\_\_\_

Average percent below recommended isoleucine therapeutic goal \_\_\_\_\_

**Leucine**

Recommended therapeutic leucine goal \_\_\_\_\_

Recommended therapeutic leucine goal units umol/dL umol/L mg/dL

Highest leucine value on home monitoring samples \_\_\_\_\_

Highest leucine value on home monitoring samples units umol/dL umol/L mg/dL

Average leucine on home monitoring samples \_\_\_\_\_

Average leucine on home monitoring samples units umol/dL umol/L mg/dL

Average percent above recommended leucine therapeutic goal \_\_\_\_\_

Average percent below recommended leucine therapeutic goal \_\_\_\_\_

**Valine**

Recommended therapeutic valine goal \_\_\_\_\_

Recommended therapeutic valine goal units umol/dL umol/L mg/dL

Average valine on home monitoring samples \_\_\_\_\_

Average valine on home monitoring samples units umol/dL umol/L mg/dL

Average percent above recommended valine therapeutic goal \_\_\_\_\_

Average percent below recommended valine therapeutic goal \_\_\_\_\_

**Phenylalanine**Sample type of phenylalanine home monitoring Unknown Filter paper blood PlasmaFrequency of phenylalanine home monitoring Unknown Only when symptomatic Routinely

Specify routine frequency \_\_\_\_\_

Recommended therapeutic phenylalanine goal \_\_\_\_\_

Recommended therapeutic phenylalanine goal units umol/dL umol/L mg/dL

Highest phenylalanine value on home monitoring samples \_\_\_\_\_

Highest phenylalanine value on home monitoring samples units umol/dL umol/L mg/dL

Average phenylalanine on home monitoring samples \_\_\_\_\_

Average phenylalanine on home monitoring samples units umol/dL umol/L mg/dL

Average percent above recommended phenylalanine therapeutic goal \_\_\_\_\_

Patient Name \_\_\_\_\_

Date      

Average percent below recommended phenylalanine therapeutic goal \_\_\_\_\_

Number of phenylalanine home monitoring samples reported since last outpatient metabolic visit \_\_\_\_\_

**Tyrosine**Sample type of tyrosine home monitoring  Unknown  Filter paper blood  PlasmaFrequency of tyrosine home monitoring  Unknown  Only when symptomatic  Routinely

Specify routine frequency \_\_\_\_\_

Recommended therapeutic tyrosine goal \_\_\_\_\_

Recommended therapeutic tyrosine goal units  umol/dL  umol/L  mg/dL

Lowest tyrosine value on home monitoring samples \_\_\_\_\_

Lowest tyrosine value on home monitoring samples units  umol/dL  umol/L  mg/dL

Average tyrosine on home monitoring samples \_\_\_\_\_

Average tyrosine on home monitoring samples units  umol/dL  umol/L  mg/dL

Average percent below recommended tyrosine therapeutic goal \_\_\_\_\_

Number of tyrosine home monitoring samples reported since last outpatient metabolic visit \_\_\_\_\_

**Dinitrophenylhydrazine**

Frequency of dinitrophenylhydrazine home monitoring

 Unknown Only when symptomatic Routinely

Specify routine frequency \_\_\_\_\_

Number of dinitrophenylhydrazine home monitoring samples reported since last outpatient metabolic visit \_\_\_\_\_

Result of dinitrophenylhydrazine home monitoring samples

 Unknown Precipitate present Precipitate absent**Ketones**Highest level of ketones  Unknown  None  Trace  Small  Moderate  LargeFrequency of ketones home monitoring  Unknown  Only when symptomatic  Routinely

Specify routine frequency \_\_\_\_\_

Number of ketones home monitoring samples reported since last outpatient metabolic visit \_\_\_\_\_

**Myoglobin/blood**Highest level of myoglobin/blood  Unknown  None  Trace  Small  Moderate  LargeFrequency of myoglobin/blood home monitoring  Unknown  Only when symptomatic  Routinely

Specify routine frequency \_\_\_\_\_

Number of myoglobin/blood home monitoring samples reported since last outpatient metabolic visit \_\_\_\_\_

**Other**

Other type of home monitoring, specify \_\_\_\_\_

Frequency of other home monitoring  Unknown  Only when symptomatic  Routinely

Patient Name \_\_\_\_\_

Date

Specify routine frequency \_\_\_\_\_

Number of other home monitoring samples reported since last outpatient metabolic visit \_\_\_\_\_

**Other Studies**

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Physiological tests associated with this visit

- Unknown  None  Electrocardiogram (ECG)
- Electroencephalography (EEG)  Electromyography (EMG)  Holter monitoring
- Cardiac stress test

**Electrocardiogram**

Electrocardiogram date

Electrocardiogram findings of note

**Electroencephalography**

Electroencephalography date

Electroencephalography findings of note

**Electromyography**

Electromyography date

Electromyography findings of note

**Holter Monitoring**

Holter monitoring date

Holter monitoring findings of note

**Cardiac Stress Test**

Cardiac stress test date

Patient Name \_\_\_\_\_

Date

Cardiac stress test findings of note

**Imaging Studies**

Imaging studies associated with this visit

- Unknown       None       Abdominal       Cardiac
- Musculoskeletal       Neurological       Renal/pelvic/genital       Other

Abdominal imaging

- Unknown       CT WNL       CT Abn       MRI WNL       MRI Abn
- Ultrasound WNL       Ultrasound Abn       X-ray WNL       X-ray Abn

Abdominal findings of note

Cardiac Imaging

- Unknown       Chest x-ray WNL       Chest x-ray Abn       Echocardiogram WNL
- Echocardiogram Abn       Other

Cardiac findings of note

Cardiomyopathy since last visit     Unknown     Yes     No

Cardiomyopathy has worsened/progressed since last visit     Unknown     Yes     No

Current treatments for cardiomyopathy

Musculoskeletal imaging

- Unknown       Bone scan WNL       Bone scan Abn       CT WNL       CT Abn
- MRI WNL       MRI Abn       Ultrasound WNL       Ultrasound Abn       X-rays WNL
- X-rays Abn       Other

Patient Name \_\_\_\_\_

Date

Musculoskeletal findings of note

Neurological imaging

- Unknown
- Cranial ultrasound Abn
- Head CT Abn
- Head MRI Abn
- Positron emission tomography (PET) scan Abn
- Cranial ultrasound WNL
- Head CT WNL
- Head MRI WNL
- Positron emission tomography (PET) scan WNL

Evidence of abnormal myelination on CNS imaging    Unknown    Yes    No

Neurological findings of note

Renal/Pelvic/Genital imaging

- Unknown
- Genitogram WNL
- Genitogram Abn
- Nuclear medicine dimercapto succinic acid (DMSA) renal scan WNL
- Nuclear medicine dimercapto succinic acid (DMSA) renal scan Abn
- Pelvic ultrasound WNL
- Pelvic ultrasound Abn
- Renal ultrasound WNL
- Renal ultrasound Abn
- Testicular ultrasound WNL
- Testicular ultrasound Abn
- VCUG WNL
- VCUG Abn
- Other

Renal/pelvic/genital findings of note

Imaging studies-other, specify \_\_\_\_\_

Patient Name \_\_\_\_\_

Date

Other imaging studies date

Other imaging studies explanation

### Eye Exam

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Eye exam associated with this visit    Unknown    Yes    No

Eye exam findings    Unknown    Within normal limits    Evidence of nystagmus    Other

Eye exam findings-other, specify \_\_\_\_\_

### Comments

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Visit studies-other comments

Patient Name \_\_\_\_\_

Date      

## CACT - Visit Management And Treatment Pharmacotherapy

### Pharmacotherapy

#### Medications

- |   |  |   |
|---|--|---|
| <input type="checkbox"/> Unknown                      | <input type="checkbox"/> None                | <input type="checkbox"/> Analgesics           |
| <input type="checkbox"/> Antacids                     | <input type="checkbox"/> Antianxiety         | <input type="checkbox"/> Antibiotics          |
| <input type="checkbox"/> Anticoagulants/Thrombolytics | <input type="checkbox"/> Anticonvulsants     | <input type="checkbox"/> Antidepressants      |
| <input type="checkbox"/> Antiemetics                  | <input type="checkbox"/> Antifungals         | <input type="checkbox"/> Antihistamines       |
| <input type="checkbox"/> Antihypertensives            | <input type="checkbox"/> Antiinflammatories  | <input type="checkbox"/> Antioxidants         |
| <input type="checkbox"/> Antipsychotics               | <input type="checkbox"/> Antipyretics        | <input type="checkbox"/> Antivirals           |
| <input type="checkbox"/> Aromatase inhibitor          | <input type="checkbox"/> Biphosphonates      | <input type="checkbox"/> Bronchodilators      |
| <input type="checkbox"/> Contraceptives-injections    | <input type="checkbox"/> Contraceptives-oral | <input type="checkbox"/> Corticosteroids      |
| <input type="checkbox"/> Diuretics                    | <input type="checkbox"/> Estrogen            | <input type="checkbox"/> GnRH Analog          |
| <input type="checkbox"/> Growth hormone               | <input type="checkbox"/> Immunosuppressives  | <input type="checkbox"/> Insulin              |
| <input type="checkbox"/> Insulin sensitizers          | <input type="checkbox"/> Iron                | <input type="checkbox"/> Laxatives            |
| <input type="checkbox"/> Mannitol                     | <input type="checkbox"/> Progesterone        | <input type="checkbox"/> Sleeping medications |
| <input type="checkbox"/> Testosterone                 | <input type="checkbox"/> Vitamins            | <input type="checkbox"/> Other                |

Other medications- specify \_\_\_\_\_

Homeopathic therapies  Unknown  Yes  No

Specify homeopathic therapies

#### Disease Treatment

Treatment recommended/prescribed

- |                                      |                                |  |
|--------------------------------------|--------------------------------|--|
| <input type="checkbox"/> Unknown     | <input type="checkbox"/> None  | <input type="checkbox"/> Glucose paste/gel/tablets |
| <input type="checkbox"/> L-carnitine | <input type="checkbox"/> Other |  |

#### Glucose paste/gel/tablets

Number of times patient has used glucose paste/gel/tablets since the last outpatient metabolic visit

- |                         |                         |                         |                          |                           |                               |                         |
|-------------------------|-------------------------|-------------------------|--------------------------|---------------------------|-------------------------------|-------------------------|
| <input type="radio"/> 0 | <input type="radio"/> 1 | <input type="radio"/> 2 | <input type="radio"/> 3  | <input type="radio"/> 4   | <input type="radio"/> 5       | <input type="radio"/> 6 |
| <input type="radio"/> 7 | <input type="radio"/> 8 | <input type="radio"/> 9 | <input type="radio"/> 10 | <input type="radio"/> >10 | <input type="radio"/> Unknown |                         |

#### L-carnitine

L-carnitine route recommended/prescribed  Unknown  Feeding tube  IV  Oral

L-carnitine dose recommended/prescribed \_\_\_\_\_



Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

L-carnitine dose recommended/prescribed units  g  mg

L-carnitine frequency recommended/prescribed

- Unknown
- Once/day
- Twice/day
- Three times/day
- Four times/day
- Other

L-carnitine frequency recommended/prescribed-other, specify \_\_\_\_\_

L-carnitine taken as recommended/prescribed  Unknown  Yes  No

Actual L-carnitine dose reported \_\_\_\_\_

Actual L-carnitine dose reported units  g  mg

Actual L-carnitine frequency reported

- Unknown
- Once/day
- Twice/day
- Three times/day
- Four times/day
- Other

Actual L-carnitine frequency reported-other, specify \_\_\_\_\_

Reason L-carnitine is not taken as recommended/prescribed \_\_\_\_\_

Method of payment for L-carnitine

- Unknown
- None
- Commercial/private
- Medicaid
- Medicare
- Military
- Newborn screening funds
- Patient assistance program
- Self-pay
- State Children's Health Insurance Program (SCHIP)
- State Children with Special Health Needs (CSHN) Program
- Other

**Other Treatment**

Other treatment recommend/prescribed \_\_\_\_\_

Other treatment route recommended/prescribed \_\_\_\_\_

Other treatment dose recommended/prescribed \_\_\_\_\_

Other treatment dose recommended/prescribed units \_\_\_\_\_

Other treatment frequency recommended/prescribed \_\_\_\_\_

Other treatment taken as recommended/prescribed  Unknown  Yes  No

Actual other treatment dose reported \_\_\_\_\_

Actual other treatment dose reported units \_\_\_\_\_

Patient Name _____
Date <input type="text"/>   <input type="text"/>   <input type="text"/>   <input type="text"/>   <input type="text"/>   <input type="text"/>

Actual other treatment frequency reported \_\_\_\_\_

Reason other treatment is not taken as recommended/prescribed \_\_\_\_\_

Method of payment for other treatment

- Unknown
- None
- Commercial/private
- Medicaid
- Medicare
- Military
- Newborn screening funds
- Patient assistance program
- Self-pay
- State Children's Health Insurance Program (SCHIP)
- State Children with Special Health Needs (CSHN) Program
- Other

**Comments**

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Medication and supplement comments

Patient Name \_\_\_\_\_

Date      

## CACT - Visit Management And Treatment Nutrition

### Nutrition

Mode of nutrition delivery   Unknown   Oral   NG tube   NJ tube   G-tube   GJ tube   TPN

Types of milk/formula taken

- |  |  |  |
|--|--|--|
| <input type="checkbox"/> Unknown               | <input type="checkbox"/> None                      | <input type="checkbox"/> Baby formula (regular)    |
| <input type="checkbox"/> Baby formula (soy)    | <input type="checkbox"/> Elemental formula         | <input type="checkbox"/> Breast milk               |
| <input type="checkbox"/> Human milk fortifier  | <input type="checkbox"/> Almond milk               | <input type="checkbox"/> Rice milk                 |
| <input type="checkbox"/> Skim milk             | <input type="checkbox"/> 1% milk                   | <input type="checkbox"/> 2% milk                   |
| <input type="checkbox"/> Soy milk              | <input type="checkbox"/> Special metabolic formula | <input type="checkbox"/> Toddler formula (regular) |
| <input type="checkbox"/> Toddler formula (soy) | <input type="checkbox"/> Whole milk                | <input type="checkbox"/> Other                     |

Number of special metabolic formulas recommended/prescribed   Unknown   1   2   3

Name of special metabolic formula 1 \_\_\_\_\_

Amount of special metabolic formula 1 (grams) recommended/prescribed per 24 hours \_\_\_\_\_

Special metabolic formula 1 taken as recommended/prescribed   Unknown   Yes   No

Actual frequency of use of special metabolic formula 1

- Unknown   0 days/week   1 day/week   2 days/week   3 days/week   4 days/week  
5 days/week   6 days/week

Reason special metabolic formula 1 is not taken as recommended/prescribed \_\_\_\_\_

Fat grams from metabolic formula 1 recommended/prescribed per 24 hours \_\_\_\_\_

Protein grams from metabolic formula 1 recommended/prescribed per 24 hours \_\_\_\_\_

Method of payment for special metabolic formula 1

- Unknown  
None  
Commercial/private  
Medicaid  
Medicare  
Military  
Newborn screening funds  
Patient assistance program  
Self-pay  
State Children's Health Insurance Program (SCHIP)  
State Children with Special Health Needs (CSHN) Program  
Other

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

Name of special metabolic formula 2 \_\_\_\_\_

Amount of special metabolic formula 2 (grams) recommended/prescribed per 24 hours \_\_\_\_\_

Special metabolic formula 2 taken as recommended/prescribed     Unknown     Yes     No

Actual frequency of use of special metabolic formula 2

Unknown     0 days/week     1 day/week     2 days/week     3 days/week     4 days/week

5 days/week     6 days/week

Reason special metabolic formula 2 is not taken as recommended/prescribed \_\_\_\_\_

Fat grams from metabolic formula 2 recommended/prescribed per 24 hours \_\_\_\_\_

Protein grams from metabolic formula 2 recommended/prescribed per 24 hours \_\_\_\_\_

Method of payment for special metabolic formula 2

Unknown

None

Commercial/private

Medicaid

Medicare

Military

Newborn screening funds

Patient assistance program

Self-pay

State Children's Health Insurance Program (SCHIP)

State Children with Special Health Needs (CSHN) Program

Other

Name of special metabolic formula 3 \_\_\_\_\_

Amount of special metabolic formula 3 (grams) recommended/prescribed per 24 hours \_\_\_\_\_

Special metabolic formula 3 taken as recommended/prescribed     Unknown     Yes     No

Actual frequency of use of special metabolic formula 3

Unknown     0 days/week     1 day/week     2 days/week     3 days/week     4 days/week

5 days/week     6 days/week

Reason special metabolic formula 3 is not taken as recommended/prescribed \_\_\_\_\_

Fat grams from metabolic formula 3 recommended/prescribed per 24 hours \_\_\_\_\_

Protein grams from metabolic formula 3 recommended/prescribed per 24 hours \_\_\_\_\_

Method of payment for special metabolic formula 3

Unknown

None

Commercial/private

Patient Name \_\_\_\_\_

Date      

- Medicaid
- Medicare
- Military
- Newborn screening funds
- Patient assistance program
- Self-pay
- State Children's Health Insurance Program (SCHIP)
- State Children with Special Health Needs (CSHN) Program
- Other

Type milk/formula, other- specify \_\_\_\_\_

**Cornstarch**Cornstarch recommended/prescribed  Unknown  Yes  NoCornstarch route recommended/prescribed  Unknown  Feeding tube  Oral

Cornstarch dose recommended/prescribed \_\_\_\_\_

Cornstarch dose recommended/prescribed units  gCornstarch frequency recommended/prescribed  Unknown  Once/day at bedtime  PRN  Other

Cornstarch frequency recommended/prescribed-other, specify \_\_\_\_\_

Cornstarch taken as recommended/prescribed  Unknown  Yes  No

Actual cornsarch dose reported \_\_\_\_\_

Actual cornsarch dose reported units  gActual cornstarch frequency reported  Unknown  Once/day at bedtime  PRN  Other

Actual cornstarch frequency reported-other, specify \_\_\_\_\_

Reason cornstarch is not taken as recommended/prescribed \_\_\_\_\_

## Method of payment for cornstarch

- Unknown
- None
- Commercial/private
- Medicaid
- Medicare
- Military
- Newborn screening funds
- Patient assistance program
- Self-pay
- State Children's Health Insurance Program (SCHIP)
- State Children with Special Health Needs (CSHN) Program

Patient Name _____
Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>

Other

**MCT Supplement**

MCT supplement recommended/prescribed  Unknown  Yes  No

MCT supplement formulation recommended/prescribed

Unknown  Metabolic formula  Oil  Powder

Total dose of MCT (grams) provided by the recommended/prescribed supplement per day \_\_\_\_\_

MCT supplement taken as recommended/prescribed  Unknown  Yes  No

Actual total dose of MCT (grams) taken daily \_\_\_\_\_

Reason MCT supplement is not taken as recommended/prescribed \_\_\_\_\_

Method of payment for MCT supplement

- Unknown
- None
- Commercial/private
- Medicaid
- Medicare
- Military
- Newborn screening funds
- Patient assistance program
- Self-pay
- State Children's Health Insurance Program (SCHIP)
- State Children with Special Health Needs (CSHN) Program
- Other

**Essential fatty acids**

Essential fatty acids recommended/prescribed  Unknown  Yes  No

Essential fatty acids route recommended/prescribed  Unknown  Feeding tube  Oral

Essential fatty acids dose recommended/prescribed \_\_\_\_\_

Essential fatty acids dose recommended/prescribed units  g  mg

Essential fatty acids frequency recommended/prescribed

- Unknown  Once/day  Twice/day  Three times/day  Four times/day
- Other

Essential fatty acids frequency recommended/prescribed-other, specify \_\_\_\_\_

Essential fatty acids taken as recommended/prescribed  Unknown  Yes  No

Actual essential fatty acids dose reported \_\_\_\_\_

Actual essential fatty acids dose reported units  g  mg

Actual essential fatty acids frequency reported

Patient Name \_\_\_\_\_

Date      

- Unknown       Once/day       Twice/day       Three times/day       Four times/day  
 Other

Actual essential fatty acids frequency reported-other, specify \_\_\_\_\_

Reason essential fatty acids are not taken as recommended/prescribed \_\_\_\_\_

**Method of payment for essential fatty acids**

- Unknown  
 None  
 Commercial/private  
 Medicaid  
 Medicare  
 Military  
 Newborn screening funds  
 Patient assistance program  
 Self-pay  
 State Children's Health Insurance Program (SCHIP)  
 State Children with Special Health Needs (CSHN) Program  
 Other

**Fat Restricted Diet**Fat restricted diet recommended/prescribed     Unknown     Yes     No

Percent of total kcal per 24 hours from fat recommended/prescribed in fat restricted diet \_\_\_\_\_

Fat grams per day recommended/prescribed in fat restricted diet \_\_\_\_\_

Patient/primary caregiver reports adherence to fat restricted diet     Unknown     Yes     No

Actual frequency of adherence to fat restricted diet

- Unknown     0 days/week     1 day/week     2 days/week     3 days/week     4 days/week  
 5 days/week     6 days/week

Reason for poor adherence to fat restricted diet \_\_\_\_\_

**Feeding Intervals**Specific information given to avoid fasting     Unknown     Yes     No

Longest interval recommended between feedings in well state during DAY (in hours)

- Unknown     1       2       3       4       5       6  
 7       8       9       10       11       12

Longest reported interval between feedings in well state during DAY (in hours)

- Unknown     1       2       3       4       5       6  
 7       8       9       10       11       12

Longest interval recommended between feedings in well state during NIGHT (in hours)

Patient Name _____
Date <input type="text" value="M"/> <input type="text" value="M"/> <input type="text" value="D"/> <input type="text" value="D"/> <input type="text" value="Y"/> <input type="text" value="Y"/>

- Unknown  1       2       3       4       5       6  
 7       8       9       10       11       12

Longest reported interval between feedings in well state during NIGHT (in hours)

- Unknown  1       2       3       4       5       6  
 7       8       9       10       11       12

**Comments**

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Nutrition comments



Patient Name \_\_\_\_\_

Date        

## CACT - Study Status

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First date of study status change      Condition follow-up status  Active  Inactive

Reason for inactive status

- Unknown  Deceased  Lost to follow-up
- Moved  Refused follow-up  Follow-up deemed unnecessary
- Subject withdrawal from study

Date of death      

Age of death (in years) \_\_\_\_\_

Moved-specify

- Moved to another IBEMC participating center
- Moved to another non-IBEMC participating center
- Moved-condition follow-up status unknown
- Specify other IBEMC participating center
- IL-Ann & Robert H. Lurie Children's Hospital of Chicago
- IL-University of Illinois
- IN- Riley Hospital for Children Indiana University Health
- KY-University of Louisville
- MI-University of Michigan
- MI-Wayne State University Children's Hospital of Michigan
- MN-University of Minnesota
- MO-University of Missouri
- NE-University of Nebraska
- NJ-Hackensack University
- NY-University of Rochester
- NY-Women's & Children's Hospital of Buffalo
- OH-Cincinnati Children's Hospital
- OH-Nationwide Children's Hospital
- OK-Saint Francis Hospital
- OK-University of Oklahoma
- PA-Children's Hospital of Pittsburgh
- SD-Sanford Children's Specialty Clinic
- WI-University of Wisconsin
- WI-Medical College of Wisconsin

Patient Name \_\_\_\_\_

Date

Specify other non-IBEMC participating center \_\_\_\_\_

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

- Declined consent at age of majority or emancipation
- Subject initiated withdrawal
- Investigator initiated withdrawal

Specify reason subject initiated withdrawal, if known

Specify reason investigator initiated withdrawal, if known

Second date of study status change

Condition follow-up status    Active    Inactive

Reason for inactive status

- Unknown
- Moved
- Subject withdrawal from study
- Deceased
- Refused follow-up
- Lost to follow-up
- Follow-up deemed unnecessary

Date of death

Age of death (in years) \_\_\_\_\_

Moved-specify

- Moved to another IBEMC participating center
- Moved to another non-IBEMC participating center
- Moved-condition follow-up status unknown

Specify other IBEMC participating center

- IL-Ann & Robert H. Lurie Children's Hospital of Chicago

Patient Name _____
Date <input type="text" value="M"/> <input type="text" value="M"/> <input type="text" value="D"/> <input type="text" value="D"/> <input type="text" value="Y"/> <input type="text" value="Y"/>

- IL-University of Illinois
- IN- Riley Hospital for Children Indiana University Health
- KY-University of Louisville
- MI-University of Michigan
- MI-Wayne State University Children's Hospital of Michigan
- MN-University of Minnesota
- MO-University of Missouri
- NE-University of Nebraska
- NJ-Hackensack University
- NY-University of Rochester
- NY-Women's & Children's Hospital of Buffalo
- OH-Cincinnati Children's Hospital
- OH-Nationwide Children's Hospital
- OK-Saint Francis Hospital
- OK-University of Oklahoma
- PA-Children's Hospital of Pittsburgh
- SD-Sanford Children's Specialty Clinic
- WI-University of Wisconsin
- WI-Medical College of Wisconsin

Specify other non-IBEMC participating center \_\_\_\_\_

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

- Declined consent at age of majority or emancipation
- Subject initiated withdrawal
- Investigator initiated withdrawal

Patient Name \_\_\_\_\_

Date

Specify reason subject initiated withdrawal, if known

Specify reason investigator initiated withdrawal, if known

Third date of study status change

Condition follow-up status    Active    Inactive

Reason for inactive status

- Unknown                                       Deceased                                       Lost to follow-up
- Moved     Refused follow-up                                       Follow-up deemed unnecessary
- Subject withdrawal from study

Date of death

Age of death (in years) \_\_\_\_\_

Moved-specify

- Moved to another IBEMC participating center
- Moved to another non-IBEMC participating center
- Moved-condition follow-up status unknown
- Specify other IBEMC participating center
  - IL-Ann & Robert H. Lurie Children's Hospital of Chicago
  - IL-University of Illinois
  - IN- Riley Hospital for Children Indiana University Health
  - KY-University of Louisville
  - MI-University of Michigan
  - MI-Wayne State University Children's Hospital of Michigan
  - MN-University of Minnesota
  - MO-University of Missouri
  - NE-University of Nebraska
  - NJ-Hackensack University
  - NY-University of Rochester
  - NY-Women's & Children's Hospital of Buffalo
  - OH-Cincinnati Children's Hospital
  - OH-Nationwide Children's Hospital

Patient Name \_\_\_\_\_

Date

- OK-Saint Francis Hospital
- OK-University of Oklahoma
- PA-Children's Hospital of Pittsburgh
- SD-Sanford Children's Specialty Clinic
- WI-University of Wisconsin
- WI-Medical College of Wisconsin

Specify other non-IBEMC participating center \_\_\_\_\_

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

- Declined consent at age of majority or emancipation
- Subject initiated withdrawal
- Investigator initiated withdrawal

Specify reason subject initiated withdrawal, if known

Specify reason investigator initiated withdrawal, if known

Fourth date of study status change

Condition follow-up status    Active    Inactive

Reason for inactive status

- Unknown
- Deceased
- Lost to follow-up
- Moved
- Refused follow-up
- Follow-up deemed unnecessary
- Subject withdrawal from study

Date of death

Age of death (in years) \_\_\_\_\_

Patient Name _____ Date <input type="text" value="M"/> <input type="text" value="M"/> <input type="text" value="D"/> <input type="text" value="D"/> <input type="text" value="Y"/> <input type="text" value="Y"/>
--

Moved-specify

- Moved to another IBEMC participating center
- Moved to another non-IBEMC participating center
- Moved-condition follow-up status unknown

Specify other IBEMC participating center

- IL-Ann & Robert H. Lurie Children's Hospital of Chicago
- IL-University of Illinois
- IN- Riley Hospital for Children Indiana University Health
- KY-University of Louisville
- MI-University of Michigan
- MI-Wayne State University Children's Hospital of Michigan
- MN-University of Minnesota
- MO-University of Missouri
- NE-University of Nebraska
- NJ-Hackensack University
- NY-University of Rochester
- NY-Women's & Children's Hospital of Buffalo
- OH-Cincinnati Children's Hospital
- OH-Nationwide Children's Hospital
- OK-Saint Francis Hospital
- OK-University of Oklahoma
- PA-Children's Hospital of Pittsburgh
- SD-Sanford Children's Specialty Clinic
- WI-University of Wisconsin
- WI-Medical College of Wisconsin

Specify other non-IBEMC participating center \_\_\_\_\_

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

Patient Name \_\_\_\_\_

Date

Declined consent at age of majority or emancipation

Subject initiated withdrawal

Investigator initiated withdrawal

Specify reason subject initiated withdrawal, if known

Specify reason investigator initiated withdrawal, if known

**Comments**

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Study status comments

Patient Name \_\_\_\_\_

Date      **CACT - Pregnancy**Patient has had one or more pregnancies  Unknown  Yes  No

Number of pregnancies \_\_\_\_\_

Number of term pregnancies \_\_\_\_\_

Number of preterm pregnancies \_\_\_\_\_

Number of pregnancies ending in abortion/miscarriage \_\_\_\_\_

Number of pregnancies resulting in live birth \_\_\_\_\_

Patient has biological children  Unknown  Yes  No

Number of biological children \_\_\_\_\_

**First Pregnancy**

Patient's age at time of first pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before first pregnancy (in months)

- Unknown  Unplanned  1  2  3  4  5  
 6  7  8  9  10  11  12  
 13-24  25-36  >36

History of infertility for first pregnancy

- Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for first pregnancy

- Unknown  Male factor  Female factor  
 Combined male-female factor

Cause(s) of male infertility for first pregnancy

- Unknown  Abnormal sperm production or function  
 Age  Cancer/cancer treatment related  
 Environmental  Problems with sperm delivery  
 Tobacco use  Other

Cause(s) of male infertility for first pregnancy-other, specify \_\_\_\_\_

Cause(s) of female infertility for first pregnancy

- Unknown  Age  
 Alcohol use  Cancer/cancer treatment related  
 Early menopause  Fallopian tube damage/blockage



Patient Name \_\_\_\_\_

Date |M|M| |D|D| |Y|Y|

- Hyperprolactinemia                      Pelvic adhesions  
Polycystic ovary syndrome (PCOS)   Premature ovarian insufficiency  
Over-exercise                                      Ovulation disorder  
Thyroid problems                                Tobacco use  
Uterine fibroid                                    Weight related  
Other

Cause(s) of female infertility for first pregnancy-other, specify \_\_\_\_\_

Cause(s) of infertility for first pregnancy, combined male-female factor- specify

\_\_\_\_\_

Types of fertility treatment(s) received prior to first pregnancy

- Unknown    None  
Assisted reproductive technology   Fertility drugs  
Surgery

Type of assisted reproductive technologies used during first pregnancy

- Assisted hatching                      ICSI                                      IVF  
Surgical sperm aspiration   Other

Type of assisted reproductive technologies used during first pregnancy-other, specify

\_\_\_\_\_

Partner tested for patient's disorder during first pregnancy

- Unknown                      Yes-biochemical                      Yes-molecular                      No

Partner also affected by disorder during first pregnancy   Unknown   Yes   NoPreimplantation genetic diagnosis for disorder done for first pregnancy   Unknown   Yes   NoPrenatal testing done for fetus for this disorder for first pregnancy   Unknown   Yes   NoType of prenatal testing performed for first pregnancy   Unknown   Biochemical   Molecular

Method of prenatal testing for first pregnancy

- Unknown    Amniocentesis                                      Chorionic villus sampling

Additional prenatal testing performed on fetus as a result of parent's disorder for first pregnancy

- Unknown    None  
Fetal echocardiogram                              Increased frequency prenatal ultrasounds  
Other

Additional prenatal testing performed on fetus as a result of parent's disorder for first pregnancy-other, specify \_\_\_\_\_

First pregnancy terminated   Unknown   Yes   No

Reason first pregnancy terminated

- Elective due to fetus affected with disorder

Patient Name \_\_\_\_\_

Date      

- Elective due to other fetal well-being unrelated to disorder
- Elective for other reason
- Elective due to maternal well-being
- Spontaneous

Gestational age (in weeks) at time of first pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for first pregnancy \_\_\_\_\_

Prenatal care received during first pregnancy  Unknown  Yes  No

Weeks gestation prenatal care started during first pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during first pregnancy

- Unknown  Metabolic diet  Medications
- Biochemical lab monitoring  Avoidance of fasting  Other

Treatment prescribed for patient's disorder during first pregnancy-other, specify

Patient in good metabolic condition prior to first pregnancy  Unknown  Yes  No

Patient in good metabolic condition during first trimester of first pregnancy  Unknown  Yes  No

Patient in good metabolic condition during second trimester of first pregnancy

- Unknown  Pregnancy not sustained to second trimester
- Yes  No

Patient in good metabolic condition during third trimester of first pregnancy

- Unknown  Pregnancy not sustained to third trimester
- Yes  No

Number of outpatient metabolic visits for patient during first pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during first pregnancy

- Unknown  0  1  2  3  4  5
- 6  7  8  9  10  >10

Number of hospitalizations for management of disorder during first pregnancy

- Unknown  0  1  2  3  4  5
- 6  7  8  9  10  >10

Complications during first pregnancy

- Unknown  Yes-related to disorder
- Yes- not known to be related to disorder  No

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

Complications related to disorder during first pregnancy- specify

- Unknown
- Acute fatty liver of pregnancy (AFLP)
- Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)
- Intrauterine growth restriction (AUGR)
- Mother affected with this condition
- Other

Complications not known to be related to disorder during first pregnancy-specify

- Unknown
- Advanced maternal age (35+ years of age)
- Ectopic pregnancy
- Gestational diabetes
- Group B strep
- Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)
- Intrauterine growth restriction (AUGR)
- Inadequate prenatal care
- Maternal prenatal substance exposure
- Preeclampsia
- Rh isoimmunization
- Toxemia
- Young maternal age (15 years of age + under)
- Preterm labor
- Other

Complications during first pregnancy-other, specify\_\_\_\_\_

Number of ED visits for complications during first pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5
- 6       7       8       9       10       >10

Number of hospitalizations for complications during first pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5
- 6       7       8       9       10       >10

Duration of longest inpatient hospitalization (in days) for any reason during first pregnancy\_\_\_\_\_

Highest value of primary metabolite of concern during first pregnancy (specify metabolite, value, and units of measure)

Patient Name \_\_\_\_\_

Date |M|M| |D|D| |Y|Y|

Lowest value of primary metabolite of concern during first pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during first pregnancy

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> Ammonia - Abn high                  |
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> C02 - Abn low                       | <input type="checkbox"/> C02 - WNL                           |
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

Other laboratory studies done on patient during first pregnancy: describe test(s) and result(s)

Additional interventions required during first pregnancy due to this metabolic condition

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                                    | <input type="checkbox"/> ED visits for hyperemesis/IV fluids   |
| <input type="checkbox"/> Hospitalizations for hyperemesis/IV fluids | <input type="checkbox"/> TPN                                   |
| <input type="checkbox"/> Tube feedings                              | <input type="checkbox"/> Additional medications                |
| <input type="checkbox"/> Home lab monitoring                        | <input type="checkbox"/> Increased frequency of lab monitoring |

Patient Name \_\_\_\_\_

Date      

- More intensive fetal monitoring       Bedrest  
 Other

Additional interventions required during first pregnancy due to this metabolic condition-other, specify  
 \_\_\_\_\_

Total maternal weight gain (in kg) during first pregnancy \_\_\_\_\_

First pregnancy complicated by maternal HELLP syndrome     Unknown     Yes     No

First pregnancy complicated by acute fatty liver of pregnancy (AFLP)     Unknown     Yes     No

Number of documented episodes of hypoglycemia during first pregnancy

- Unknown     0       1       2       3       4       5  
 6       7       8       9       10       >10

Lowest maternal blood glucose value documented during first pregnancy \_\_\_\_\_

Lowest maternal blood glucose value documented during first pregnancy units     mmol/L     mg/dL

Number of documented episodes of rhabdomyolysis during first pregnancy

- Unknown     0       1       2       3       4       5  
 6       7       8       9       10       >10

Highest maternal CK value documented during first pregnancy \_\_\_\_\_

Highest maternal CK value documented during first pregnancy units     U/L     umol/L     mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during first pregnancy

- Unknown     1       2       3       4       5       6  
 7       8       9       10       11       12

Longest permitted fasting interval (in hours) during the NIGHT recommended during first pregnancy

- Unknown     1       2       3       4       5       6  
 7       8       9       10       11       13

L-carnitine supplementation used on a regular basis during first pregnancy

- Unknown       Yes       No  
 L-carnitine not prescribed

Patient adhered to a fat-restricted diet during first pregnancy

- Unknown       Yes       No  
 Fat-restriction not prescribed

Patient took a metabolic formula regularly during first pregnancy

- Unknown       Yes  
 No       Metabolic formula not prescribed

Evidence of cardiac hypertrophy during first pregnancy

- Unknown       Yes  
 No       Echocardiogram not performed during pregnancy

Patient Name \_\_\_\_\_

Date

Evidence of any cardiac dysfunction during first pregnancy

- Unknown  Yes
- No  Cardiac testing not performed during pregnancy

Additional interventions planned for labor/delivery related to patient's disorder for first pregnancy

- Unknown  None
- Additional maternal lab monitoring  Altered anesthesia plan
- Change in delivery site  IV fluids
- Letter to OB/MFM specialist  Planned C-section
- Referral for high risk OB management  Other

Additional interventions planned for labor/delivery related to patient's disorder for first pregnancy-other, specify

Actual interventions for labor/delivery related to patient's disorder for first pregnancy

- Unknown  No
- Letter to OB/MFM specialist  IV fluids
- Planned C-section  Additional maternal lab monitoring
- Change in delivery site  Referral for high risk OB management
- Altered anesthesia plan  Other

Actual interventions for labor/delivery related to patient's disorder for first pregnancy-other, specify

Acute health concerns experienced by the patient during delivery for first delivery

Abnormal patient lab results during first delivery

- Unknown  None  Normal labs during delivery
- Elevated CK  Elevated liver function tests  Hyperammonemia
- Hypoglycemia  Metabolic decompensation  Other

Patient Name \_\_\_\_\_

Date

Abnormal patient lab results during first delivery-other, specify

Additional maternal interventions during or after first delivery related to this disorder

- Unknown                       Ammonul                       Blood/blood product transfusion
- Dialysis                       ICU monitoring                       Infusions
- Medications                       Resuscitation                       TPN
- Other

Additional maternal interventions during or after first delivery related to this disorder-other, specify

Patient death during or shortly after first delivery     Unknown     Yes     No

Disorder contributed to death of mother during first delivery     Unknown     Yes     No

Weeks gestation at time of first delivery - round to nearest week\_\_\_\_\_

Method of first delivery

- Unknown                                       Caesarean section (scheduled or non-urgent)
- Casesarean section (emergent)                       Vaginal

Maternal inpatient days post- first delivery\_\_\_\_\_

Live delivery of first newborn     Unknown     Yes     No

Acute health concerns for first newborn related to maternal disorder (example: fetal distress secondary to maternal acute metabolic decompensation)

Additional lab tests and results done on first newborn specifically due to maternal disorder history

Health concerns for first newborn not known to be related to disorder

Additional interventions for first newborn during or shortly after delivery (other than labs)

Patient Name \_\_\_\_\_

Date

- Unknown
- Blood/blood product transfusion
- Glucose infusion
- Medications
- NICU/special care nursery monitoring
- Resuscitation
- TPN
- Other

Additional interventions for first newborn during or shortly after delivery-other, specify

Birth measurements for first newborn  Unknown  Head circumference  Length  Weight

Birth head circumference \_\_\_\_\_

Birth head circumference units  cm  in

Birth length \_\_\_\_\_

Birth length units  cm  in

Birth weight \_\_\_\_\_

Birth weight units  lbs  kg  gm  oz

APGAR score at 1 minute for first newborn

- Unknown
- 0
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10

Health concern(s) with APGAR score at one minute < 8 for first newborn

APGAR score at 5 minutes for first newborn

- Unknown
- 0
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10

Health concern(s) with APGAR score at five minutes < 8 for first newborn

Length of first newborn's stay in the hospital after birth

- Unknown
- <24 hours
- 24-28 hours
- 3-5 days
- 6-14 days
- >14 days
- N/A - baby was not born in a hospital
- baby is still in the hospital

Routine newborn screening done for first newborn



Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

- Unknown
- Yes - abnormal
- No
- Yes - normal for all screened disorders
- Yes - results pending

Describe abnormal newborn screen result for first newborn

Reason routine newborn screening was not done for first newborn

- Unknown
- Died prior to collection of NBS
- Refused
- Transferred to another facility prior to NBS collection
- Transfused prior to collection of NBS
- Other

Reason routine newborn screening was not done for first newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the first baby after birth to rule out the mother's disorder

- Unknown
- Yes - biochemical normal
- Yes - biochemical abnormal
- Yes - molecular normal
- Yes - molecular abnormal affected
- Yes - molecular abnormal unaffected carrier
- No

Additional studies and results (examples: brain MRI, echocardiogram) done on first newborn to assess for effects of maternal disorder

First newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

- Unknown
- Yes
- No

Abnormal newborn exam findings for first newborn

- Unknown
- None
- Congenital heart disease
- Dysmorphism
- Lethargy
- Microcephaly
- Other congenital anomalies
- Poor feeding
- Respiratory distress
- Seizure(s)
- Small for gestational age
- Other

Abnormal newborn exam findings for first newborn-other, specify \_\_\_\_\_

Patient Name \_\_\_\_\_

Date      Newborn death at or shortly after delivery for first newborn  Unknown  Yes  NoNewborn's death related to maternal disorder for first newborn  Unknown  Yes  NoFirst newborn currently alive  Unknown  Yes  No**Second Pregnancy**

---

Patient's age at time of second pregnancy (in years)\_\_\_\_\_

Length of time patient tried to become pregnant before second pregnancy (in months)

- Unknown  Unplanned  1  2  3  4  5  
 6  7  8  9  10  11  12  
 13-24  25-36  >36

History of infertility for second pregnancy

- Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for second pregnancy

- Unknown  Male factor  Female factor  
 Combined male-female factor

Cause(s) of male infertility for second pregnancy

- Unknown  Abnormal sperm production or function  
 Age  Cancer/cancer treatment related  
 Environmental  Problems with sperm delivery  
 Tobacco use  Other

Cause(s) of male infertility for second pregnancy-other, specify\_\_\_\_\_

Cause(s) of female infertility for second pregnancy

- Unknown  Age  
 Alcohol use  Cancer/cancer treatment related  
 Early menopause  Fallopian tube damage/blockage  
 Hyperprolactinemia  Pelvic adhesions  
 Polycystic ovary syndrome (PCOS)  Premature ovarian insufficiency  
 Over-exercise  Ovulation disorder  
 Thyroid problems  Tobacco use  
 Uterine fibroid  Weight related  
 Other

Cause(s) of female infertility for second pregnancy-other, specify\_\_\_\_\_

Cause(s) of infertility for second pregnancy, combined male-female factor- specify

Patient Name \_\_\_\_\_

Date      

Types of fertility treatment(s) received prior to second pregnancy

- Unknown  None  
 Assisted reproductive technology  Fertility drugs  
 Surgery

Type of assisted reproductive technologies used during second pregnancy

- Assisted hatching  ICSI  IVF  
 Surgical sperm aspiration  Other

Type of assisted reproductive technologies used during second pregnancy-other, specify \_\_\_\_\_

Partner tested for patient's disorder during second pregnancy

- Unknown  Yes-biochemical  Yes-molecular  No

Partner also affected by disorder during second pregnancy  Unknown  Yes  No

Preimplantation genetic diagnosis for disorder done for second pregnancy  Unknown  Yes  No

Prenatal testing done for fetus for this disorder for second pregnancy  Unknown  Yes  No

Type of prenatal testing performed for second pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for second pregnancy

- Unknown  Amniocentesis  Chorionic villus sampling

Additional prenatal testing performed on fetus as a result of parent's disorder for second pregnancy

- Unknown  None  
 Fetal echocardiogram  Increased frequency prenatal ultrasounds  
 Other

Additional prenatal testing performed on fetus as a result of parent's disorder for second pregnancy-other, specify \_\_\_\_\_

Second pregnancy terminated  Unknown  Yes  No

Reason second pregnancy terminated

- Elective due to fetus affected with disorder  
 Elective due to other fetal well-being unrelated to disorder  
 Elective for other reason  
 Elective due to maternal well-being  
 Spontaneous

Gestational age (in weeks) at time of second pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for second pregnancy \_\_\_\_\_

Patient Name \_\_\_\_\_

Date

Prenatal care received during second pregnancy Unknown Yes No

Weeks gestation prenatal care started during second pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during second pregnancy

Unknown Metabolic diet Medications

Biochemical lab monitoring Avoidance of fasting Other

Treatment prescribed for patient's disorder during second pregnancy-other, specify

Patient in good metabolic condition prior to second pregnancy Unknown Yes No

Patient in good metabolic condition during first trimester of second pregnancy Unknown Yes No

Patient in good metabolic condition during second trimester of second pregnancy

Unknown Pregnancy not sustained to second trimester

Yes No

Patient in good metabolic condition during third trimester of second pregnancy

Unknown Pregnancy not sustained to third trimester

Yes No

Number of outpatient metabolic visits for patient during second pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during second pregnancy

Unknown 0 1 2 3 4 5

6 7 8 9 10  >10

Number of hospitalizations for management of disorder during second pregnancy

Unknown 0 1 2 3 4 5

6 7 8 9 10  >10

Complications during second pregnancy

Unknown Yes-related to disorder

Yes- not known to be related to disorder No

Complications related to disorder during second pregnancy- specify

Unknown

Acute fatty liver of pregnancy (AFLP)

Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)

Intrauterine growth restriction (AUGR)

Mother affected with this condition

Other

Complications not known to be related to disorder during second pregnancy-specify

Patient Name _____
Date <input type="text" value="M"/> <input type="text" value="M"/> <input type="text" value="D"/> <input type="text" value="D"/> <input type="text" value="Y"/> <input type="text" value="Y"/>

- Unknown
- Advanced maternal age (35+ years of age)
- Ectopic pregnancy
- Gestational diabetes
- Group B strep
- Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)
- Intrauterine growth restriction (AUGR)
- Inadequate prenatal care
- Maternal prenatal substance exposure
- Preeclampsia
- Rh isoimmunization
- Toxemia
- Young maternal age (15 years of age + under)
- Preterm labor
- Other

Complications during second pregnancy-other, specify \_\_\_\_\_

Number of ED visits for complications during second pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5
- 6       7       8       9       10       >10

Number of hospitalizations for complications during second pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5
- 6       7       8       9       10       >10

Duration of longest inpatient hospitalization (in days) for any reason during second pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during second pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during second pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during second pregnancy

- Unknown                                       Ammonia - Abn high

Patient Name \_\_\_\_\_

Date      

- |  |  |
|--|--|
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> CO2 - Abn low                       | <input type="checkbox"/> CO2 - WNL                           |
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

Other laboratory studies done on patient during second pregnancy: describe test(s) and result(s)

Additional interventions required during second pregnancy due to this metabolic condition

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                                    | <input type="checkbox"/> ED visits for hyperemesis/IV fluids   |
| <input type="checkbox"/> Hospitalizations for hyperemesis/IV fluids | <input type="checkbox"/> TPN                                   |
| <input type="checkbox"/> Tube feedings                              | <input type="checkbox"/> Additional medications                |
| <input type="checkbox"/> Home lab monitoring                        | <input type="checkbox"/> Increased frequency of lab monitoring |
| <input type="checkbox"/> More intensive fetal monitoring            | <input type="checkbox"/> Bedrest                               |
| <input type="checkbox"/> Other                                      |  |

Additional interventions required during second pregnancy due to this metabolic condition-other, specify

\_\_\_\_\_

Total maternal weight gain (in kg) during second pregnancy \_\_\_\_\_

Second pregnancy complicated by maternal HELLP syndrome     Unknown     Yes     NoSecond pregnancy complicated by acute fatty liver of pregnancy (AFLP)     Unknown     Yes     No

Patient Name \_\_\_\_\_

Date      

Number of documented episodes of hypoglycemia during second pregnancy

- Unknown  0  1  2  3  4  5  
 6  7  8  9  10  >10

Lowest maternal blood glucose value documented during second pregnancy \_\_\_\_\_

Lowest maternal blood glucose value documented during second pregnancy units  mmol/L  mg/dL

Number of documented episodes of rhabdomyolysis during second pregnancy

- Unknown  0  1  2  3  4  5  
 6  7  8  9  10  >10

Highest maternal CK value documented during second pregnancy \_\_\_\_\_

Highest maternal CK value documented during second pregnancy units  U/L  umol/L  mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during second pregnancy

- Unknown  1  2  3  4  5  6  
 7  8  9  10  11  12

Longest permitted fasting interval (in hours) during the NIGHT recommended during second pregnancy

- Unknown  1  2  3  4  5  6  
 7  8  9  10  11  13

L-carnitine supplementation used on a regular basis during second pregnancy

- Unknown  Yes  No  
 L-carnitine not prescribed

Patient adhered to a fat-restricted diet during second pregnancy

- Unknown  Yes  No  
 Fat-restriction not prescribed

Patient took a metabolic formula regularly during second pregnancy

- Unknown  Yes  
 No  Metabolic formula not prescribed

Evidence of cardiac hypertrophy during second pregnancy

- Unknown  Yes  
 No  Echocardiogram not performed during pregnancy

Evidence of any cardiac dysfunction during second pregnancy

- Unknown  Yes  
 No  Cardiac testing not performed during pregnancy

Additional interventions planned for labor/delivery related to patient's disorder for second pregnancy

- Unknown  None  
 Additional maternal lab monitoring  Altered anesthesia plan  
 Change in delivery site  IV fluids

Patient Name \_\_\_\_\_

Date

- Letter to OB/MFM specialist
- Planned C-section
- Referral for high risk OB management
- Other

Additional interventions planned for labor/delivery related to patient's disorder for second pregnancy-other, specify

Actual interventions for labor/delivery related to patient's disorder for second pregnancy

- Unknown
- No
- Letter to OB/MFM specialist
- IV fluids
- Planned C-section
- Additional maternal lab monitoring
- Change in delivery site
- Referral for high risk OB management
- Altered anesthesia plan
- Other

Actual interventions for labor/delivery related to patient's disorder for second pregnancy-other, specify

Acute health concerns experienced by the patient during delivery for second delivery

Abnormal patient lab results during second delivery

- Unknown
- None
- Normal labs during delivery
- Elevated CK
- Elevated liver function tests
- Hyperammonemia
- Hypoglycemia
- Metabolic decompensation
- Other

Abnormal patient lab results during second delivery-other, specify

Additional maternal interventions during or after second delivery related to this disorder

- Unknown
- Ammonul
- Blood/blood product transfusion
- Dialysis
- ICU monitoring
- Infusions
- Medications
- Resuscitation
- TPN
- Other



Patient Name \_\_\_\_\_

Date

Additional maternal interventions during or after second delivery related to this disorder-other, specify

Patient death during or shortly after second delivery Unknown Yes No

Disorder contributed to death of mother during second delivery Unknown Yes No

Weeks gestation at time of second delivery - round to nearest week \_\_\_\_\_

Method of second delivery

- Unknown Caesarean section (scheduled or non-urgent)
- Casesarean section (emergent) Vaginal

Maternal inpatient days post- second delivery \_\_\_\_\_

Live delivery of second newborn Unknown Yes No

Acute health concerns for second newborn related to maternal disorder (example: fetal distress secondary to maternal acute metabolic decompensation)

Additional lab tests and results done on second newborn specifically due to maternal disorder history

Health concerns for second newborn not known to be related to disorder

Additional interventions for second newborn during or shortly after delivery (other than labs)

- Unknown Blood/blood product transfusion
- Glucose infusion Medications
- NICU/special care nursery monitoring Resuscitation
- TPN Other

Additional interventions for second newborn during or shortly after delivery-other, specify

Birth measurements for second newborn Unknown Head circumference Length Weight

Patient Name \_\_\_\_\_

Date

Birth head circumference \_\_\_\_\_

Birth head circumference units  cm  in

Birth length \_\_\_\_\_

Birth length units  cm  in

Birth weight \_\_\_\_\_

Birth weight units  lbs  kg  gm  oz

APGAR score at 1 minute for second newborn

Unknown  0  1  2  3  4  5

6  7  8  9  10

Health concern(s) with APGAR score at one minute < 8 for second newborn

APGAR score at 5 minutes for second newborn

Unknown  0  1  2  3  4  5

6  7  8  9  10

Health concern(s) with APGAR score at five minutes < 8 for second newborn

Length of second newborn's stay in the hospital after birth

Unknown  <24 hours

24-28 hours  3-5 days

6-14 days  >14 days

N/A - baby was not born in a hospital  baby is still in the hospital

Routine newborn screening done for second newborn

Unknown  Yes - normal for all screened disorders

Yes -abnormal  Yes - results pending

No

Describe abnormal newborn screen result for second newborn

Reason routine newborn screening was not done for second newborn

Unknown

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

- Died prior to collection of NBS
- Refused
- Transferred to another facility prior to NBS collection
- Transfused prior to collection of NBS
- Other

Reason routine newborn screening was not done for second newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the second baby after birth to rule out the mother's disorder

- Unknown
- Yes - biochemical normal
- Yes - biochemical abnormal
- Yes - molecular normal
- Yes - molecular abnormal affected
- Yes - molecular abnormal unaffected carrier
- No

Additional studies and results (examples: brain MRI, echocardiogram) done on second newborn to assess for effects of maternal disorder

Second newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

- Unknown
- Yes
- No

Abnormal newborn exam findings for second newborn

- Unknown
- None
- Congenital heart disease
- Dysmorphism
- Lethargy
- Microcephaly
- Other congenital anomalies
- Poor feeding
- Respiratory distress
- Seizure(s)
- Small for gestational age
- Other

Abnormal newborn exam findings for second newborn-other, specify \_\_\_\_\_

Newborn death at or shortly after delivery for second newborn  Unknown  Yes  No

Newborn's death related to maternal disorder for second newborn  Unknown  Yes  No

Second newborn currently alive  Unknown  Yes  No

### Third Pregnancy

---

Patient's age at time of third pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before third pregnancy (in months)

- Unknown
- Unplanned
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10
- 11
- 12

Patient Name \_\_\_\_\_

Date      

13-24     25-36     >36

## History of infertility for third pregnancy

- Unknown
- Yes- not known to be related to inborn error of metabolism
- Yes-related to inborn error of metabolism
- No

## Cause(s) of infertility for third pregnancy

- Unknown                       Male factor                       Female factor
- Combined male-female factor

## Cause(s) of male infertility for third pregnancy

- Unknown                       Abnormal sperm production or function
- Age                               Cancer/cancer treatment related
- Environmental               Problems with sperm delivery
- Tobacco use                   Other

Cause(s) of male infertility for third pregnancy-other, specify \_\_\_\_\_

## Cause(s) of female infertility for third pregnancy

- Unknown                       Age
- Alcohol use                       Cancer/cancer treatment related
- Early menopause               Fallopian tube damage/blockage
- Hyperprolactinemia               Pelvic adhesions
- Polycystic ovary syndrome (PCOS)    Premature ovarian insufficiency
- Over-exercise                       Ovulation disorder
- Thyroid problems               Tobacco use
- Uterine fibroid                   Weight related
- Other

Cause(s) of female infertility for third pregnancy-other, specify \_\_\_\_\_

## Cause(s) of infertility for third pregnancy, combined male-female factor- specify

\_\_\_\_\_

## Types of fertility treatment(s) received prior to third pregnancy

- Unknown                       None
- Assisted reproductive technology    Fertility drugs
- Surgery

## Type of assisted reproductive technologies used during third pregnancy

- Assisted hatching               ICSI                               IVF
- Surgical sperm aspiration    Other

Patient Name \_\_\_\_\_

Date      Type of assisted reproductive technologies used during third pregnancy-other, specify  
\_\_\_\_\_

Partner tested for patient's disorder during third pregnancy

 Unknown       Yes-biochemical       Yes-molecular       NoPartner also affected by disorder during third pregnancy     Unknown     Yes     NoPreimplantation genetic diagnosis for disorder done for third pregnancy     Unknown     Yes     NoPrenatal testing done for fetus for this disorder for third pregnancy     Unknown     Yes     NoType of prenatal testing performed for third pregnancy     Unknown     Biochemical     Molecular

Method of prenatal testing for third pregnancy

 Unknown       Amniocentesis       Chorionic villus sampling

Additional prenatal testing performed on fetus as a result of parent's disorder for third pregnancy

 Unknown       None Fetal echocardiogram       Increased frequency prenatal ultrasounds OtherAdditional prenatal testing performed on fetus as a result of parent's disorder for third pregnancy-other,  
specify \_\_\_\_\_Third pregnancy terminated     Unknown     Yes     No

Reason third pregnancy terminated

- Elective due to fetus affected with disorder
- Elective due to other fetal well-being unrelated to disorder
- Elective for other reason
- Elective due to maternal well-being
- Spontaneous

Gestational age (in weeks) at time of third pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for third pregnancy \_\_\_\_\_

Prenatal care received during third pregnancy     Unknown     Yes     No

Weeks gestation prenatal care started during third pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during third pregnancy

 Unknown       Metabolic diet       Medications  
 Biochemical lab monitoring     Avoidance of fasting       Other

Patient Name \_\_\_\_\_

Date |M|M| |D|D| |Y|Y|

Treatment prescribed for patient's disorder during third pregnancy-other, specify

--

Patient in good metabolic condition prior to third pregnancy Unknown Yes NoPatient in good metabolic condition during first trimester of third pregnancy Unknown Yes No

Patient in good metabolic condition during second trimester of third pregnancy

UnknownPregnancy not sustained to second trimesterYesNo

Patient in good metabolic condition during third trimester of third pregnancy

UnknownPregnancy not sustained to third trimesterYesNo

Number of outpatient metabolic visits for patient during third pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during third pregnancy

Unknown 0 1 2 3 4 56 7 8 9 10  >10

Number of hospitalizations for management of disorder during third pregnancy

Unknown 0 1 2 3 4 56 7 8 9 10  >10

Complications during third pregnancy

UnknownYes-related to disorderYes- not known to be related to disorder No

Complications related to disorder during third pregnancy- specify

UnknownAcute fatty liver of pregnancy (AFLP)Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)Intrauterine growth restriction (AUGR)Mother affected with this conditionOther

Complications not known to be related to disorder during third pregnancy-specify

UnknownAdvanced maternal age (35+ years of age)Ectopic pregnancyGestational diabetesGroup B strep

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/>   <input type="text"/> <input type="text"/> <input type="text"/>   <input type="text"/> <input type="text"/>
--

- Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)
- Intrauterine growth restriction (AUGR)
- Inadequate prenatal care
- Maternal prenatal substance exposure
- Preeclampsia
- Rh isoimmunization
- Toxemia
- Young maternal age (15 years of age + under)
- Preterm labor
- Other

Complications during third pregnancy-other, specify \_\_\_\_\_

Number of ED visits for complications during third pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5
- 6       7       8       9       10       >10

Number of hospitalizations for complications during third pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5
- 6       7       8       9       10       >10

Duration of longest inpatient hospitalization (in days) for any reason during third pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during third pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during third pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during third pregnancy

- |  |   |
|--|---|
| <input type="checkbox"/> Unknown             | <input type="checkbox"/> Ammonia - Abn high       |
| <input type="checkbox"/> Ammonia - WNL       | <input type="checkbox"/> Blood glucose - Abn low  |
| <input type="checkbox"/> Blood glucose - WNL | <input type="checkbox"/> Blood Glucose - Abn high |
| <input type="checkbox"/> BNP - Abn high      | <input type="checkbox"/> BNP - WNL                |
| <input type="checkbox"/> CBC - Abn           | <input type="checkbox"/> CBC - WNL                |
| <input type="checkbox"/> CO2 - Abn low       | <input type="checkbox"/> CO2 - WNL                |

Patient Name \_\_\_\_\_

Date      

- |  |  |
|--|--|
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

Other laboratory studies done on patient during third pregnancy: describe test(s) and result(s)

Additional interventions required during third pregnancy due to this metabolic condition

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                                    | <input type="checkbox"/> ED visits for hyperemesis/IV fluids   |
| <input type="checkbox"/> Hospitalizations for hyperemesis/IV fluids | <input type="checkbox"/> TPN                                   |
| <input type="checkbox"/> Tube feedings                              | <input type="checkbox"/> Additional medications                |
| <input type="checkbox"/> Home lab monitoring                        | <input type="checkbox"/> Increased frequency of lab monitoring |
| <input type="checkbox"/> More intensive fetal monitoring            | <input type="checkbox"/> Bedrest                               |
| <input type="checkbox"/> Other                                      |  |

Additional interventions required during third pregnancy due to this metabolic condition—other, specify

\_\_\_\_\_

Total maternal weight gain (in kg) during third pregnancy \_\_\_\_\_

Third pregnancy complicated by maternal HELLP syndrome     Unknown     Yes     NoThird pregnancy complicated by acute fatty liver of pregnancy (AFLP)     Unknown     Yes     No

Number of documented episodes of hypoglycemia during third pregnancy

- Unknown     0             1             2             3             4             5  
 6             7             8             9             10           >10

Lowest maternal blood glucose value documented during third pregnancy \_\_\_\_\_

Lowest maternal blood glucose value documented during third pregnancy units     mmol/L     mg/dL



Patient Name \_\_\_\_\_

Date      

Number of documented episodes of rhabdomyolysis during third pregnancy

- Unknown  0  1  2  3  4  5  
 6  7  8  9  10  >10

Highest maternal CK value documented during third pregnancy \_\_\_\_\_

Highest maternal CK value documented during third pregnancy units  U/L  umol/L  mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during third pregnancy

- Unknown  1  2  3  4  5  6  
 7  8  9  10  11  12

Longest permitted fasting interval (in hours) during the NIGHT recommended during third pregnancy

- Unknown  1  2  3  4  5  6  
 7  8  9  10  11  13

L-carnitine supplementation used on a regular basis during third pregnancy

- Unknown  Yes  No  
 L-carnitine not prescribed

Patient adhered to a fat-restricted diet during third pregnancy

- Unknown  Yes  No  
 Fat-restriction not prescribed

Patient took a metabolic formula regularly during third pregnancy

- Unknown  Yes  
 No  Metabolic formula not prescribed

Evidence of cardiac hypertrophy during third pregnancy

- Unknown  Yes  
 No  Echocardiogram not performed during pregnancy

Evidence of any cardiac dysfunction during third pregnancy

- Unknown  Yes  
 No  Cardiac testing not performed during pregnancy

Additional interventions planned for labor/delivery related to patient's disorder for third pregnancy

- Unknown  None  
 Additional maternal lab monitoring  Altered anesthesia plan  
 Change in delivery site  IV fluids  
 Letter to OB/MFM specialist  Planned C-section  
 Referral for high risk OB management  Other

Patient Name \_\_\_\_\_

Date

Additional interventions planned for labor/delivery related to patient's disorder for third pregnancy-other, specify

Actual interventions for labor/delivery related to patient's disorder for third pregnancy

- Unknown No
- Letter to OB/MFM specialist IV fluids
- Planned C-section Additional maternal lab monitoring
- Change in delivery site Referral for high risk OB management
- Altered anesthesia plan Other

Actual interventions for labor/delivery related to patient's disorder for third pregnancy-other, specify

Acute health concerns experienced by the patient during delivery for third delivery

Abnormal patient lab results during third delivery

- Unknown None Normal labs during delivery
- Elevated CK Elevated liver function tests Hyperammonemia
- Hypoglycemia Metabolic decompensation Other

Abnormal patient lab results during third delivery-other, specify

Additional maternal interventions during or after third delivery related to this disorder

- Unknown Ammonul Blood/blood product transfusion
- Dialysis ICU monitoring Infusions
- Medications Resuscitation TPN
- Other

Patient Name \_\_\_\_\_

Date

Additional maternal interventions during or after third delivery related to this disorder-other, specify

Patient death during or shortly after third delivery Unknown Yes No

Disorder contributed to death of mother during third delivery Unknown Yes No

Weeks gestation at time of third delivery - round to nearest week \_\_\_\_\_

Method of third delivery

- Unknown Caesarean section (scheduled or non-urgent)
- Casesarean section (emergent) Vaginal

Maternal inpatient days post- third delivery \_\_\_\_\_

Live delivery of third newborn Unknown Yes No

Acute health concerns for third newborn related to maternal disorder (example: fetal distress secondary to maternal acute metabolic decompensation)

Additional lab tests and results done on third newborn specifically due to maternal disorder history

Birth measurements for third newborn Unknown Head circumference Length Weight

Birth head circumference \_\_\_\_\_

Birth head circumference units cm in

Birth length \_\_\_\_\_

Birth length units cm in

Birth weight \_\_\_\_\_

Birth weight units lbs kg gm oz

APGAR score at 1 minute for third newborn

- Unknown
- 0
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10

Health concern(s) with APGAR score at one minute < 8 for third newborn

Patient Name \_\_\_\_\_

Date      

APGAR score at 5 minutes for third newborn

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10

Health concern(s) with APGAR score at five minutes &lt; 8 for third newborn

Length of third newborn's stay in the hospital after birth

- Unknown                                       <24 hours  
 24-28 hours                                       3-5 days  
 6-14 days     >14 days  
 N/A - baby was not born in a hospital    baby is still in the hospital

Routine newborn screening done for third newborn

- Unknown     Yes - normal for all screened disorders  
 Yes - abnormal                                       Yes - results pending  
 No

Describe abnormal newborn screen result for third newborn

Reason routine newborn screening was not done for third newborn

- Unknown  
 Died prior to collection of NBS  
 Refused  
 Transferred to another facility prior to NBS collection  
 Transfused prior to collection of NBS  
 Other

Reason routine newborn screening was not done for third newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the third baby after birth to rule out the mother's disorder

- Unknown     Yes - biochemical normal  
 Yes - biochemical abnormal                       Yes - molecular normal  
 Yes - molecular abnormal affected               Yes - molecular abnormal unaffected carrier  
 No

Patient Name \_\_\_\_\_

Date      

Additional studies and results (examples: brain MRI, echocardiogram) done on third newborn to assess for effects of maternal disorder

Third newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

Unknown  Yes  No

Abnormal newborn exam findings for third newborn

Unknown  None  Congenital heart disease  
 Dysmorphism  Lethargy  Microcephaly  
 Other congenital anomalies  Poor feeding  Respiratory distress  
 Seizure(s)  Small for gestational age  Other

Abnormal newborn exam findings for third newborn-other, specify \_\_\_\_\_

Newborn death at or shortly after delivery for third newborn  Unknown  Yes  No

Newborn's death related to maternal disorder for third newborn  Unknown  Yes  No

Third newborn currently alive  Unknown  Yes  No

## Fourth Pregnancy

Patient's age at time of fourth pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before fourth pregnancy (in months)

Unknown  Unplanned  1  2  3  4  5  
 6  7  8  9  10  11  12  
 13-24  25-36  >36

History of infertility for fourth pregnancy

Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for fourth pregnancy

Unknown  Male factor  Female factor  
 Combined male-female factor

Cause(s) of male infertility for fourth pregnancy

Unknown  Abnormal sperm production or function  
 Age  Cancer/cancer treatment related  
 Environmental  Problems with sperm delivery  
 Tobacco use  Other

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

Cause(s) of male infertility for fourth pregnancy-other, specify \_\_\_\_\_

Cause(s) of female infertility for fourth pregnancy

- Unknown
- Age
- Alcohol use
- Cancer/cancer treatment related
- Early menopause
- Fallopian tube damage/blockage
- Hyperprolactinemia
- Pelvic adhesions
- Polycystic ovary syndrome (PCOS)
- Premature ovarian insufficiency
- Over-exercise
- Ovulation disorder
- Thyroid problems
- Tobacco use
- Uterine fibroid
- Weight related
- Other

Cause(s) of female infertility for fourth pregnancy-other, specify \_\_\_\_\_

Cause(s) of infertility for fourth pregnancy, combined male-female factor- specify

\_\_\_\_\_

Types of fertility treatment(s) received prior to fourth pregnancy

- Unknown
- None
- Assisted reproductive technology
- Fertility drugs
- Surgery

Type of assisted reproductive technologies used during fourth pregnancy

- Assisted hatching
- ICSI
- IVF
- Surgical sperm aspiration
- Other

Type of assisted reproductive technologies used during fourth pregnancy-other, specify

\_\_\_\_\_

Partner tested for patient's disorder during fourth pregnancy

- Unknown
- Yes-biochemical
- Yes-molecular
- No

Partner also affected by disorder during fourth pregnancy  Unknown  Yes  No

Preimplantation genetic diagnosis for disorder done for fourth pregnancy  Unknown  Yes  No

Prenatal testing done for fetus for this disorder for fourth pregnancy  Unknown  Yes  No

Type of prenatal testing performed for fourth pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for fourth pregnancy

- Unknown
- Amniocentesis
- Chorionic villus sampling

Additional prenatal testing performed on fetus as a result of parent's disorder for fourth pregnancy

- Unknown
- None
- Fetal echocardiogram
- Increased frequency prenatal ultrasounds
- Other

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

Additional prenatal testing performed on fetus as a result of parent's disorder for fourth pregnancy-other, specify \_\_\_\_\_

Fourth pregnancy terminated     Unknown     Yes     No

Reason fourth pregnancy terminated

- Elective due to fetus affected with disorder
- Elective due to other fetal well-being unrelated to disorder
- Elective for other reason
- Elective due to maternal well-being
- Spontaneous

Gestational age (in weeks) at time of fourth pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for fourth pregnancy \_\_\_\_\_

Prenatal care received during fourth pregnancy     Unknown     Yes     No

Weeks gestation prenatal care started during fourth pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during fourth pregnancy

- Unknown                       Metabolic diet                       Medications
- Biochemical lab monitoring     Avoidance of fasting                       Other

Treatment prescribed for patient's disorder during fourth pregnancy-other, specify

Patient in good metabolic condition prior to fourth pregnancy     Unknown     Yes     No

Patient in good metabolic condition during first trimester of fourth pregnancy     Unknown     Yes     No

Patient in good metabolic condition during second trimester of fourth pregnancy

- Unknown                                       Pregnancy not sustained to second trimester
- Yes     No

Patient in good metabolic condition during third trimester of fourth pregnancy

- Unknown                                       Pregnancy not sustained to third trimester
- Yes     No

Number of outpatient metabolic visits for patient during fourth pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during fourth pregnancy

- Unknown     0             1             2             3             4             5
- 6             7             8             9             10             >10

Number of hospitalizations for management of disorder during fourth pregnancy

Patient Name \_\_\_\_\_

Date |M|M| |D|D| |Y|Y|

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10       >10

## Complications during fourth pregnancy

- Unknown                                       Yes-related to disorder  
 Yes- not known to be related to disorder    No

## Complications related to disorder during fourth pregnancy- specify

- Unknown  
 Acute fatty liver of pregnancy (AFLP)  
 Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)  
 Intrauterine growth restriction (AUGR)  
 Mother affected with this condition  
 Other

## Complications not known to be related to disorder during fourth pregnancy-specify

- Unknown  
 Advanced maternal age (35+ years of age)  
 Ectopic pregnancy  
 Gestational diabetes  
 Group B strep  
 Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)  
 Intrauterine growth restriction (AUGR)  
 Inadequate prenatal care  
 Maternal prenatal substance exposure  
 Preeclampsia  
 Rh isoimmunization  
 Toxemia  
 Young maternal age (15 years of age + under)  
 Preterm labor  
 Other

Complications during fourth pregnancy-other, specify\_\_\_\_\_

## Number of ED visits for complications during fourth pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10       >10

## Number of hospitalizations for complications during fourth pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10       >10



Patient Name \_\_\_\_\_

Date      

Duration of longest inpatient hospitalization (in days) for any reason during fourth pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during fourth pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during fourth pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during fourth pregnancy

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> Ammonia - Abn high                  |
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> CO <sub>2</sub> - Abn low           | <input type="checkbox"/> CO <sub>2</sub> - WNL               |
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

Patient Name \_\_\_\_\_

Date |M|M| |D|D| |Y|Y|

Other laboratory studies done on patient during fourth pregnancy: describe test(s) and result(s)

--

Additional interventions required during fourth pregnancy due to this metabolic condition

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                                    | <input type="checkbox"/> ED visits for hyperemesis/IV fluids   |
| <input type="checkbox"/> Hospitalizations for hyperemesis/IV fluids | <input type="checkbox"/> TPN                                   |
| <input type="checkbox"/> Tube feedings                              | <input type="checkbox"/> Additional medications                |
| <input type="checkbox"/> Home lab monitoring                        | <input type="checkbox"/> Increased frequency of lab monitoring |
| <input type="checkbox"/> More intensive fetal monitoring            | <input type="checkbox"/> Bedrest                               |
| <input type="checkbox"/> Other                                      |  |

Additional interventions required during fourth pregnancy due to this metabolic condition-other, specify

\_\_\_\_\_

Total maternal weight gain (in kg) during fourth pregnancy \_\_\_\_\_

Fourth pregnancy complicated by maternal HELLP syndrome     Unknown     Yes     NoFourth pregnancy complicated by acute fatty liver of pregnancy (AFLP)     Unknown     Yes     No

Number of documented episodes of hypoglycemia during fourth pregnancy

- Unknown    0         1         2         3         4         5  
 6         7         8         9         10       >10

Lowest maternal blood glucose value documented during fourth pregnancy \_\_\_\_\_

Lowest maternal blood glucose value documented during fourth pregnancy units     mmol/L     mg/dL

Number of documented episodes of rhabdomyolysis during fourth pregnancy

- Unknown    0         1         2         3         4         5  
 6         7         8         9         10       >10

Highest maternal CK value documented during fourth pregnancy \_\_\_\_\_

Highest maternal CK value documented during fourth pregnancy units     U/L     umol/L     mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during fourth pregnancy

- Unknown    1         2         3         4         5         6  
 7         8         9         10       11       12

Longest permitted fasting interval (in hours) during the NIGHT recommended during fourth pregnancy

- Unknown    1         2         3         4         5         6  
 7         8         9         10       11       13

L-carnitine supplementation used on a regular basis during fourth pregnancy

- Unknown                       Yes                       No  
 L-carnitine not prescribed

Patient Name \_\_\_\_\_

Date

Patient adhered to a fat-restricted diet during fourth pregnancy

- Unknown                       Yes                       No
- Fat-restriction not prescribed

Patient took a metabolic formula regularly during fourth pregnancy

- Unknown                       Yes
- No                       Metabolic formula not prescribed

Evidence of cardiac hypertrophy during fourth pregnancy

- Unknown                       Yes
- No                       Echocardiogram not performed during pregnancy

Evidence of any cardiac dysfunction during fourth pregnancy

- Unknown                       Yes
- No                       Cardiac testing not performed during pregnancy

Additional interventions planned for labor/delivery related to patient's disorder for fourth pregnancy

- Unknown                       None
- Additional maternal lab monitoring       Altered anesthesia plan
- Change in delivery site                       IV fluids
- Letter to OB/MFM specialist                       Planned C-section
- Referral for high risk OB management       Other

Additional interventions planned for labor/delivery related to patient's disorder for fourth pregnancy-other, specify

Actual interventions for labor/delivery related to patient's disorder for fourth pregnancy

- Unknown                       No
- Letter to OB/MFM specialist                       IV fluids
- Planned C-section                       Additional maternal lab monitoring
- Change in delivery site                       Referral for high risk OB management
- Altered anesthesia plan                       Other

Actual interventions for labor/delivery related to patient's disorder for fourth pregnancy-other, specify

Patient Name \_\_\_\_\_

Date

Acute health concerns experienced by the patient during delivery for fourth delivery

Abnormal patient lab results during fourth delivery

- Unknown                       None                       Normal labs during delivery
- Elevated CK                       Elevated liver function tests    Hyperammonemia
- Hypoglycemia                       Metabolic decompensation    Other

Abnormal patient lab results during fourth delivery-other, specify

Additional maternal interventions during or after fourth delivery related to this disorder

- Unknown                       Ammonul                       Blood/blood product transfusion
- Dialysis                       ICU monitoring                       Infusions
- Medications                       Resuscitation                       TPN
- Other

Additional maternal interventions during or after fourth delivery related to this disorder-other, specify

Patient death during or shortly after fourth delivery    Unknown    Yes    No

Disorder contributed to death of mother during fourth delivery    Unknown    Yes    No

Weeks gestation at time of fourth delivery - round to nearest week\_\_\_\_\_

Method of fourth delivery

- Unknown                       Caesarean section (scheduled or non-urgent)
- Casesarean section (emergent)                       Vaginal

Maternal inpatient days post- fourth delivery\_\_\_\_\_

Live delivery of fourth newborn    Unknown    Yes    No

Acute health concerns for fourth newborn related to maternal disorder (example: fetal distress secondary to maternal acute metabolic decompensation)

Patient Name \_\_\_\_\_

Date

Additional lab tests and results done on fourth newborn specifically due to maternal disorder history

Health concerns for fourth newborn not known to be related to disorder

Additional interventions for fourth newborn during or shortly after delivery (other than labs)

- Unknown  Blood/blood product transfusion
- Glucose infusion  Medications
- NICU/special care nursery monitoring  Resuscitation
- TPN  Other

Additional interventions for fourth newborn during or shortly after delivery-other, specify

Birth measurements for fourth newborn  Unknown  Head circumference  Length  Weight

Birth head circumference \_\_\_\_\_

Birth head circumference units  cm  in

Birth length \_\_\_\_\_

Birth length units  cm  in

Birth weight \_\_\_\_\_

Birth weight units  lbs  kg  gm  oz

APGAR score at 1 minute for fourth newborn

- Unknown
- 0
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10

Health concern(s) with APGAR score at one minute < 8 for fourth newborn

APGAR score at 5 minutes for fourth newborn

- Unknown
- 0
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10

Patient Name \_\_\_\_\_

Date      

Health concern(s) with APGAR score at five minutes &lt; 8 for fourth newborn

Length of fourth newborn's stay in the hospital after birth

- Unknown  <24 hours  
 24-28 hours  3-5 days  
 6-14 days  >14 days  
 N/A - baby was not born in a hospital  baby is still in the hospital

Routine newborn screening done for fourth newborn

- Unknown  Yes - normal for all screened disorders  
 Yes - abnormal  Yes - results pending  
 No

Describe abnormal newborn screen result for fourth newborn

Reason routine newborn screening was not done for fourth newborn

- Unknown  
 Died prior to collection of NBS  
 Refused  
 Transferred to another facility prior to NBS collection  
 Transfused prior to collection of NBS  
 Other

Reason routine newborn screening was not done for fourth newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the fourth baby after birth to rule out the mother's disorder

- Unknown  Yes - biochemical normal  
 Yes - biochemical abnormal  Yes - molecular normal  
 Yes - molecular abnormal affected  Yes - molecular abnormal unaffected carrier  
 No

Patient Name \_\_\_\_\_

Date      

Additional studies and results (examples: brain MRI, echocardiogram) done on fourth newborn to assess for effects of maternal disorder

Fourth newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

Unknown  Yes  No

Abnormal newborn exam findings for fourth newborn

- |   |  |   |
|---|--|---|
| <input type="checkbox"/> Unknown                    | <input type="checkbox"/> None                      | <input type="checkbox"/> Congenital heart disease |
| <input type="checkbox"/> Dysmorphism                | <input type="checkbox"/> Lethargy                  | <input type="checkbox"/> Microcephaly             |
| <input type="checkbox"/> Other congenital anomalies | <input type="checkbox"/> Poor feeding              | <input type="checkbox"/> Respiratory distress     |
| <input type="checkbox"/> Seizure(s)                 | <input type="checkbox"/> Small for gestational age | <input type="checkbox"/> Other                    |

Abnormal newborn exam findings for fourth newborn-other, specify \_\_\_\_\_

Newborn death at or shortly after delivery for fourth newborn  Unknown  Yes  No

Newborn's death related to maternal disorder for fourth newborn  Unknown  Yes  No

Fourth newborn currently alive  Unknown  Yes  No

## **Fifth Pregnancy**

---

Patient's age at time of fifth pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before fifth pregnancy (in months)

- Unknown  Unplanned  1  2  3  4  5  
 6  7  8  9  10  11  12  
 13-24  25-36  >36

History of infertility for fifth pregnancy

- Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for fifth pregnancy

- Unknown  Male factor  Female factor  
 Combined male-female factor

Cause(s) of male infertility for fifth pregnancy

- Unknown  Abnormal sperm production or function  
 Age  Cancer/cancer treatment related  
 Environmental  Problems with sperm delivery

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

- Tobacco use
- Other

Cause(s) of male infertility for fifth pregnancy-other, specify \_\_\_\_\_

Cause(s) of female infertility for fifth pregnancy

- Unknown
- Age
- Alcohol use
- Cancer/cancer treatment related
- Early menopause
- Fallopian tube damage/blockage
- Hyperprolactinemia
- Pelvic adhesions
- Polycystic ovary syndrome (PCOS)
- Premature ovarian insufficiency
- Over-exercise
- Ovulation disorder
- Thyroid problems
- Tobacco use
- Uterine fibroid
- Weight related
- Other

Cause(s) of female infertility for fifth pregnancy-other, specify \_\_\_\_\_

Cause(s) of infertility for fifth pregnancy, combined male-female factor- specify \_\_\_\_\_

Types of fertility treatment(s) received prior to fifth pregnancy

- Unknown
- None
- Assisted reproductive technology
- Fertility drugs
- Surgery

Type of assisted reproductive technologies used during fifth pregnancy

- Assisted hatching
- ICSI
- IVF
- Surgical sperm aspiration
- Other

Type of assisted reproductive technologies used during fifth pregnancy-other, specify \_\_\_\_\_

Partner tested for patient's disorder during fifth pregnancy

- Unknown
- Yes-biochemical
- Yes-molecular
- No

Partner also affected by disorder during fifth pregnancy  Unknown  Yes  No

Preimplantation genetic diagnosis for disorder done for fifth pregnancy  Unknown  Yes  No

Prenatal testing done for fetus for this disorder for fifth pregnancy  Unknown  Yes  No

Type of prenatal testing performed for fifth pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for fifth pregnancy

- Unknown
- Amniocentesis
- Chorionic villus sampling

Additional prenatal testing performed on fetus as a result of parent's disorder for fifth pregnancy

- Unknown
- None
- Fetal echocardiogram
- Increased frequency prenatal ultrasounds



Patient Name \_\_\_\_\_

Date       Other

Additional prenatal testing performed on fetus as a result of parent's disorder for fifth pregnancy-other, specify \_\_\_\_\_

Fifth pregnancy terminated  Unknown  Yes  No

Reason fifth pregnancy terminated

- Elective due to fetus affected with disorder
- Elective due to other fetal well-being unrelated to disorder
- Elective for other reason
- Elective due to maternal well-being
- Spontaneous

Gestational age (in weeks) at time of fifth pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for fifth pregnancy \_\_\_\_\_

Prenatal care received during fifth pregnancy  Unknown  Yes  No

Weeks gestation prenatal care started during fifth pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during fifth pregnancy

- Unknown  Metabolic diet  Medications
- Biochemical lab monitoring  Avoidance of fasting  Other

Treatment prescribed for patient's disorder during fifth pregnancy-other, specify

Patient in good metabolic condition prior to fifth pregnancy  Unknown  Yes  NoPatient in good metabolic condition during first trimester of fifth pregnancy  Unknown  Yes  No

Patient in good metabolic condition during second trimester of fifth pregnancy

- Unknown  Pregnancy not sustained to second trimester
- Yes  No

Patient in good metabolic condition during third trimester of fifth pregnancy

- Unknown  Pregnancy not sustained to third trimester
- Yes  No

Number of outpatient metabolic visits for patient during fifth pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during fifth pregnancy

- Unknown  0  1  2  3  4  5
- 6  7  8  9  10  >10

Patient Name \_\_\_\_\_

Date |M|M| |D|D| |Y|Y|

Number of hospitalizations for management of disorder during fifth pregnancy

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10       >10

Complications during fifth pregnancy

- Unknown                                       Yes-related to disorder  
 Yes- not known to be related to disorder    No

Complications related to disorder during fifth pregnancy- specify

- Unknown  
 Acute fatty liver of pregnancy (AFLP)  
 Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)  
 Intrauterine growth restriction (AUGR)  
 Mother affected with this condition  
 Other

Complications not known to be related to disorder during fifth pregnancy-specify

- Unknown  
 Advanced maternal age (35+ years of age)  
 Ectopic pregnancy  
 Gestational diabetes  
 Group B strep  
 Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)  
 Intrauterine growth restriction (AUGR)  
 Inadequate prenatal care  
 Maternal prenatal substance exposure  
 Preeclampsia  
 Rh isoimmunization  
 Toxemia  
 Young maternal age (15 years of age + under)  
 Preterm labor  
 Other

Complications during fifth pregnancy-other, specify \_\_\_\_\_

Number of ED visits for complications during fifth pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10       >10

Number of hospitalizations for complications during fifth pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5

Patient Name \_\_\_\_\_

Date      

6     
  7     
  8     
  9     
  10     
  >10

Duration of longest inpatient hospitalization (in days) for any reason during fifth pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during fifth pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during fifth pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during fifth pregnancy

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> Ammonia - Abn high                  |
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> CO <sub>2</sub> - Abn low           | <input type="checkbox"/> CO <sub>2</sub> - WNL               |
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

Patient Name \_\_\_\_\_

Date |||||

Other laboratory studies done on patient during fifth pregnancy: describe test(s) and result(s)

--

Additional interventions required during fifth pregnancy due to this metabolic condition

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                                    | <input type="checkbox"/> ED visits for hyperemesis/IV fluids   |
| <input type="checkbox"/> Hospitalizations for hyperemesis/IV fluids | <input type="checkbox"/> TPN                                   |
| <input type="checkbox"/> Tube feedings                              | <input type="checkbox"/> Additional medications                |
| <input type="checkbox"/> Home lab monitoring                        | <input type="checkbox"/> Increased frequency of lab monitoring |
| <input type="checkbox"/> More intensive fetal monitoring            | <input type="checkbox"/> Bedrest                               |
| <input type="checkbox"/> Other                                      |  |

Additional interventions required during fifth pregnancy due to this metabolic condition-other, specify

\_\_\_\_\_

Total maternal weight gain (in kg) during fifth pregnancy \_\_\_\_\_

Fifth pregnancy complicated by maternal HELLP syndrome  Unknown  Yes  NoFifth pregnancy complicated by acute fatty liver of pregnancy (AFLP)  Unknown  Yes  No

Number of documented episodes of hypoglycemia during fifth pregnancy

- Unknown  0  1  2  3  4  5  
 6  7  8  9  10  >10

Lowest maternal blood glucose value documented during fifth pregnancy \_\_\_\_\_

Lowest maternal blood glucose value documented during fifth pregnancy units  mmol/L  mg/dL

Number of documented episodes of rhabdomyolysis during fifth pregnancy

- Unknown  0  1  2  3  4  5  
 6  7  8  9  10  >10

Highest maternal CK value documented during fifth pregnancy \_\_\_\_\_

Highest maternal CK value documented during fifth pregnancy units  U/L  umol/L  mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during fifth pregnancy

- Unknown  1  2  3  4  5  6  
 7  8  9  10  11  12

Longest permitted fasting interval (in hours) during the NIGHT recommended during fifth pregnancy

- Unknown  1  2  3  4  5  6  
 7  8  9  10  11  13

L-carnitine supplementation used on a regular basis during fifth pregnancy

- Unknown  Yes  No  
 L-carnitine not prescribed

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

Patient adhered to a fat-restricted diet during fifth pregnancy

- Unknown                       Yes                       No
- Fat-restriction not prescribed

Patient took a metabolic formula regularly during fifth pregnancy

- Unknown                       Yes
- No                       Metabolic formula not prescribed

Evidence of cardiac hypertrophy during fifth pregnancy

- Unknown                       Yes
- No                       Echocardiogram not performed during pregnancy

Evidence of any cardiac dysfunction during fifth pregnancy

- Unknown                       Yes
- No                       Cardiac testing not performed during pregnancy

Additional interventions planned for labor/delivery related to patient's disorder for fifth pregnancy

- Unknown                       None
- Additional maternal lab monitoring       Altered anesthesia plan
- Change in delivery site                       IV fluids
- Letter to OB/MFM specialist                       Planned C-section
- Referral for high risk OB management       Other

Additional interventions planned for labor/delivery related to patient's disorder for fifth pregnancy-other, specify

Actual interventions for labor/delivery related to patient's disorder for fifth pregnancy

- Unknown                       No
- Letter to OB/MFM specialist                       IV fluids
- Planned C-section                       Additional maternal lab monitoring
- Change in delivery site                       Referral for high risk OB management
- Altered anesthesia plan                       Other

Actual interventions for labor/delivery related to patient's disorder for fifth pregnancy-other, specify

Patient Name \_\_\_\_\_

Date

Acute health concerns experienced by the patient during delivery for fifth delivery

Abnormal patient lab results during fifth delivery

- Unknown                       None                       Normal labs during delivery
- Elevated CK                       Elevated liver function tests    Hyperammonemia
- Hypoglycemia                       Metabolic decompensation    Other

Abnormal patient lab results during fifth delivery-other, specify

Additional maternal interventions during or after fifth delivery related to this disorder

- Unknown                       Ammonul                       Blood/blood product transfusion
- Dialysis                       ICU monitoring                       Infusions
- Medications                       Resuscitation                       TPN
- Other

Additional maternal interventions during or after fifth delivery related to this disorder-other, specify

Patient death during or shortly after fifth delivery    Unknown    Yes    No

Disorder contributed to death of mother during fifth delivery    Unknown    Yes    No

Weeks gestation at time of fifth delivery - round to nearest week \_\_\_\_\_

Method of fifth delivery

- Unknown                                       Caesarean section (scheduled or non-urgent)
- Casesarean section (emergent)                       Vaginal

Maternal inpatient days post- fifth delivery \_\_\_\_\_

Live delivery of fifth newborn    Unknown    Yes    No

Acute health concerns for fifth newborn related to maternal disorder (example: fetal distress secondary to maternal acute metabolic decompensation)

Patient Name \_\_\_\_\_

Date |M|M| |D|D| |Y|Y|

Additional lab tests and results done on fifth newborn specifically due to maternal disorder history

[Empty text box for lab tests and results]

Health concerns for fifth newborn not known to be related to disorder

[Empty text box for health concerns]

Additional interventions for fifth newborn during or shortly after delivery (other than labs)

- Unknown
- Blood/blood product transfusion
- Glucose infusion
- Medications
- NICU/special care nursery monitoring
- Resuscitation
- TPN
- Other

Additional interventions for fifth newborn during or shortly after delivery-other, specify

[Empty text box for additional interventions]

Birth measurements for fifth newborn  Unknown  Head circumference  Length  Weight

Birth head circumference \_\_\_\_\_

Birth head circumference units  cm  in

Birth length \_\_\_\_\_

Birth length units  cm  in

Birth weight \_\_\_\_\_

Birth weight units  lbs  kg  gm  oz

APGAR score at 1 minute for fifth newborn

- Unknown  0  1  2  3  4  5
- 6  7  8  9  10

Health concern(s) with APGAR score at one minute < 8 for fifth newborn

[Empty text box for health concerns with APGAR score < 8]

APGAR score at 5 minutes for fifth newborn

- Unknown  0  1  2  3  4  5
- 6  7  8  9  10

Patient Name \_\_\_\_\_

Date      

Health concern(s) with APGAR score at five minutes &lt; 8 for fifth newborn

Length of fifth newborn's stay in the hospital after birth

- Unknown  <24 hours  
 24-28 hours  3-5 days  
 6-14 days  >14 days  
 N/A - baby was not born in a hospital  baby is still in the hospital

Routine newborn screening done for fifth newborn

- Unknown  Yes - normal for all screened disorders  
 Yes - abnormal  Yes - results pending  
 No

Describe abnormal newborn screen result for fifth newborn

Reason routine newborn screening was not done for fifth newborn

- Unknown  
 Died prior to collection of NBS  
 Refused  
 Transferred to another facility prior to NBS collection  
 Transfused prior to collection of NBS  
 Other

Reason routine newborn screening was not done for fifth newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the fifth baby after birth to rule out the mother's disorder

- Unknown  Yes - biochemical normal  
 Yes - biochemical abnormal  Yes - molecular normal  
 Yes - molecular abnormal affected  Yes - molecular abnormal unaffected carrier  
 No



Patient Name \_\_\_\_\_

Date      

Additional studies and results (examples: brain MRI, echocardiogram) done on fifth newborn to assess for effects of maternal disorder

Fifth newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

Unknown  Yes  No

Abnormal newborn exam findings for fifth newborn

Unknown  None  Congenital heart disease  
 Dysmorphism  Lethargy  Microcephaly  
 Other congenital anomalies  Poor feeding  Respiratory distress  
 Seizure(s)  Small for gestational age  Other

Abnormal newborn exam findings for fifth newborn-other, specify \_\_\_\_\_

Newborn death at or shortly after delivery for fifth newborn  Unknown  Yes  No

Newborn's death related to maternal disorder for fifth newborn  Unknown  Yes  No

Fifth newborn currently alive  Unknown  Yes  No

## Sixth Pregnancy

Patient's age at time of sixth pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before sixth pregnancy (in months)

Unknown  Unplanned  1  2  3  4  5  
 6  7  8  9  10  11  12  
 13-24  25-36  >36

History of infertility for sixth pregnancy

Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for sixth pregnancy

Unknown  Male factor  Female factor  
 Combined male-female factor

Cause(s) of male infertility for sixth pregnancy

Unknown  Abnormal sperm production or function  
 Age  Cancer/cancer treatment related  
 Environmental  Problems with sperm delivery  
 Tobacco use  Other

Patient Name \_\_\_\_\_

Date      

Cause(s) of male infertility for sixth pregnancy-other, specify \_\_\_\_\_

Cause(s) of female infertility for sixth pregnancy

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                          | <input type="checkbox"/> Age                             |
| <input type="checkbox"/> Alcohol use                      | <input type="checkbox"/> Cancer/cancer treatment related |
| <input type="checkbox"/> Early menopause                  | <input type="checkbox"/> Fallopian tube damage/blockage  |
| <input type="checkbox"/> Hyperprolactinemia               | <input type="checkbox"/> Pelvic adhesions                |
| <input type="checkbox"/> Polycystic ovary syndrome (PCOS) | <input type="checkbox"/> Premature ovarian insufficiency |
| <input type="checkbox"/> Over-exercise                    | <input type="checkbox"/> Ovulation disorder              |
| <input type="checkbox"/> Thyroid problems                 | <input type="checkbox"/> Tobacco use                     |
| <input type="checkbox"/> Uterine fibroid                  | <input type="checkbox"/> Weight related                  |
| <input type="checkbox"/> Other                            |  |

Cause(s) of female infertility for sixth pregnancy-other, specify \_\_\_\_\_

Cause(s) of infertility for sixth pregnancy, combined male-female factor- specify

\_\_\_\_\_

Types of fertility treatment(s) received prior to sixth pregnancy

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                          | <input type="checkbox"/> None            |
| <input type="checkbox"/> Assisted reproductive technology | <input type="checkbox"/> Fertility drugs |
| <input type="checkbox"/> Surgery                          |  |

Type of assisted reproductive technologies used during sixth pregnancy

- |  |                                |                              |
|--|--------------------------------|------------------------------|
| <input type="checkbox"/> Assisted hatching         | <input type="checkbox"/> ICSI  | <input type="checkbox"/> IVF |
| <input type="checkbox"/> Surgical sperm aspiration | <input type="checkbox"/> Other |                              |

Type of assisted reproductive technologies used during sixth pregnancy-other, specify

\_\_\_\_\_

Partner tested for patient's disorder during sixth pregnancy

- |                                  |  |  |                             |
|----------------------------------|--|--|-----------------------------|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Yes-biochemical | <input type="checkbox"/> Yes-molecular | <input type="checkbox"/> No |
|----------------------------------|--|--|-----------------------------|

Partner also affected by disorder during sixth pregnancy  Unknown  Yes  NoPreimplantation genetic diagnosis for disorder done for sixth pregnancy  Unknown  Yes  NoPrenatal testing done for fetus for this disorder for sixth pregnancy  Unknown  Yes  NoType of prenatal testing performed for sixth pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for sixth pregnancy

- |                                  |  |  |
|----------------------------------|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Amniocentesis | <input type="checkbox"/> Chorionic villus sampling |
|----------------------------------|--|--|

Additional prenatal testing performed on fetus as a result of parent's disorder for sixth pregnancy

- |   |   |
|---|---|
| <input type="checkbox"/> Unknown              | <input type="checkbox"/> None                                     |
| <input type="checkbox"/> Fetal echocardiogram | <input type="checkbox"/> Increased frequency prenatal ultrasounds |
| <input type="checkbox"/> Other                |   |

Patient Name \_\_\_\_\_

Date      

Additional prenatal testing performed on fetus as a result of parent's disorder for sixth pregnancy-other, specify \_\_\_\_\_

Sixth pregnancy terminated  Unknown  Yes  No

Reason sixth pregnancy terminated

- Elective due to fetus affected with disorder  
 Elective due to other fetal well-being unrelated to disorder  
 Elective for other reason  
 Elective due to maternal well-being  
 Spontaneous

Gestational age (in weeks) at time of sixth pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for sixth pregnancy \_\_\_\_\_

Prenatal care received during sixth pregnancy  Unknown  Yes  No

Weeks gestation prenatal care started during sixth pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during sixth pregnancy

- Unknown  Metabolic diet  Medications  
 Biochemical lab monitoring  Avoidance of fasting  Other

Treatment prescribed for patient's disorder during sixth pregnancy-other, specify

Patient in good metabolic condition prior to sixth pregnancy  Unknown  Yes  No

Patient in good metabolic condition during first trimester of sixth pregnancy  Unknown  Yes  No

Patient in good metabolic condition during second trimester of sixth pregnancy

- Unknown  Pregnancy not sustained to second trimester  
 Yes  No

Patient in good metabolic condition during third trimester of sixth pregnancy

- Unknown  Pregnancy not sustained to third trimester  
 Yes  No

Number of outpatient metabolic visits for patient during sixth pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during sixth pregnancy

- Unknown  0  1  2  3  4  5  
 6  7  8  9  10  >10

Number of hospitalizations for management of disorder during sixth pregnancy

Patient Name \_\_\_\_\_

Date |M|M| |D|D| |Y|Y|

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10       >10

## Complications during sixth pregnancy

- Unknown                                   Yes-related to disorder  
 Yes- not known to be related to disorder    No

## Complications related to disorder during sixth pregnancy- specify

- Unknown  
 Acute fatty liver of pregnancy (AFLP)  
 Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)  
 Intrauterine growth restriction (AUGR)  
 Mother affected with this condition  
 Other

## Complications not known to be related to disorder during sixth pregnancy-specify

- Unknown  
 Advanced maternal age (35+ years of age)  
 Ectopic pregnancy  
 Gestational diabetes  
 Group B strep  
 Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)  
 Intrauterine growth restriction (AUGR)  
 Inadequate prenatal care  
 Maternal prenatal substance exposure  
 Preeclampsia  
 Rh isoimmunization  
 Toxemia  
 Young maternal age (15 years of age + under)  
 Preterm labor  
 Other

## Complications during sixth pregnancy-other, specify\_\_\_\_\_

## Number of ED visits for complications during sixth pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10       >10

## Number of hospitalizations for complications during sixth pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10       >10

Patient Name \_\_\_\_\_

Date      

Duration of longest inpatient hospitalization (in days) for any reason during sixth pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during sixth pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during sixth pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during sixth pregnancy

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> Ammonia - Abn high                  |
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> CO <sub>2</sub> - Abn low           | <input type="checkbox"/> CO <sub>2</sub> - WNL               |
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

Patient Name \_\_\_\_\_

Date |M|M| |D|D| |Y|Y|

Other laboratory studies done on patient during sixth pregnancy: describe test(s) and result(s)

--

Additional interventions required during sixth pregnancy due to this metabolic condition

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                                    | <input type="checkbox"/> ED visits for hyperemesis/IV fluids   |
| <input type="checkbox"/> Hospitalizations for hyperemesis/IV fluids | <input type="checkbox"/> TPN                                   |
| <input type="checkbox"/> Tube feedings                              | <input type="checkbox"/> Additional medications                |
| <input type="checkbox"/> Home lab monitoring                        | <input type="checkbox"/> Increased frequency of lab monitoring |
| <input type="checkbox"/> More intensive fetal monitoring            | <input type="checkbox"/> Bedrest                               |
| <input type="checkbox"/> Other                                      |  |

Additional interventions required during sixth pregnancy due to this metabolic condition-other, specify

\_\_\_\_\_

Total maternal weight gain (in kg) during sixth pregnancy \_\_\_\_\_

Sixth pregnancy complicated by maternal HELLP syndrome     Unknown     Yes     NoSixth pregnancy complicated by acute fatty liver of pregnancy (AFLP)     Unknown     Yes     No

Number of documented episodes of hypoglycemia during sixth pregnancy

- Unknown    0         1         2         3         4         5  
 6         7         8         9         10       >10

Lowest maternal blood glucose value documented during sixth pregnancy \_\_\_\_\_

Lowest maternal blood glucose value documented during sixth pregnancy units     mmol/L     mg/dL

Number of documented episodes of rhabdomyolysis during sixth pregnancy

- Unknown    0         1         2         3         4         5  
 6         7         8         9         10       >10

Highest maternal CK value documented during sixth pregnancy \_\_\_\_\_

Highest maternal CK value documented during sixth pregnancy units     U/L     umol/L     mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during sixth pregnancy

- Unknown    1         2         3         4         5         6  
 7         8         9         10       11       12

Longest permitted fasting interval (in hours) during the NIGHT recommended during sixth pregnancy

- Unknown    1         2         3         4         5         6  
 7         8         9         10       11       13

L-carnitine supplementation used on a regular basis during sixth pregnancy

- Unknown                       Yes                       No  
 L-carnitine not prescribed

Patient Name \_\_\_\_\_

Date

Patient adhered to a fat-restricted diet during sixth pregnancy

- Unknown  Yes  No
- Fat-restriction not prescribed

Patient took a metabolic formula regularly during sixth pregnancy

- Unknown  Yes
- No  Metabolic formula not prescribed

Evidence of cardiac hypertrophy during sixth pregnancy

- Unknown  Yes
- No  Echocardiogram not performed during pregnancy

Evidence of any cardiac dysfunction during sixth pregnancy

- Unknown  Yes
- No  Cardiac testing not performed during pregnancy

Additional interventions planned for labor/delivery related to patient's disorder for sixth pregnancy

- Unknown  None
- Additional maternal lab monitoring  Altered anesthesia plan
- Change in delivery site  IV fluids
- Letter to OB/MFM specialist  Planned C-section
- Referral for high risk OB management  Other

Additional interventions planned for labor/delivery related to patient's disorder for sixth pregnancy-other, specify

Actual interventions for labor/delivery related to patient's disorder for sixth pregnancy

- Unknown  No
- Letter to OB/MFM specialist  IV fluids
- Planned C-section  Additional maternal lab monitoring
- Change in delivery site  Referral for high risk OB management
- Altered anesthesia plan  Other

Actual interventions for labor/delivery related to patient's disorder for sixth pregnancy-other, specify

Patient Name \_\_\_\_\_

Date

Acute health concerns experienced by the patient during delivery for sixth delivery

Abnormal patient lab results during sixth delivery

- Unknown                       None                       Normal labs during delivery
- Elevated CK                       Elevated liver function tests    Hyperammonemia
- Hypoglycemia                       Metabolic decompensation    Other

Abnormal patient lab results during sixth delivery-other, specify

Additional maternal interventions during or after sixth delivery related to this disorder

- Unknown                       Ammonul                       Blood/blood product transfusion
- Dialysis                       ICU monitoring                       Infusions
- Medications                       Resuscitation                       TPN
- Other

Additional maternal interventions during or after sixth delivery related to this disorder-other, specify

Patient death during or shortly after sixth delivery    Unknown    Yes    No

Disorder contributed to death of mother during sixth delivery    Unknown    Yes    No

Weeks gestation at time of sixth delivery - round to nearest week\_\_\_\_\_

Method of sixth delivery

- Unknown                                       Caesarean section (scheduled or non-urgent)
- Casesarean section (emergent)                       Vaginal

Maternal inpatient days post- sixth delivery\_\_\_\_\_

Live delivery of sixth newborn    Unknown    Yes    No

Acute health concerns for sixth newborn related to maternal disorder (example: fetal distress secondary to maternal acute metabolic decompensation)



Patient Name \_\_\_\_\_

Date

Additional lab tests and results done on sixth newborn specifically due to maternal disorder history

Health concerns for sixth newborn not known to be related to disorder

Additional interventions for sixth newborn during or shortly after delivery (other than labs)

- Unknown  Blood/blood product transfusion
- Glucose infusion  Medications
- NICU/special care nursery monitoring  Resuscitation
- TPN  Other

Additional interventions for sixth newborn during or shortly after delivery-other, specify

Birth measurements for sixth newborn  Unknown  Head circumference  Length  Weight

Birth head circumference \_\_\_\_\_

Birth head circumference units  cm  in

Birth length \_\_\_\_\_

Birth length units  cm  in

Birth weight \_\_\_\_\_

Birth weight units  lbs  kg  gm  oz

APGAR score at 1 minute for sixth newborn

- Unknown
- 0
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10

Health concern(s) with APGAR score at one minute < 8 for sixth newborn

APGAR score at 5 minutes for sixth newborn

- Unknown
- 0
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10

Patient Name \_\_\_\_\_

Date |||||

Health concern(s) with APGAR score at five minutes &lt; 8 for sixth newborn

Length of sixth newborn's stay in the hospital after birth

- Unknown  <24 hours  
 24-28 hours  3-5 days  
 6-14 days  >14 days  
 N/A - baby was not born in a hospital  baby is still in the hospital

Routine newborn screening done for sixth newborn

- Unknown  Yes - normal for all screened disorders  
 Yes - abnormal  Yes - results pending  
 No

Describe abnormal newborn screen result for sixth newborn

Reason routine newborn screening was not done for sixth newborn

- Unknown  
 Died prior to collection of NBS  
 Refused  
 Transferred to another facility prior to NBS collection  
 Transfused prior to collection of NBS  
 Other

Reason routine newborn screening was not done for sixth newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the sixth baby after birth to rule out the mother's disorder

- Unknown  Yes - biochemical normal  
 Yes - biochemical abnormal  Yes - molecular normal  
 Yes - molecular abnormal affected  Yes - molecular abnormal unaffected carrier  
 No

Patient Name \_\_\_\_\_

Date      

Additional studies and results (examples: brain MRI, echocardiogram) done on sixth newborn to assess for effects of maternal disorder

Sixth newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

Unknown  Yes  No

Abnormal newborn exam findings for sixth newborn

Unknown  None  Congenital heart disease  
 Dysmorphism  Lethargy  Microcephaly  
 Other congenital anomalies  Poor feeding  Respiratory distress  
 Seizure(s)  Small for gestational age  Other

Abnormal newborn exam findings for sixth newborn-other, specify \_\_\_\_\_

Newborn death at or shortly after delivery for sixth newborn  Unknown  Yes  No

Newborn's death related to maternal disorder for sixth newborn  Unknown  Yes  No

Sixth newborn currently alive  Unknown  Yes  No

## Seventh Pregnancy

Patient's age at time of seventh pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before seventh pregnancy (in months)

Unknown  Unplanned  1  2  3  4  5  
 6  7  8  9  10  11  12  
 13-24  25-36  >36

History of infertility for seventh pregnancy

Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for seventh pregnancy

Unknown  Male factor  Female factor  
 Combined male-female factor

Cause(s) of male infertility for seventh pregnancy

Unknown  Abnormal sperm production or function  
 Age  Cancer/cancer treatment related  
 Environmental  Problems with sperm delivery  
 Tobacco use  Other

Patient Name \_\_\_\_\_

Date      

Cause(s) of male infertility for seventh pregnancy-other, specify \_\_\_\_\_

Cause(s) of female infertility for seventh pregnancy

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                          | <input type="checkbox"/> Age                             |
| <input type="checkbox"/> Alcohol use                      | <input type="checkbox"/> Cancer/cancer treatment related |
| <input type="checkbox"/> Early menopause                  | <input type="checkbox"/> Fallopian tube damage/blockage  |
| <input type="checkbox"/> Hyperprolactinemia               | <input type="checkbox"/> Pelvic adhesions                |
| <input type="checkbox"/> Polycystic ovary syndrome (PCOS) | <input type="checkbox"/> Premature ovarian insufficiency |
| <input type="checkbox"/> Over-exercise                    | <input type="checkbox"/> Ovulation disorder              |
| <input type="checkbox"/> Thyroid problems                 | <input type="checkbox"/> Tobacco use                     |
| <input type="checkbox"/> Uterine fibroid                  | <input type="checkbox"/> Weight related                  |
| <input type="checkbox"/> Other                            |  |

Cause(s) of female infertility for seventh pregnancy-other, specify \_\_\_\_\_

Cause(s) of infertility for seventh pregnancy, combined male-female factor- specify

\_\_\_\_\_

Types of fertility treatment(s) received prior to seventh pregnancy

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                          | <input type="checkbox"/> None            |
| <input type="checkbox"/> Assisted reproductive technology | <input type="checkbox"/> Fertility drugs |
| <input type="checkbox"/> Surgery                          |  |

Type of assisted reproductive technologies used during seventh pregnancy

- |  |                                |                              |
|--|--------------------------------|------------------------------|
| <input type="checkbox"/> Assisted hatching         | <input type="checkbox"/> ICSI  | <input type="checkbox"/> IVF |
| <input type="checkbox"/> Surgical sperm aspiration | <input type="checkbox"/> Other |                              |

Type of assisted reproductive technologies used during seventh pregnancy-other, specify

\_\_\_\_\_

Partner tested for patient's disorder during seventh pregnancy

- |                                  |  |  |                             |
|----------------------------------|--|--|-----------------------------|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Yes-biochemical | <input type="checkbox"/> Yes-molecular | <input type="checkbox"/> No |
|----------------------------------|--|--|-----------------------------|

Partner also affected by disorder during seventh pregnancy  Unknown  Yes  NoPreimplantation genetic diagnosis for disorder done for seventh pregnancy  Unknown  Yes  NoPrenatal testing done for fetus for this disorder for seventh pregnancy  Unknown  Yes  NoType of prenatal testing performed for seventh pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for seventh pregnancy

- |                                  |  |  |
|----------------------------------|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Amniocentesis | <input type="checkbox"/> Chorionic villus sampling |
|----------------------------------|--|--|

Additional prenatal testing performed on fetus as a result of parent's disorder for seventh pregnancy

- |   |   |
|---|---|
| <input type="checkbox"/> Unknown              | <input type="checkbox"/> None                                     |
| <input type="checkbox"/> Fetal echocardiogram | <input type="checkbox"/> Increased frequency prenatal ultrasounds |
| <input type="checkbox"/> Other                |   |

Patient Name \_\_\_\_\_

Date

Additional prenatal testing performed on fetus as a result of parent's disorder for seventh pregnancy-other, specify \_\_\_\_\_

Seventh pregnancy terminated Unknown Yes No

Reason seventh pregnancy terminated

- Elective due to fetus affected with disorder
- Elective due to other fetal well-being unrelated to disorder
- Elective for other reason
- Elective due to maternal well-being
- Spontaneous

Gestational age (in weeks) at time of seventh pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for seventh pregnancy \_\_\_\_\_

Prenatal care received during seventh pregnancy Unknown Yes No

Weeks gestation prenatal care started during seventh pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during seventh pregnancy

- Unknown Metabolic diet Medications
- Biochemical lab monitoring Avoidance of fasting Other

Treatment prescribed for patient's disorder during seventh pregnancy-other, specify

Patient in good metabolic condition prior to seventh pregnancy Unknown Yes No

Patient in good metabolic condition during first trimester of seventh pregnancy Unknown Yes No

Patient in good metabolic condition during second trimester of seventh pregnancy

- Unknown Pregnancy not sustained to second trimester
- Yes No

Patient in good metabolic condition during third trimester of seventh pregnancy

- Unknown Pregnancy not sustained to third trimester
- Yes No

Number of outpatient metabolic visits for patient during seventh pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during seventh pregnancy

- Unknown 0 1 2 3 4 5
- 6 7 8 9 10 >10

Number of hospitalizations for management of disorder during seventh pregnancy

Patient Name _____
Date  M M   D D   Y Y

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10       >10

Complications during seventh pregnancy

- Unknown       Yes-related to disorder  
 Yes- not known to be related to disorder     No

Complications related to disorder during seventh pregnancy- specify

- Unknown  
 Acute fatty liver of pregnancy (AFLP)  
 Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)  
 Intrauterine growth restriction (AUGR)  
 Mother affected with this condition  
 Other

Complications not known to be related to disorder during seventh pregnancy-specify

- Unknown  
 Advanced maternal age (35+ years of age)  
 Ectopic pregnancy  
 Gestational diabetes  
 Group B strep  
 Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)  
 Intrauterine growth restriction (AUGR)  
 Inadequate prenatal care  
 Maternal prenatal substance exposure  
 Preeclampsia  
 Rh isoimmunization  
 Toxemia  
 Young maternal age (15 years of age + under)  
 Preterm labor  
 Other

Complications during seventh pregnancy-other, specify\_\_\_\_\_

Number of ED visits for complications during seventh pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10       >10

Number of hospitalizations for complications during seventh pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10       >10

Patient Name \_\_\_\_\_

Date |||||

Duration of longest inpatient hospitalization (in days) for any reason during seventh pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during seventh pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during seventh pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during seventh pregnancy

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> Ammonia - Abn high                  |
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> CO2 - Abn low                       | <input type="checkbox"/> CO2 - WNL                           |
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

Patient Name \_\_\_\_\_

Date |M|M| |D|D| |Y|Y|

Other laboratory studies done on patient during seventh pregnancy: describe test(s) and result(s)

--

Additional interventions required during seventh pregnancy due to this metabolic condition

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                                    | <input type="checkbox"/> ED visits for hyperemesis/IV fluids   |
| <input type="checkbox"/> Hospitalizations for hyperemesis/IV fluids | <input type="checkbox"/> TPN                                   |
| <input type="checkbox"/> Tube feedings                              | <input type="checkbox"/> Additional medications                |
| <input type="checkbox"/> Home lab monitoring                        | <input type="checkbox"/> Increased frequency of lab monitoring |
| <input type="checkbox"/> More intensive fetal monitoring            | <input type="checkbox"/> Bedrest                               |
| <input type="checkbox"/> Other                                      |  |

Additional interventions required during seventh pregnancy due to this metabolic condition-other, specify

\_\_\_\_\_

Total maternal weight gain (in kg) during seventh pregnancy \_\_\_\_\_

Seventh pregnancy complicated by maternal HELLP syndrome     Unknown     Yes     NoSeventh pregnancy complicated by acute fatty liver of pregnancy (AFLP)     Unknown     Yes     No

Number of documented episodes of hypoglycemia during seventh pregnancy

- Unknown    0         1         2         3         4         5  
 6         7         8         9         10       >10

Lowest maternal blood glucose value documented during seventh pregnancy \_\_\_\_\_

Lowest maternal blood glucose value documented during seventh pregnancy units     mmol/L     mg/dL

Number of documented episodes of rhabdomyolysis during seventh pregnancy

- Unknown    0         1         2         3         4         5  
 6         7         8         9         10       >10

Highest maternal CK value documented during seventh pregnancy \_\_\_\_\_

Highest maternal CK value documented during seventh pregnancy units     U/L     umol/L     mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during seventh pregnancy

- Unknown    1         2         3         4         5         6  
 7         8         9         10       11       12

Longest permitted fasting interval (in hours) during the NIGHT recommended during seventh pregnancy

- Unknown    1         2         3         4         5         6  
 7         8         9         10       11       13

L-carnitine supplementation used on a regular basis during seventh pregnancy

- Unknown                       Yes                       No  
 L-carnitine not prescribed



Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

Patient adhered to a fat-restricted diet during seventh pregnancy

- Unknown                       Yes                       No
- Fat-restriction not prescribed

Patient took a metabolic formula regularly during seventh pregnancy

- Unknown                       Yes
- No                       Metabolic formula not prescribed

Evidence of cardiac hypertrophy during seventh pregnancy

- Unknown                       Yes
- No                       Echocardiogram not performed during pregnancy

Evidence of any cardiac dysfunction during seventh pregnancy

- Unknown                       Yes
- No                       Cardiac testing not performed during pregnancy

Additional interventions planned for labor/delivery related to patient's disorder for seventh pregnancy

- Unknown                       None
- Additional maternal lab monitoring       Altered anesthesia plan
- Change in delivery site                       IV fluids
- Letter to OB/MFM specialist                       Planned C-section
- Referral for high risk OB management       Other

Additional interventions planned for labor/delivery related to patient's disorder for seventh pregnancy-other, specify

Actual interventions for labor/delivery related to patient's disorder for seventh pregnancy

- Unknown                       No
- Letter to OB/MFM specialist                       IV fluids
- Planned C-section                       Additional maternal lab monitoring
- Change in delivery site                       Referral for high risk OB management
- Altered anesthesia plan                       Other

Actual interventions for labor/delivery related to patient's disorder for seventh pregnancy-other, specify

Patient Name \_\_\_\_\_

Date

Acute health concerns experienced by the patient during delivery for seventh delivery

Abnormal patient lab results during seventh delivery

- Unknown                       None                       Normal labs during delivery
- Elevated CK                       Elevated liver function tests    Hyperammonemia
- Hypoglycemia                       Metabolic decompensation    Other

Abnormal patient lab results during seventh delivery-other, specify

Additional maternal interventions during or after seventh delivery related to this disorder

- Unknown                       Ammonul                       Blood/blood product transfusion
- Dialysis                       ICU monitoring                       Infusions
- Medications                       Resuscitation                       TPN
- Other

Additional maternal interventions during or after seventh delivery related to this disorder-other, specify

Patient death during or shortly after seventh delivery    Unknown    Yes    No

Disorder contributed to death of mother during seventh delivery    Unknown    Yes    No

Weeks gestation at time of seventh delivery - round to nearest week\_\_\_\_\_

Method of seventh delivery

- Unknown                       Caesarean section (scheduled or non-urgent)
- Casesarean section (emergent)                       Vaginal

Maternal inpatient days post- seventh delivery\_\_\_\_\_

Live delivery of seventh newborn    Unknown    Yes    No

Acute health concerns for seventh newborn related to maternal disorder (example: fetal distress secondary to maternal acute metabolic decompensation)

Patient Name \_\_\_\_\_

Date

Additional lab tests and results done on seventh newborn specifically due to maternal disorder history

Health concerns for seventh newborn not known to be related to disorder

Additional interventions for seventh newborn during or shortly after delivery (other than labs)

- Unknown  Blood/blood product transfusion
- Glucose infusion  Medications
- NICU/special care nursery monitoring  Resuscitation
- TPN  Other

Additional interventions for seventh newborn during or shortly after delivery-other, specify

Birth measurements for seventh newborn  Unknown  Head circumference  Length  Weight

Birth head circumference \_\_\_\_\_

Birth head circumference units  cm  in

Birth length \_\_\_\_\_

Birth length units  cm  in

Birth weight \_\_\_\_\_

Birth weight units  lbs  kg  gm  oz

APGAR score at 1 minute for seventh newborn

- Unknown
- 0
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10

Health concern(s) with APGAR score at one minute < 8 for seventh newborn

APGAR score at 5 minutes for seventh newborn

- Unknown
- 0
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10

Patient Name \_\_\_\_\_

Date      

Health concern(s) with APGAR score at five minutes &lt; 8 for seventh newborn

Length of seventh newborn's stay in the hospital after birth

- Unknown  <24 hours  
 24-28 hours  3-5 days  
 6-14 days  >14 days  
 N/A - baby was not born in a hospital  baby is still in the hospital

Routine newborn screening done for seventh newborn

- Unknown  Yes - normal for all screened disorders  
 Yes - abnormal  Yes - results pending  
 No

Describe abnormal newborn screen result for seventh newborn

Reason routine newborn screening was not done for seventh newborn

- Unknown  
 Died prior to collection of NBS  
 Refused  
 Transferred to another facility prior to NBS collection  
 Transfused prior to collection of NBS  
 Other

Reason routine newborn screening was not done for seventh newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the seventh baby after birth to rule out the mother's disorder

- Unknown  Yes - biochemical normal  
 Yes - biochemical abnormal  Yes - molecular normal  
 Yes - molecular abnormal affected  Yes - molecular abnormal unaffected carrier  
 No

Patient Name \_\_\_\_\_

Date      

Additional studies and results (examples: brain MRI, echocardiogram) done on seventh newborn to assess for effects of maternal disorder

Seventh newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

Unknown  Yes  No

Abnormal newborn exam findings for seventh newborn

Unknown  None  Congenital heart disease  
 Dysmorphism  Lethargy  Microcephaly  
 Other congenital anomalies  Poor feeding  Respiratory distress  
 Seizure(s)  Small for gestational age  Other

Abnormal newborn exam findings for seventh newborn-other, specify \_\_\_\_\_

Newborn death at or shortly after delivery for seventh newborn  Unknown  Yes  No

Newborn's death related to maternal disorder for seventh newborn  Unknown  Yes  No

Seventh newborn currently alive  Unknown  Yes  No

## **Eighth Pregnancy**

---

Patient's age at time of eighth pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before eighth pregnancy (in months)

Unknown  Unplanned  1  2  3  4  5  
 6  7  8  9  10  11  12  
 13-24  25-36  >36

History of infertility for eighth pregnancy

Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for eighth pregnancy

Unknown  Male factor  Female factor  
 Combined male-female factor

Cause(s) of male infertility for eighth pregnancy

Unknown  Abnormal sperm production or function  
 Age  Cancer/cancer treatment related  
 Environmental  Problems with sperm delivery

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

- Tobacco use
- Other

Cause(s) of male infertility for eighth pregnancy-other, specify \_\_\_\_\_

Cause(s) of female infertility for eighth pregnancy

- Unknown
- Age
- Alcohol use
- Cancer/cancer treatment related
- Early menopause
- Fallopian tube damage/blockage
- Hyperprolactinemia
- Pelvic adhesions
- Polycystic ovary syndrome (PCOS)
- Premature ovarian insufficiency
- Over-exercise
- Ovulation disorder
- Thyroid problems
- Tobacco use
- Uterine fibroid
- Weight related
- Other

Cause(s) of female infertility for eighth pregnancy-other, specify \_\_\_\_\_

Cause(s) of infertility for eighth pregnancy, combined male-female factor- specify \_\_\_\_\_

Types of fertility treatment(s) received prior to eighth pregnancy

- Unknown
- None
- Assisted reproductive technology
- Fertility drugs
- Surgery

Type of assisted reproductive technologies used during eighth pregnancy

- Assisted hatching
- ICSI
- IVF
- Surgical sperm aspiration
- Other

Type of assisted reproductive technologies used during eighth pregnancy-other, specify \_\_\_\_\_

Partner tested for patient's disorder during eighth pregnancy

- Unknown
- Yes-biochemical
- Yes-molecular
- No

Partner also affected by disorder during eighth pregnancy  Unknown  Yes  No

Preimplantation genetic diagnosis for disorder done for eighth pregnancy  Unknown  Yes  No

Prenatal testing done for fetus for this disorder for eighth pregnancy  Unknown  Yes  No

Type of prenatal testing performed for eighth pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for eighth pregnancy

- Unknown
- Amniocentesis
- Chorionic villus sampling

Additional prenatal testing performed on fetus as a result of parent's disorder for eighth pregnancy

- Unknown
- None
- Fetal echocardiogram
- Increased frequency prenatal ultrasounds

Patient Name \_\_\_\_\_

Date

Other

Additional prenatal testing performed on fetus as a result of parent's disorder for eighth pregnancy-other, specify \_\_\_\_\_

Eighth pregnancy terminated  Unknown  Yes  No

Reason eighth pregnancy terminated

- Elective due to fetus affected with disorder
- Elective due to other fetal well-being unrelated to disorder
- Elective for other reason
- Elective due to maternal well-being
- Spontaneous

Gestational age (in weeks) at time of eighth pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for eighth pregnancy \_\_\_\_\_

Prenatal care received during eighth pregnancy  Unknown  Yes  No

Weeks gestation prenatal care started during eighth pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during eighth pregnancy

- Unknown  Metabolic diet  Medications
- Biochemical lab monitoring  Avoidance of fasting  Other

Treatment prescribed for patient's disorder during eighth pregnancy-other, specify

Patient in good metabolic condition prior to eighth pregnancy  Unknown  Yes  No

Patient in good metabolic condition during first trimester of eighth pregnancy  Unknown  Yes  No

Patient in good metabolic condition during second trimester of eighth pregnancy

- Unknown  Pregnancy not sustained to second trimester
- Yes  No

Patient in good metabolic condition during third trimester of eighth pregnancy

- Unknown  Pregnancy not sustained to third trimester
- Yes  No

Number of outpatient metabolic visits for patient during eighth pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during eighth pregnancy

- Unknown  0  1  2  3  4  5
- 6  7  8  9  10  >10

Patient Name \_\_\_\_\_

Date |M|M| |D|D| |Y|Y|

Number of hospitalizations for management of disorder during eighth pregnancy

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10       >10

Complications during eighth pregnancy

- Unknown                                       Yes-related to disorder  
 Yes- not known to be related to disorder    No

Complications related to disorder during eighth pregnancy- specify

- Unknown  
 Acute fatty liver of pregnancy (AFLP)  
 Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)  
 Intrauterine growth restriction (AUGR)  
 Mother affected with this condition  
 Other

Complications not known to be related to disorder during eighth pregnancy-specify

- Unknown  
 Advanced maternal age (35+ years of age)  
 Ectopic pregnancy  
 Gestational diabetes  
 Group B strep  
 Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)  
 Intrauterine growth restriction (AUGR)  
 Inadequate prenatal care  
 Maternal prenatal substance exposure  
 Preeclampsia  
 Rh isoimmunization  
 Toxemia  
 Young maternal age (15 years of age + under)  
 Preterm labor  
 Other

Complications during eighth pregnancy-other, specify \_\_\_\_\_

Number of ED visits for complications during eighth pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10       >10

Number of hospitalizations for complications during eighth pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5



Patient Name \_\_\_\_\_

Date      

6     
  7     
  8     
  9     
  10     
  >10

Duration of longest inpatient hospitalization (in days) for any reason during eighth pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during eighth pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during eighth pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during eighth pregnancy

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> Ammonia - Abn high                  |
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> CO <sub>2</sub> - Abn low           | <input type="checkbox"/> CO <sub>2</sub> - WNL               |
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

Patient Name \_\_\_\_\_

Date      

Other laboratory studies done on patient during eighth pregnancy: describe test(s) and result(s)

Additional interventions required during eighth pregnancy due to this metabolic condition

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                                    | <input type="checkbox"/> ED visits for hyperemesis/IV fluids   |
| <input type="checkbox"/> Hospitalizations for hyperemesis/IV fluids | <input type="checkbox"/> TPN                                   |
| <input type="checkbox"/> Tube feedings                              | <input type="checkbox"/> Additional medications                |
| <input type="checkbox"/> Home lab monitoring                        | <input type="checkbox"/> Increased frequency of lab monitoring |
| <input type="checkbox"/> More intensive fetal monitoring            | <input type="checkbox"/> Bedrest                               |
| <input type="checkbox"/> Other                                      |  |

Additional interventions required during eighth pregnancy due to this metabolic condition-other, specify

\_\_\_\_\_

Total maternal weight gain (in kg) during eighth pregnancy \_\_\_\_\_

Eighth pregnancy complicated by maternal HELLP syndrome  Unknown  Yes  NoEighth pregnancy complicated by acute fatty liver of pregnancy (AFLP)  Unknown  Yes  No

Number of documented episodes of hypoglycemia during eighth pregnancy

- Unknown  0  1  2  3  4  5  
 6  7  8  9  10  >10

Lowest maternal blood glucose value documented during eighth pregnancy \_\_\_\_\_

Lowest maternal blood glucose value documented during eighth pregnancy units  mmol/L  mg/dL

Number of documented episodes of rhabdomyolysis during eighth pregnancy

- Unknown  0  1  2  3  4  5  
 6  7  8  9  10  >10

Highest maternal CK value documented during eighth pregnancy \_\_\_\_\_

Highest maternal CK value documented during eighth pregnancy units  U/L  umol/L  mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during eighth pregnancy

- Unknown  1  2  3  4  5  6  
 7  8  9  10  11  12

Longest permitted fasting interval (in hours) during the NIGHT recommended during eighth pregnancy

- Unknown  1  2  3  4  5  6  
 7  8  9  10  11  13

L-carnitine supplementation used on a regular basis during eighth pregnancy

- Unknown  Yes  No  
 L-carnitine not prescribed

Patient Name \_\_\_\_\_

Date

Patient adhered to a fat-restricted diet during eighth pregnancy

- Unknown                       Yes                       No
- Fat-restriction not prescribed

Patient took a metabolic formula regularly during eighth pregnancy

- Unknown                       Yes
- No                       Metabolic formula not prescribed

Evidence of cardiac hypertrophy during eighth pregnancy

- Unknown                       Yes
- No                       Echocardiogram not performed during pregnancy

Evidence of any cardiac dysfunction during eighth pregnancy

- Unknown                       Yes
- No                       Cardiac testing not performed during pregnancy

Additional interventions planned for labor/delivery related to patient's disorder for eighth pregnancy

- Unknown                       None
- Additional maternal lab monitoring       Altered anesthesia plan
- Change in delivery site                       IV fluids
- Letter to OB/MFM specialist               Planned C-section
- Referral for high risk OB management    Other

Additional interventions planned for labor/delivery related to patient's disorder for eighth pregnancy-other, specify

Actual interventions for labor/delivery related to patient's disorder for eighth pregnancy

- Unknown                       No
- Letter to OB/MFM specialist               IV fluids
- Planned C-section                       Additional maternal lab monitoring
- Change in delivery site                       Referral for high risk OB management
- Altered anesthesia plan                       Other

Actual interventions for labor/delivery related to patient's disorder for eighth pregnancy-other, specify

Patient Name _____
Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>

Acute health concerns experienced by the patient during delivery for eighth delivery

Abnormal patient lab results during eighth delivery

- |                                       |  |  |
|---------------------------------------|--|--|
| <input type="checkbox"/> Unknown      | <input type="checkbox"/> None                          | <input type="checkbox"/> Normal labs during delivery |
| <input type="checkbox"/> Elevated CK  | <input type="checkbox"/> Elevated liver function tests | <input type="checkbox"/> Hyperammonemia              |
| <input type="checkbox"/> Hypoglycemia | <input type="checkbox"/> Metabolic decompensation      | <input type="checkbox"/> Other                       |

Abnormal patient lab results during eighth delivery-other, specify

Additional maternal interventions during or after eighth delivery related to this disorder

- |                                      |   |  |
|--------------------------------------|---|--|
| <input type="checkbox"/> Unknown     | <input type="checkbox"/> Ammonul        | <input type="checkbox"/> Blood/blood product transfusion |
| <input type="checkbox"/> Dialysis    | <input type="checkbox"/> ICU monitoring | <input type="checkbox"/> Infusions                       |
| <input type="checkbox"/> Medications | <input type="checkbox"/> Resuscitation  | <input type="checkbox"/> TPN                             |
| <input type="checkbox"/> Other       |   |  |

Additional maternal interventions during or after eighth delivery related to this disorder-other, specify

Patient death during or shortly after eighth delivery     Unknown     Yes     No

Disorder contributed to death of mother during eighth delivery     Unknown     Yes     No

Weeks gestation at time of eighth delivery - round to nearest week\_\_\_\_\_

Method of eighth delivery

- |   |   |
|---|---|
| <input type="radio"/> Unknown                       | <input type="radio"/> Caesarean section (scheduled or non-urgent) |
| <input type="radio"/> Casesarean section (emergent) | <input type="radio"/> Vaginal                                     |

Maternal inpatient days post- eighth delivery\_\_\_\_\_

Live delivery of eighth newborn     Unknown     Yes     No

Acute health concerns for eighth newborn related to maternal disorder (example: fetal distress secondary to maternal acute metabolic decompensation)

Patient Name \_\_\_\_\_

Date

Additional lab tests and results done on eighth newborn specifically due to maternal disorder history

Health concerns for eighth newborn not known to be related to disorder

Additional interventions for eighth newborn during or shortly after delivery (other than labs)

- Unknown  Blood/blood product transfusion
- Glucose infusion  Medications
- NICU/special care nursery monitoring  Resuscitation
- TPN  Other

Additional interventions for eighth newborn during or shortly after delivery-other, specify

Birth measurements for eighth newborn  Unknown  Head circumference  Length  Weight

Birth head circumference \_\_\_\_\_

Birth head circumference units  cm  in

Birth length \_\_\_\_\_

Birth length units  cm  in

Birth weight \_\_\_\_\_

Birth weight units  lbs  kg  gm  oz

APGAR score at 1 minute for eighth newborn

- Unknown
- 0
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10

Health concern(s) with APGAR score at one minute < 8 for eighth newborn

APGAR score at 5 minutes for eighth newborn

- Unknown
- 0
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10

Patient Name \_\_\_\_\_

Date      

Health concern(s) with APGAR score at five minutes &lt; 8 for eighth newborn

Length of eighth newborn's stay in the hospital after birth

- Unknown  <24 hours  
 24-28 hours  3-5 days  
 6-14 days  >14 days  
 N/A - baby was not born in a hospital  baby is still in the hospital

Routine newborn screening done for eighth newborn

- Unknown  Yes - normal for all screened disorders  
 Yes - abnormal  Yes - results pending  
 No

Describe abnormal newborn screen result for eighth newborn

Reason routine newborn screening was not done for eighth newborn

- Unknown  
 Died prior to collection of NBS  
 Refused  
 Transferred to another facility prior to NBS collection  
 Transfused prior to collection of NBS  
 Other

Reason routine newborn screening was not done for eighth newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the eighth baby after birth to rule out the mother's disorder

- Unknown  Yes - biochemical normal  
 Yes - biochemical abnormal  Yes - molecular normal  
 Yes - molecular abnormal affected  Yes - molecular abnormal unaffected carrier  
 No

Patient Name \_\_\_\_\_

Date      

Additional studies and results (examples: brain MRI, echocardiogram) done on eighth newborn to assess for effects of maternal disorder

Eighth newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

Unknown  Yes  No

Abnormal newborn exam findings for eighth newborn

- |   |  |   |
|---|--|---|
| <input type="checkbox"/> Unknown                    | <input type="checkbox"/> None                      | <input type="checkbox"/> Congenital heart disease |
| <input type="checkbox"/> Dysmorphism                | <input type="checkbox"/> Lethargy                  | <input type="checkbox"/> Microcephaly             |
| <input type="checkbox"/> Other congenital anomalies | <input type="checkbox"/> Poor feeding              | <input type="checkbox"/> Respiratory distress     |
| <input type="checkbox"/> Seizure(s)                 | <input type="checkbox"/> Small for gestational age | <input type="checkbox"/> Other                    |

Abnormal newborn exam findings for eighth newborn-other, specify \_\_\_\_\_

Newborn death at or shortly after delivery for eighth newborn  Unknown  Yes  No

Newborn's death related to maternal disorder for eighth newborn  Unknown  Yes  No

Eighth newborn currently alive  Unknown  Yes  No

## Current Pregnancy

---

Patient is pregnant  Unknown  Yes  No

Patient's age at time of current pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before current pregnancy (in months)

- Unknown  Unplanned  1  2  3  4  5  
 6  7  8  9  10  11  12  
 13-24  25-36  >36

History of infertility for current pregnancy

- Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for current pregnancy

- Unknown  Male factor  Female factor  
 Combined male-female factor

Cause(s) of male infertility for current pregnancy

- Unknown  Abnormal sperm production or function  
 Age  Cancer/cancer treatment related

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

- Environmental
- Tobacco use
- Problems with sperm delivery
- Other

Cause(s) of male infertility for current pregnancy-other, specify \_\_\_\_\_

Cause(s) of female infertility for current pregnancy

- Unknown
- Alcohol use
- Early menopause
- Hyperprolactinemia
- Polycystic ovary syndrome (PCOS)
- Over-exercise
- Thyroid problems
- Uterine fibroid
- Other
- Age
- Cancer/cancer treatment related
- Fallopian tube damage/blockage
- Pelvic adhesions
- Premature ovarian insufficiency
- Ovulation disorder
- Tobacco use
- Weight related

Cause(s) of female infertility for current pregnancy-other, specify \_\_\_\_\_

Cause(s) of infertility for current pregnancy, combined male-female factor- specify

\_\_\_\_\_

Types of fertility treatment(s) received prior to current pregnancy

- Unknown
- Assisted reproductive technology
- Surgery
- None
- Fertility drugs

Type of assisted reproductive technologies used during current pregnancy

- Assisted hatching
- Surgical sperm aspiration
- ICSI
- Other
- IVF

Type of assisted reproductive technologies used during current pregnancy-other, specify

\_\_\_\_\_

Partner tested for patient's disorder during current pregnancy

- Unknown
- Yes-biochemical
- Yes-molecular
- No

Partner also affected by disorder during current pregnancy  Unknown  Yes  No

Preimplantation genetic diagnosis for disorder done for current pregnancy  Unknown  Yes  No

Prenatal testing done for fetus for this disorder for current pregnancy  Unknown  Yes  No

Type of prenatal testing performed for current pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for current pregnancy

- Unknown
- Amniocentesis
- Chorionic villus sampling

Additional prenatal testing performed on fetus as a result of parent's disorder for current pregnancy

- Unknown
- None



Patient Name \_\_\_\_\_

Date

Fetal echocardiogram

Increased frequency prenatal ultrasounds

Other

Additional prenatal testing performed on fetus as a result of parent's disorder for current pregnancy-other, specify \_\_\_\_\_

Current pregnancy terminated  Unknown  Yes  No

Reason current pregnancy terminated

- Elective due to fetus affected with disorder
- Elective due to other fetal well-being unrelated to disorder
- Elective for other reason
- Elective due to maternal well-being
- Spontaneous

Gestational age (in weeks) at time of current pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for current pregnancy \_\_\_\_\_

Prenatal care received during current pregnancy  Unknown  Yes  No

Weeks gestation prenatal care started during current pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during current pregnancy

- Unknown  Metabolic diet  Medications
- Biochemical lab monitoring  Avoidance of fasting  Other

Treatment prescribed for patient's disorder during current pregnancy-other, specify

Patient in good metabolic condition prior to current pregnancy  Unknown  Yes  No

Patient in good metabolic condition during first trimester of current pregnancy

- Unknown  Yes  No

Patient in good metabolic condition during second trimester of current pregnancy

- Unknown  Pregnancy not sustained to second trimester
- Yes  No

Patient in good metabolic condition during third trimester of current pregnancy

- Unknown  Pregnancy not sustained to third trimester
- Yes  No

Number of outpatient metabolic visits for patient during current pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during current pregnancy \_\_\_\_\_

Patient Name \_\_\_\_\_

Date |M|M| |D|D| |Y|Y|

- Unknown  0  1  2  3  4  5  
 6  7  8  9  10  >10

Number of hospitalizations for management of disorder during current pregnancy

- Unknown  0  1  2  3  4  5  
 6  7  8  9  10  >10

Complications during current pregnancy

- Unknown  Yes-related to disorder  
 Yes- not known to be related to disorder  No

Complications related to disorder during current pregnancy- specify

- Unknown  
 Acute fatty liver of pregnancy (AFLP)  
 Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)  
 Intrauterine growth restriction (AUGR)  
 Mother affected with this condition  
 Other

Complications not known to be related to disorder during current pregnancy-specify

- Unknown  
 Advanced maternal age (35+ years of age)  
 Ectopic pregnancy  
 Gestational diabetes  
 Group B strep  
 Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)  
 Intrauterine growth restriction (AUGR)  
 Inadequate prenatal care  
 Maternal prenatal substance exposure  
 Preeclampsia  
 Rh isoimmunization  
 Toxemia  
 Young maternal age (15 years of age + under)  
 Preterm labor  
 Other

Complications during current pregnancy-other, specify \_\_\_\_\_

Number of ED visits for complications during current pregnancy, unrelated to management of disorder

- Unknown  0  1  2  3  4  5  
 6  7  8  9  10  >10

Patient Name \_\_\_\_\_

Date      

Number of hospitalizations for complications during current pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10       >10

Duration of longest inpatient hospitalization (in days) for any reason during current pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during current pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during current pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during current pregnancy

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> Ammonia - Abn high                  |
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> CO2 - Abn low                       | <input type="checkbox"/> CO2 - WNL                           |
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |

Patient Name \_\_\_\_\_

Date    |    |    Other

Other laboratory studies done on patient during current pregnancy: describe test(s) and result(s)

Additional interventions required during current pregnancy due to this metabolic condition

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                                    | <input type="checkbox"/> ED visits for hyperemesis/IV fluids   |
| <input type="checkbox"/> Hospitalizations for hyperemesis/IV fluids | <input type="checkbox"/> TPN                                   |
| <input type="checkbox"/> Tube feedings                              | <input type="checkbox"/> Additional medications                |
| <input type="checkbox"/> Home lab monitoring                        | <input type="checkbox"/> Increased frequency of lab monitoring |
| <input type="checkbox"/> More intensive fetal monitoring            | <input type="checkbox"/> Bedrest                               |
| <input type="checkbox"/> Other                                      |  |

Additional interventions required during current pregnancy due to this metabolic condition-other, specify

\_\_\_\_\_

Total maternal weight gain to date (in kg) during current pregnancy \_\_\_\_\_

Current pregnancy complicated by maternal HELLP syndrome  Unknown  Yes  NoCurrent pregnancy complicated by acute fatty liver of pregnancy (AFLP)  Unknown  Yes  No

Number of documented episodes of hypoglycemia during current pregnancy

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10       >10

Lowest maternal blood glucose value documented during current pregnancy \_\_\_\_\_

Lowest maternal blood glucose value documented during current pregnancy units  mmol/L  mg/dL

Number of documented episodes of rhabdomyolysis during current pregnancy

- Unknown  0       1       2       3       4       5  
 6       7       8       9       10       >10

Highest maternal CK value documented during current pregnancy \_\_\_\_\_

Highest maternal CK value documented during current pregnancy units  U/L  umol/L  mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during current pregnancy

- Unknown  1       2       3       4       5       6  
 7       8       9       10       11       12

Longest permitted fasting interval (in hours) during the NIGHT recommended during current pregnancy

- Unknown  1       2       3       4       5       6  
 7       8       9       10       11       13

L-carnitine supplementation used on a regular basis during current pregnancy

- Unknown       Yes       No

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

L-carnitine not prescribed

Patient adhered to a fat-restricted diet during current pregnancy

Unknown                       Yes                       No

Fat-restriction not prescribed

Patient took a metabolic formula regularly during current pregnancy

Unknown                       Yes  
 No                       Metabolic formula not prescribed

Evidence of cardiac hypertrophy during current pregnancy

Unknown                       Yes  
 No                       Echocardiogram not performed during pregnancy

Evidence of any cardiac dysfunction during current pregnancy

Unknown                       Yes  
 No                       Cardiac testing not performed during pregnancy

Additional interventions planned for labor/delivery related to patient's disorder for current pregnancy

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                              | <input type="checkbox"/> None                    |
| <input type="checkbox"/> Additional maternal lab monitoring   | <input type="checkbox"/> Altered anesthesia plan |
| <input type="checkbox"/> Change in delivery site              | <input type="checkbox"/> IV fluids               |
| <input type="checkbox"/> Letter to OB/MFM specialist          | <input type="checkbox"/> Planned C-section       |
| <input type="checkbox"/> Referral for high risk OB management | <input type="checkbox"/> Other                   |

Additional interventions planned for labor/delivery related to patient's disorder for current pregnancy-other, specify

**Comments**

---

Pregnancy comments

Patient Name \_\_\_\_\_

Date      **CACT - Dialysis**

Number of different episodes during which dialysis (any type) was used

 1    2    3    4    5    6    7    8    9    10    >10**First Dialysis Treatment**

Type(s) of dialysis received during first episode

- Unknown       APD/CCPD       ECMO       CAPD  
 CVVH       CVVD       CVVHDF       Hemodialysis  
 Peritoneal dialysis

Reason for first episode of dialysis

- Unknown       Hyperammonemia    MSUD       Organ failure       Sepsis  
 Other

Reason for first episode of dialysis-other, specify

Start date of first episode of dialysis treatment      

Duration (in days) of first episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during first episode of dialysis

- Unknown       None       Ammonia       Blood urea nitrogen  
 Plasma alloisoleucine    Plasma isoleucine       Plasma leucine       Plasma valine  
 Serum creatinine       Other

Metabolite(s) of concern during first episode of dialysis-other,specify \_\_\_\_\_

Peak value of ammonia during first episode of dialysis \_\_\_\_\_

Peak value of ammonia during first episode of dialysis units

 umol/L       ug/dL       ug/L       ug/mL       g/dL

Peak value of blood urea nitrogen during first episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during first episode of dialysis units    mg/dL    mmol/L

Peak value of plasma alloisoleucine during first episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during first episode of dialysis units

 umol/dL       umol/L       mg/dL

Peak value of plasma isoleucine during first episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during first episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of plasma leucine during first episode of dialysis \_\_\_\_\_

Patient Name _____
Date <input type="text"/> <input type="text"/> <input type="text"/>   <input type="text"/> <input type="text"/> <input type="text"/>   <input type="text"/> <input type="text"/> <input type="text"/>

Peak value of plasma leucine during first episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of plasma valine during first episode of dialysis\_\_\_\_\_

Peak value of plasma valine during first episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of serum creatinine during first episode of dialysis\_\_\_\_\_

Peak value of serum creatinine during first episode of dialysis units  umol/L  mg/dL

Peak value of other metabolite during first episode of dialysis\_\_\_\_\_

Peak value of other metabolite during first episode of dialysis units\_\_\_\_\_

Reason first episode of dialysis treatment was stopped

- Unknown  Acute episode resolved  Treatment withdrawn  Death
- Other

Reason first episode of dialysis treatment was stopped-other, specify\_\_\_\_\_

### Second Dialysis Treatment

---

Type(s) of dialysis received during second episode

- Unknown  APD/CCPD  ECMO  CAPD
- CVVH  CVVD  CVVHDF  Hemodialysis
- Peritoneal dialysis

Reason for second episode of dialysis

- Unknown  Hyperammonemia  MSUD  Organ failure  Sepsis
- Other

Reason for second episode of dialysis-other, specify

Start date of second episode of dialysis treatment    |    |

Duration (in days) of second episode of dialysis treatment\_\_\_\_\_

Metabolite(s) of concern during second episode of dialysis

- Unknown  None  Ammonia  Blood urea nitrogen
- Plasma allosioleucine  Plasma isoleucine  Plasma leucine  Plasma valine
- Serum creatinine  Other

Metabolite(s) of concern during second episode of dialysis- other, specify\_\_\_\_\_

Peak value of ammonia during second episode of dialysis\_\_\_\_\_

Peak value of ammonia during second episode of dialysis units

- umol/L  ug/dL  ug/L  ug/mL  g/dL

Peak value of blood urea nitrogen during second episode of dialysis\_\_\_\_\_

Peak value of blood urea nitrogen during second episode of dialysis units  mg/dL  mmol/L

Patient Name \_\_\_\_\_

Date      

Peak value of plasma alloisoleucine during second episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during second episode of dialysis units

 umol/dL umol/L mg/dL

Peak value of plasma isoleucine during second episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during second episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of plasma leucine during second episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during second episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of plasma valine during second episode of dialysis \_\_\_\_\_

Peak value of plasma valine during second episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of serum creatinine during second episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during second episode of dialysis units  umol/L  mg/dL

Peak value of other metabolite during second episode of dialysis \_\_\_\_\_

Peak value of other metabolite during second episode of dialysis units \_\_\_\_\_

Reason second episode of dialysis treatment was stopped

 Unknown Acute episode resolved  Treatment withdrawn  Death Other

Reason second episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

**Third Dialysis Treatment**

Type(s) of dialysis received during third episode

 Unknown APD/CCPD ECMO CAPD CVVH CVVD CVVHDF Hemodialysis Peritoneal dialysis

Reason for third episode of dialysis

 Unknown Hyperammonemia  MSUD Organ failure Sepsis Other

Reason for third episode of dialysis-other, specify

Start date of third episode of dialysis treatment      

Duration (in days) of third episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during third episode of dialysis

 Unknown None Ammonia Blood urea nitrogen Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine Serum creatinine Other



Patient Name \_\_\_\_\_

Date      

Metabolite(s) of concern during third episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during third episode of dialysis \_\_\_\_\_

Peak value of ammonia during third episode of dialysis units

 umol/L       ug/dL       ug/L       ug/mL       g/dL

Peak value of blood urea nitrogen during third episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during third episode of dialysis units     mg/dL     mmol/L

Peak value of plasma alloisoleucine during third episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during third episode of dialysis units

 umol/dL                       umol/L                       mg/dL

Peak value of plasma isoleucine during third episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during third episode of dialysis units     umol/dL     umol/L     mg/dL

Peak value of plasma leucine during third episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during third episode of dialysis units     umol/dL     umol/L     mg/dL

Peak value of plasma valine during third episode of dialysis \_\_\_\_\_

Peak value of plasma valine during third episode of dialysis units     umol/dL     umol/L     mg/dL

Peak value of serum creatinine during third episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during third episode of dialysis units     umol/L     mg/dL

Peak value of other metabolite during third episode of dialysis \_\_\_\_\_

Peak value of other metabolite during third episode of dialysis units \_\_\_\_\_

Reason third episode of dialysis treatment was stopped

 Unknown                       Acute episode resolved     Treatment withdrawn     Death
 Other

Reason third episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

**Fourth Dialysis Treatment**

Type(s) of dialysis received during fourth episode

 Unknown                       APD/CCPD                       ECMO                       CAPD  
 CVVH                       CVVD                       CVVHDF                       Hemodialysis  
 Peritoneal dialysis

Reason for fourth episode of dialysis

 Unknown                       Hyperammonemia     MSUD                       Organ failure                       Sepsis
 Other

Reason for fourth episode of dialysis-other, specify

Patient Name \_\_\_\_\_

Date      Start date of fourth episode of dialysis treatment      

Duration (in days) of fourth episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during fourth episode of dialysis

- Unknown                       None                       Ammonia                       Blood urea nitrogen  
 Plasma alloisoleucine    Plasma isoleucine         Plasma leucine               Plasma valine  
 Serum creatinine         Other

Metabolite(s) of concern during fourth episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during fourth episode of dialysis \_\_\_\_\_

Peak value of ammonia during fourth episode of dialysis units

- umol/L                       ug/dL                       ug/L                       ug/mL                       g/dL

Peak value of blood urea nitrogen during fourth episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during fourth episode of dialysis units    mg/dL    mmol/L

Peak value of plasma alloisoleucine during fourth episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during fourth episode of dialysis units

- umol/dL                       umol/L                       mg/dL

Peak value of plasma isoleucine during fourth episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during fourth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of plasma leucine during fourth episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during fourth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of plasma valine during fourth episode of dialysis \_\_\_\_\_

Peak value of plasma valine during fourth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of serum creatinine during fourth episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during fourth episode of dialysis units    umol/L    mg/dL

Peak value of other metabolite during fourth episode of dialysis \_\_\_\_\_

Peak value of other metabolite during fourth episode of dialysis units \_\_\_\_\_

Reason fourth episode of dialysis treatment was stopped

- Unknown                       Acute episode resolved    Treatment withdrawn    Death  
 Other

Reason fourth episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

## Fifth Dialysis Treatment

---

Type(s) of dialysis received during fifth episode

- Unknown                       APD/CCPD                       ECMO                       CAPD  
 CVVH                       CVVD                       CVVHDF                       Hemodialysis  
 Peritoneal dialysis

Reason for fifth episode of dialysis

Patient Name \_\_\_\_\_

Date      

- Unknown     
  Hyperammonemia   
  MSUD                     
  Organ failure       
  Sepsis  
 Other

Reason for fifth episode of dialysis-other, specify

Start date of fifth episode of dialysis treatment      

Duration (in days) of fifth episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during fifth episode of dialysis

- Unknown             
  None                     
  Ammonia                     
  Blood urea nitrogen  
 Plasma alloisoleucine  
  Plasma isoleucine     
  Plasma leucine             
  Plasma valine  
 Serum creatinine     
  Other

Metabolite(s) of concern during fifth episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during fifth episode of dialysis \_\_\_\_\_

Peak value of ammonia during fifth episode of dialysis units

- umol/L             
  ug/dL                     
  ug/L                     
  ug/mL                     
  g/dL

Peak value of blood urea nitrogen during fifth episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during fifth episode of dialysis units   
 mg/dL   
 mmol/L

Peak value of plasma alloisoleucine during fifth episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during fifth episode of dialysis units

- umol/dL                     
  umol/L                     
  mg/dL

Peak value of plasma isoleucine during fifth episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during fifth episode of dialysis units   
 umol/dL   
 umol/L   
 mg/dL

Peak value of plasma leucine during fifth episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during fifth episode of dialysis units   
 umol/dL   
 umol/L   
 mg/dL

Peak value of plasma valine during fifth episode of dialysis \_\_\_\_\_

Peak value of plasma valine during fifth episode of dialysis units   
 umol/dL   
 umol/L   
 mg/dL

Peak value of serum creatinine during fifth episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during fifth episode of dialysis units   
 umol/L   
 mg/dL

Peak value of other metabolite during fifth episode of dialysis \_\_\_\_\_

Peak value of other metabolite during fifth episode of dialysis units \_\_\_\_\_

Reason fifth episode of dialysis treatment was stopped

- Unknown                     
  Acute episode resolved   
 Treatment withdrawn   
 Death  
 Other

Reason fifth episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

Patient Name _____
Date <input type="text"/> <input type="text"/> <input type="text"/>   <input type="text"/> <input type="text"/> <input type="text"/>   <input type="text"/> <input type="text"/> <input type="text"/>

### Sixth Dialysis Treatment

Type(s) of dialysis received during sixth episode

- Unknown                       APD/CCPD                       ECMO                       CAPD
- CVVH                       CVVD                       CVVHDF                       Hemodialysis
- Peritoneal dialysis

Reason for sixth episode of dialysis

- Unknown                       Hyperammonemia     MSUD                       Organ failure                       Sepsis
- Other

Reason for sixth episode of dialysis-other, specify

Start date of sixth episode dialysis treatment    |    |

Duration (in days) of sixth episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during sixth episode of dialysis

- Unknown                       None                       Ammonia                       Blood urea nitrogen
- Plasma alloisoleucine     Plasma isoleucine                       Plasma leucine                       Plasma valine
- Serum creatinine                       Other

Metabolite(s) of concern during sixth episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during sixth episode of dialysis \_\_\_\_\_

Peak value of ammonia during sixth episode of dialysis units

- umol/L                       ug/dL                       ug/L                       ug/mL                       g/dL

Peak value of blood urea nitrogen during sixth episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during sixth episode of dialysis units     mg/dL     mmol/L

Peak value of plasma alloisoleucine during sixth episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during sixth episode of dialysis units

- umol/dL                       umol/L                       mg/dL

Peak value of plasma isoleucine during sixth episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during sixth episode of dialysis units     umol/dL     umol/L     mg/dL

Peak value of plasma leucine during sixth episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during sixth episode of dialysis units     umol/dL     umol/L     mg/dL

Peak value of plasma valine during sixth episode of dialysis \_\_\_\_\_

Peak value of plasma valine during sixth episode of dialysis units     umol/dL     umol/L     mg/dL

Peak value of serum creatinine during sixth episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during sixth episode of dialysis units     umol/L     mg/dL

Patient Name \_\_\_\_\_

Date    |    |

Peak value of other metabolite during sixth episode of dialysis \_\_\_\_\_

Peak value of other metabolite during sixth episode of dialysis units \_\_\_\_\_

Reason sixth episode of dialysis treatment was stopped

- Unknown
- Acute episode resolved
- Treatment withdrawn
- Death
- Other

Reason sixth episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

### Seventh Dialysis Treatment

Type(s) of dialysis received during seventh episode

- Unknown
- APD/CCPD
- ECMO
- CAPD
- CVVH
- CVVD
- CVVHDF
- Hemodialysis
- Peritoneal dialysis

Reason for seventh episode of dialysis

- Unknown
- Hyperammonemia
- MSUD
- Organ failure
- Sepsis
- Other

Reason for seventh episode of dialysis -other, specify

Start date of seventh episode of dialysis treatment    |    |

Duration (in days) of seventh episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during seventh episode of dialysis

- Unknown
- None
- Ammonia
- Blood urea nitrogen
- Plasma alloisoleucine
- Plasma isoleucine
- Plasma leucine
- Plasma valine
- Serum creatinine
- Other

Metabolite(s) of concern during seventh episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during seventh episode of dialysis \_\_\_\_\_

Peak value of ammonia during seventh episode of dialysis units

- umol/L
- ug/dL
- ug/L
- ug/mL
- g/dL

Peak value of blood urea nitrogen during seventh episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during seventh episode of dialysis units  mg/dL  mmol/L

Peak value of plasma alloisoleucine during seventh episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during seventh episode of dialysis units

- umol/dL
- umol/L
- mg/dL

Peak value of plasma isoleucine during seventh episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during seventh episode of dialysis units

Patient Name \_\_\_\_\_

Date       umol/dL umol/L mg/dL

Peak value of plasma leucine during seventh episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during seventh episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of plasma valine during seventh episode of dialysis \_\_\_\_\_

Peak value of plasma valine during seventh episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of serum creatinine during seventh episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during seventh episode of dialysis units  umol/L  mg/dL

Peak value of other metabolite during seventh episode of dialysis \_\_\_\_\_

Peak value of other metabolite during seventh episode of dialysis units \_\_\_\_\_

Reason seventh episode of dialysis treatment was stopped

 Unknown Acute episode resolved Treatment withdrawn Death Other

Reason seventh episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

**Eighth Dialysis Treatment**

Type(s) of dialysis received during eighth episode

 Unknown APD/CCPD ECMO CAPD CVVH CVVD CVVHDF Hemodialysis Peritoneal dialysis

Reason for eighth episode of dialysis

 Unknown Hyperammonemia MSUD Organ failure Sepsis Other

Reason for eighth episode of dialysis-other, specify

Start date of eighth episode of dialysis treatment      

Duration (in days) of eighth episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during eighth episode of dialysis

 Unknown None Ammonia Blood urea nitrogen Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine Serum creatinine Other

Metabolite(s) of concern during eighth episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during eighth episode of dialysis \_\_\_\_\_

Peak value of ammonia during eighth episode of dialysis units

 umol/L ug/dL ug/L ug/mL g/dL

Patient Name _____
Date <input type="text" value="M"/> <input type="text" value="M"/> <input type="text" value="D"/> <input type="text" value="D"/> <input type="text" value="Y"/> <input type="text" value="Y"/>

Peak value of blood urea nitrogen during eighth episode of dialysis\_\_\_\_\_

Peak value of blood urea nitrogen during eighth episode of dialysis units  mg/dL  mmol/L

Peak value of plasma alloisoleucine during eighth episode of dialysis\_\_\_\_\_

Peak value of plasma alloisoleucine during eighth episode of dialysis units

umol/dL  umol/L  mg/dL

Peak value of plasma isoleucine during eighth episode of dialysis\_\_\_\_\_

Peak value of plasma isoleucine during eighth episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of plasma leucine during eighth episode of dialysis\_\_\_\_\_

Peak value of plasma leucine during eighth episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of plasma valine during eighth episode of dialysis\_\_\_\_\_

Peak value of plasma valine during eighth episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of serum creatinine during eighth episode of dialysis\_\_\_\_\_

Peak value of serum creatinine during eighth episode of dialysis units  umol/L  mg/dL

Peak value of other metabolite during eighth episode of dialysis\_\_\_\_\_

Peak value of other metabolite during eighth episode of dialysis units\_\_\_\_\_

Reason eighth episode of dialysis treatment was stopped

- Unknown  Acute episode resolved  Treatment withdrawn  Death
- Other

Reason eighth episode of dialysis treatment was stopped-other, specify\_\_\_\_\_

### Ninth Dialysis Treatment

Type(s) of dialysis received during ninth episode

- Unknown  APD/CCPD  ECMO  CAPD
- CVVH  CVVD  CVVHDF  Hemodialysis
- Peritoneal dialysis

Reason for ninth episode of dialysis

- Unknown  Hyperammonemia  MSUD  Organ failure  Sepsis
- Other

Reason for ninth episode of dialysis-other, specify

Start date of ninth episode of dialysis treatment

Duration (in days) of ninth episode of dialysis treatment\_\_\_\_\_

Metabolite(s) of concern during ninth episode of dialysis

- Unknown  None  Ammonia  Blood urea nitrogen

Patient Name \_\_\_\_\_

Date       Plasma alloisoleucine    Plasma isoleucine    Plasma leucine    Plasma valine Serum creatinine    Other

Metabolite(s) of concern during ninth episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during ninth episode of dialysis \_\_\_\_\_

Peak value of ammonia during ninth episode of dialysis units

 umol/L    ug/dL    ug/L    ug/mL    g/dL

Peak value of blood urea nitrogen during ninth episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during ninth episode of dialysis units    mg/dL    mmol/L

Peak value of plasma alloisoleucine during ninth episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during ninth episode of dialysis units

 umol/dL    umol/L    mg/dL

Peak value of plasma isoleucine during ninth episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during ninth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of plasma leucine during ninth episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during ninth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of plasma valine during ninth episode of dialysis \_\_\_\_\_

Peak value of plasma valine during ninth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of serum creatinine during ninth episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during ninth episode of dialysis units    umol/L    mg/dL

Peak value of other metabolite during ninth episode of dialysis \_\_\_\_\_

Peak value of other metabolite during ninth episode of dialysis units \_\_\_\_\_

Reason ninth episode of dialysis treatment was stopped

 Unknown    Acute episode resolved    Treatment withdrawn    Death Other

Reason ninth episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

**Tenth Dialysis Treatment**

Type(s) of dialysis received during tenth episode

 Unknown    APD/CCPD    ECMO    CAPD CVVH    CVVD    CVVHDF    Hemodialysis Peritoneal dialysis

Reason for tenth episode of dialysis

 Unknown    Hyperammonemia    MSUD    Organ failure    Sepsis Other



Patient Name \_\_\_\_\_

Date

Reason for tenth episode of dialysis-other, specify

Start date of tenth episode of dialysis treatment

Duration (in days) of tenth episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during tenth episode of dialysis

- Unknown                       None                       Ammonia                       Blood urea nitrogen
- Plasma alloisoleucine    Plasma isoleucine         Plasma leucine               Plasma valine
- Serum creatinine         Other

Metabolite(s) of concern during tenth episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during tenth episode of dialysis \_\_\_\_\_

Peak value of ammonia during tenth episode of dialysis units

- umol/L                       ug/dL                       ug/L                       ug/mL                       g/dL

Peak value of blood urea nitrogen during tenth episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during tenth episode of dialysis units    mg/dL    mmol/L

Peak value of plasma alloisoleucine during tenth episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during tenth episode of dialysis units

- umol/dL                       umol/L                       mg/dL

Peak value of plasma isoleucine during tenth episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during tenth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of plasma leucine during tenth episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during tenth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of plasma valine during tenth episode of dialysis \_\_\_\_\_

Peak value of plasma valine during tenth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of serum creatinine during tenth episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during tenth episode of dialysis units    umol/L    mg/dL

Peak value of other metabolite during tenth episode of dialysis \_\_\_\_\_

Peak value of other metabolite during tenth episode of dialysis units \_\_\_\_\_

Reason tenth episode of dialysis treatment was stopped

- Unknown                       Acute episode resolved    Treatment withdrawn     Death
- Other

Reason tenth episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

Patient Name \_\_\_\_\_

Date

If >10 dialysis treatments, for each episode: enter start date, duration (days), and reason for stopping dialysis

**Comments**

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Dialysis comments

Patient Name \_\_\_\_\_

Date      **CACT - Transplant**Number of organ transplants received  1  2  3  4  5  >5**First Transplant**

First organ received by transplant

- Unknown     Bone marrow     Heart                       Kidney                       Liver                       Lung
- Pancreas     Stem Cell     Other

First organ received by transplant, other- specify \_\_\_\_\_

Age (in days) at first transplant \_\_\_\_\_

Reason for first transplant

- Unknown                       Treatment of disorder     Renal failure                       Liver failure
- Heart failure                       Other

Reason for first transplant, other- specify \_\_\_\_\_

Patient followed by Metabolism on an outpatient basis post first transplant  Unknown  Yes  No

Number of outpatient metabolic visits in the last year post first transplant

- 1     2     3     4     5     6     7     8     9     10     >10

Metabolic labs monitored post first transplant  Unknown  Yes  No

Metabolic lab monitoring post first transplant: Note date(s), test(s), and normal or abnormal result(s)

Known complications during the first transplant procedure  Unknown  Yes  No

Known complications during the first transplant procedure- specify

- Clotting                       Death                       Major bleeding                       Other

Known complications during the first transplant procedure, other- specify \_\_\_\_\_

Known complications post first transplant  Unknown  Yes  No

Known complications post first transplant- specify

- |  |   |
|--|---|
| <input type="checkbox"/> Unknown                       | <input type="checkbox"/> Cancer                         |
| <input type="checkbox"/> Cataract                      | <input type="checkbox"/> Clotting                       |
| <input type="checkbox"/> Death                         | <input type="checkbox"/> Delayed graft function         |
| <input type="checkbox"/> Diabetes mellitus             | <input type="checkbox"/> High cholesterol               |
| <input type="checkbox"/> Hypertension                  | <input type="checkbox"/> Infection                      |
| <input type="checkbox"/> Major bleeding                | <input type="checkbox"/> Osteoporosis                   |
| <input type="checkbox"/> Reappearance of heart disease | <input type="checkbox"/> Reappearance of kidney disease |

Patient Name \_\_\_\_\_

Date

- Reappearance of liver disease
- Rejection
- Transplant related renal impairment
- Other

Known complications post first transplant, other- specify \_\_\_\_\_

### Second Transplant

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Second organ received by transplant

- Unknown
- Bone marrow
- Heart
- Kidney
- Liver
- Lung
- Pancreas
- Stem Cell
- Other

Second organ received by transplant, other- specify \_\_\_\_\_

Age (in days) at second transplant \_\_\_\_\_

Reason for second transplant

- Unknown
- Treatment of disorder
- Renal failure
- Liver failure
- Heart failure
- Other

Reason for second transplant, other- specify \_\_\_\_\_

Patient followed by Metabolism on an outpatient basis post second transplant     Unknown     Yes     No

Number of outpatient metabolic visits in the last year post transplant

- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10
- >10

Metabolic labs monitored post second transplant     Unknown     Yes     No

Metabolic lab monitoring post second transplant: Note date(s), test(s), and normal or abnormal result(s)

Complications during the second transplant procedure     Unknown     Yes     No

Complications during the second transplant procedure- specify

- Clotting
- Death
- Major bleeding
- Other

Complications during the second transplant procedure, other- specify \_\_\_\_\_

Complications post second transplant     Unknown     Yes     No

Complications post second transplant- specify

- Unknown
- Cancer
- Cataract
- Clotting
- Death
- Delayed graft function
- Diabetes mellitus
- High cholesterol
- Hypertension
- Infection
- Major bleeding
- Osteoporosis
- Reappearance of heart disease
- Reappearance of kidney disease
- Reappearance of liver disease
- Rejection

Patient Name \_\_\_\_\_

Date       Transplant related renal impairment  Other

Complications post second transplant, other- specify \_\_\_\_\_

**Third Transplant**

Third organ received by transplant

Unknown  Bone marrow  Heart  Kidney  Liver  Lung  
 Pancreas  Stem Cell  Other

Third organ received by transplant, other- specify \_\_\_\_\_

Age (in days) at third transplant \_\_\_\_\_

Reason for third transplant

Unknown  Treatment of disorder  Renal failure  Liver failure  
 Heart failure  Other

Reason for third transplant, other- specify \_\_\_\_\_

Patient followed by Metabolism on an outpatient basis post third transplant  Unknown  Yes  No

Number of outpatient metabolic visits in the last year post transplant

1  2  3  4  5  6  7  8  9  10  >10

Metabolic labs monitored post third transplant  Unknown  Yes  No

Metabolic lab monitoring post third transplant: Note date(s), test(s), and normal or abnormal result(s)

Complications during the third transplant procedure  Unknown  Yes  No

Complications during the third transplant procedure- specify

Clotting  Death  Major bleeding  Other

Complications during the third transplant procedure, other- specify \_\_\_\_\_

Complications post third transplant  Unknown  Yes  No

Complications post third transplant- specify

Unknown  Cancer  
 Cataract  Clotting  
 Death  Delayed graft function  
 Diabetes mellitus  High cholesterol  
 Hypertension  Infection  
 Major bleeding  Osteoporosis  
 Reappearance of heart disease  Reappearance of kidney disease  
 Reappearance of liver disease  Rejection  
 Transplant related renal impairment  Other

Patient Name \_\_\_\_\_

Date      

Complications post third transplant, other- specify \_\_\_\_\_

**Fourth Transplant**

Fourth organ received by transplant

- Unknown     Bone marrow     Heart     Kidney     Liver     Lung  
 Pancreas     Stem Cell     Other

Fourth organ received by transplant, other- specify \_\_\_\_\_

Age (in days) at fourth transplant \_\_\_\_\_

Reason for fourth transplant

- Unknown     Treatment of disorder     Renal failure     Liver failure  
 Heart failure     Other

Reason for fourth transplant, other- specify \_\_\_\_\_

Patient followed by Metabolism on an outpatient basis post fourth transplant     Unknown     Yes     No

Number of outpatient metabolic visits in the last year post transplant

- 1     2     3     4     5     6     7     8     9     10     >10

Metabolic labs monitored post fourth transplant     Unknown     Yes     No

Metabolic lab monitoring post fourth transplant: Note date(s), test(s), and normal or abnormal result(s)

Complications during the fourth transplant procedure     Unknown     Yes     No

Complications during the fourth transplant procedure- specify

- Clotting     Death     Major bleeding     Other

Complications during the fourth transplant procedure, other- specify \_\_\_\_\_

Complications post fourth transplant     Unknown     Yes     No

Complications post fourth transplant- specify

- Unknown     Cancer  
 Cataract     Clotting  
 Death     Delayed graft function  
 Diabetes mellitus     High cholesterol  
 Hypertension     Infection  
 Major bleeding     Osteoporosis  
 Reappearance of heart disease     Reappearance of kidney disease  
 Reappearance of liver disease     Rejection  
 Transplant related renal impairment     Other

Complications post fourth transplant, other- specify \_\_\_\_\_

Patient Name \_\_\_\_\_

Date      **Fifth Transplant**

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Fifth organ received by transplant

- Unknown     Bone marrow     Heart     Kidney     Liver     Lung  
 Pancreas     Stem Cell     Other

Fifth organ received by transplant, other- specify \_\_\_\_\_

Age (in days) at fifth transplant \_\_\_\_\_

Reason for fifth transplant

- Unknown     Treatment of disorder     Renal failure     Liver failure  
 Heart failure     Other

Reason for fifth transplant, other- specify \_\_\_\_\_

Patient followed by Metabolism on an outpatient basis post fifth transplant     Unknown     Yes     No

Number of outpatient metabolic visits in the last year post transplant

- 1     2     3     4     5     6     7     8     9     10     >10

Metabolic labs monitored post fifth transplant     Unknown     Yes     No

Metabolic lab monitoring post fifth transplant: Note date(s), test(s), and normal or abnormal result(s)

Complications during the fifth transplant procedure     Unknown     Yes     No

Complications during the fifth transplant procedure- specify

- Clotting     Death     Major bleeding     Other

Complications during the fifth transplant procedure, other- specify \_\_\_\_\_

Complications post fifth transplant     Unknown     Yes     No

Complications post fifth transplant- specify

- |  |   |
|--|---|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> Cancer                         |
| <input type="checkbox"/> Cataract                            | <input type="checkbox"/> Clotting                       |
| <input type="checkbox"/> Death                               | <input type="checkbox"/> Delayed graft function         |
| <input type="checkbox"/> Diabetes mellitus                   | <input type="checkbox"/> High cholesterol               |
| <input type="checkbox"/> Hypertension                        | <input type="checkbox"/> Infection                      |
| <input type="checkbox"/> Major bleeding                      | <input type="checkbox"/> Osteoporosis                   |
| <input type="checkbox"/> Reappearance of heart disease       | <input type="checkbox"/> Reappearance of kidney disease |
| <input type="checkbox"/> Reappearance of liver disease       | <input type="checkbox"/> Rejection                      |
| <input type="checkbox"/> Transplant related renal impairment | <input type="checkbox"/> Other                          |

Complications post fifth transplant, other- specify \_\_\_\_\_

Patient Name \_\_\_\_\_

Date

Provide details for any other transplants not listed above

## Comments

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Transplant comments