

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

Date      

## MCD - 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    Organic acid disordersSpecify organic acid disorder diagnosis for the patient    Holocarboxylase synthetase deficiency (MCD)

Patient disorder identification method

 Unknown                       Abnormal newborn screen                       Abnormal labs

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

Date       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)
- Master's degree (e.g., MA, MS, MEng, MEd, MSW, MBA)

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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
- Romania  Serbia  Slovakia  Slovenia  Spain

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

Date       Sweden  Switzerland  Ukraine  United Kingdom  OtherAncestral 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  OtherAncestral 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  WhiteRace-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 GA  HI  ID  IL  IN  IA

Patient Name _____
Date <input type="text" value="MM"/> <input type="text" value="DD"/> <input type="text" value="YY"/>

- |                          |                          |                          |                          |                          |                          |
|--------------------------|--------------------------|--------------------------|--------------------------|--------------------------|--------------------------|
| <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

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

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## ***MCD - 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  BorderlineSibling 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  BorderlineSibling 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 \_\_\_\_\_

Date      

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



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

Date      

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  BorderlineSibling 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  BorderlineSibling 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

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

Date      

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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Date    |    |   

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"/>

- 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\_\_\_\_\_

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

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

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

**Comments**

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Intake family history comments

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

Date      

## ***MCD - 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

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

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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**

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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**

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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  No

Date 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.

Hospitalizations 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 \_\_\_\_\_

Comorbidities at time of intake

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

Date      **Prior testing**

---

Echocardiogram prior to intake  Unknown  Yes  NoEchocardiogram date      Echocardiogram results  Unknown  Normal  Abnormal

Echocardiogram comments

Electrocardiogram prior to intake  Unknown  Yes  NoElectrocardiogram date      Electrocardiogram results  Unknown  Normal  Abnormal

Electrocardiogram comments

Neurological imaging prior to intake  Unknown  Yes  NoNeurological imaging date      Neurological imaging results  Unknown  Normal  Abnormal

Neurological imaging comments

History of a seizure disorder  Unknown  Yes  NoPatient has biospecimen stored  Unknown  Yes  NoType of patient biospecimen  Blood  Urine  Other tissue

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

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

**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  NoPatient/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

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

Date

## Comments

---

Intake past health history comments

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

Date      

## ***MCD - 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

Acetylcarnitine (C2) on first newborn screen \_\_\_\_\_

Acetylcarnitine (C2) on first newborn screen units  umol/L

Propionylcarnitine (C3) on first newborn screen \_\_\_\_\_

Propionylcarnitine (C3) on first newborn screen units  umol/L

Propionylcarnitine (C3)/Acetylcarnitine (C2) on first newborn screen \_\_\_\_\_

Propionylcarnitine (C3)/Palmitoylcarnitine (C16) on first newborn screen \_\_\_\_\_

3-Hydroxyisovalerylcarnitine (C5-OH) on first newborn screen \_\_\_\_\_

3-Hydroxyisovalerylcarnitine (C5-OH) on first newborn screen units  umol/L

3-Hydroxyisovalerylcarnitine (C5-OH)/Carnitine.free (C0) on first newborn screen \_\_\_\_\_

3-Hydroxyisovalerylcarnitine (C5-OH)/Octanoylcarnitine (C8) on first newborn screen \_\_\_\_\_

Octanoylcarnitine (C8) on first newborn screen \_\_\_\_\_

Octanoylcarnitine (C8) on first newborn screen units  umol/L

Methylmalonylcarnitine (C4-DC) on first newborn screen \_\_\_\_\_

Methylmalonylcarnitine (C4-DC) on first newborn screen units  umol/L

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

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

Methylmalonylcarnitine (C4-DC)+3-Hydroxyisovalerylcarnitine (C5-OH) on first newborn screen \_\_\_\_\_

Methylmalonylcarnitine (C4-DC)+3-Hydroxyisovalerylcarnitine (C5-OH) on first newborn screen units

 umol/L

Other result/report on first newborn screen

--

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

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

Date

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 \_\_\_\_\_

Acetylcarnitine (C2) on second newborn screen \_\_\_\_\_

Acetylcarnitine (C2) on second newborn screen units  umol/L

Propionylcarnitine (C3) on second newborn screen \_\_\_\_\_

Propionylcarnitine (C3) on second newborn screen units  umol/L

Propionylcarnitine (C3)/Acetylcarnitine (C2) on second newborn screen \_\_\_\_\_

Propionylcarnitine (C3)/Palmitoylcarnitine (C16) on second newborn screen \_\_\_\_\_

3-Hydroxyisovalerylcarnitine (C5-OH) on second newborn screen \_\_\_\_\_

3-Hydroxyisovalerylcarnitine (C5-OH) on second newborn screen units  umol/L

3-Hydroxyisovalerylcarnitine (C5-OH)/Carnitine.free (C0) on second newborn screen \_\_\_\_\_

3-Hydroxyisovalerylcarnitine (C5-OH)/Octanoylcarnitine (C8) on second newborn screen \_\_\_\_\_

Octanoylcarnitine (C8) on second newborn screen \_\_\_\_\_

Octanoylcarnitine (C8) on second newborn screen units  umol/L

Methylmalonylcarnitine (C4-DC) on second newborn screen \_\_\_\_\_

Methylmalonylcarnitine (C4-DC) on second newborn screen units  umol/L

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

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

Methylmalonylcarnitine (C4-DC)+3-Hydroxyisovalerylcarnitine (C5-OH) on second newborn screen \_\_\_\_\_

Methylmalonylcarnitine (C4-DC)+3-Hydroxyisovalerylcarnitine (C5-OH) 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

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

Date      

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

Acetylcarnitine (C2) on third newborn screen \_\_\_\_\_

Acetylcarnitine (C2) on third newborn screen units  umol/L

Propionylcarnitine (C3) on third newborn screen \_\_\_\_\_

Propionylcarnitine (C3) on third newborn screen units  umol/L

Propionylcarnitine (C3)/Acetylcarnitine (C2) on third newborn screen \_\_\_\_\_

Propionylcarnitine (C3)/Palmitoylcarnitine (C16) on third newborn screen \_\_\_\_\_

3-Hydroxyisovalerylcarnitine (C5-OH) on third newborn screen \_\_\_\_\_

3-Hydroxyisovalerylcarnitine (C5-OH) on third newborn screen units  umol/L

3-Hydroxyisovalerylcarnitine (C5-OH)/Carnitine.free (C0) on third newborn screen \_\_\_\_\_

3-Hydroxyisovalerylcarnitine (C5-OH)/Octanoylcarnitine (C8) on third newborn screen \_\_\_\_\_

Octanoylcarnitine (C8) on third newborn screen \_\_\_\_\_

Octanoylcarnitine (C8) on third newborn screen units  umol/L

Methylmalonylcarnitine (C4-DC) on third newborn screen \_\_\_\_\_

Methylmalonylcarnitine (C4-DC) on third newborn screen units  umol/L

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

Palmitoylcarnitine (C16) on third newborn screen units  umol/LMethylmalonylcarnitine (C4-DC)+3-Hydroxyisovalerylcarnitine (C5-OH) on third newborn screen  
\_\_\_\_\_Methylmalonylcarnitine (C4-DC)+3-Hydroxyisovalerylcarnitine (C5-OH) 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 required

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

Date [M][M][D][D][Y][Y]

Date of Audiological Diagnostic Evaluation [M][M][D][D][Y][Y]

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
- Other

Left 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**

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Intake newborn screening comments



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

Date      ***MCD - Intake Initial Testing***

Patient status at time of NBS reporting to specialty center

 Unknown       Well       Symptomatic       Deceased

Patient symptoms at time of initial contact

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

- Unknown
- None
- 3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay
- Km for biotin
- Plasma acylcarnitine profile
- Propionyl-CoA Carboxylase (PCC) enzyme assay
- Pyruvate Carboxylase (PC) enzyme assay
- Urine acylcarnitine
- Urine organic acids
- Other

Number of times 3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay done  1  2

3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay

Unknown  Within normal limits  Abnormal

3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay significance

Diagnostic  Non-diagnostic

3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay value \_\_\_\_\_

3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay units  pmol/min/mg protein

3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay reference range

\_\_\_\_\_

3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay medium

Unknown  Fibroblasts  Lymphocytes  Other

3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay medium, other- specify

\_\_\_\_\_

Second 3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay

Unknown  Within normal limits  Abnormal

Second 3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay significance

Diagnostic  Non-diagnostic

Second 3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay value \_\_\_\_\_

Second 3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay units  pmol/min/mg protein

Second 3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay reference range

\_\_\_\_\_

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

Date      

Second 3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay medium

 Unknown       Fibroblasts       Lymphocytes       OtherSecond 3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay medium, other- specify  
\_\_\_\_\_Km for biotin     Unknown     From fibroblasts     From lymphocytesKm for biotin from fibroblasts     Unknown     Normal     High     Low

Km for biotin from fibroblasts value \_\_\_\_\_

Km for biotin from fibroblasts units     nmol/L

Km for biotin from fibroblasts reference range \_\_\_\_\_

Km for biotin from lymphocytes     Unknown     Normal     High     Low

Km for biotin from lymphocytes value \_\_\_\_\_

Km for biotin from lymphocytes units     nmol/L

Km for biotin from lymphocytes reference range \_\_\_\_\_

Plasma acylcarnitine profile     Unknown     Within normal limits     AbnormalPlasma acylcarnitine profile significance     Diagnostic     Non-diagnosticPlasma acylcarnitine profile comments  

Propionyl-CoA Carboxylase (PCC) enzyme assay

 Unknown Propionyl-CoA Carboxylase enzyme assay from fibroblasts Propionyl-CoA Carboxylase enzyme assay from lymphocytesNumber of times Propionyl-CoA Carboxylase (PCC) enzyme assay from fibroblasts done     1     2

Propionyl-CoA Carboxylase (PCC) enzyme assay from fibroblasts

 Unknown       Within normal limits       Abnormal

Propionyl-CoA Carboxylase (PCC) enzyme assay from fibroblasts significance

 Diagnostic       Non-diagnostic

Propionyl-CoA Carboxylase (PCC) enzyme assay from fibroblasts value \_\_\_\_\_

Propionyl-CoA Carboxylase (PCC) enzyme assay from fibroblasts units     pmol/min/mg proteinPropionyl-CoA Carboxylase (PCC) enzyme assay from fibroblasts reference range  
\_\_\_\_\_

Second Propionyl-CoA Carboxylase (PCC) enzyme assay from fibroblasts

 Unknown       Within normal limits       Abnormal

Second Propionyl-CoA Carboxylase (PCC) enzyme assay from fibroblasts significance

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

Diagnostic  Non-diagnostic

Second Propionyl-CoA Carboxylase (PCC) enzyme assay from fibroblasts value \_\_\_\_\_

Second Propionyl-CoA Carboxylase (PCC) enzyme assay from fibroblasts units

pmol/min/mg protein

Second Propionyl-CoA Carboxylase (PCC) enzyme assay from fibroblasts reference range

\_\_\_\_\_

Number of times Propionyl-CoA Carboxylase (PCC) enzyme assay from lymphocytes done  1  2

Propionyl-CoA Carboxylase (PCC) enzyme assay from lymphocytes

Unknown  Within normal limits  Abnormal

Propionyl-CoA Carboxylase (PCC) enzyme assay from lymphocytes significance

Diagnostic  Non-diagnostic

Propionyl-CoA Carboxylase (PCC) enzyme assay from lymphocytes value \_\_\_\_\_

Propionyl-CoA Carboxylase (PCC) enzyme assay from lymphocytes units  pmol/min/mg protein

Propionyl-CoA Carboxylase (PCC) enzyme assay from lymphocytes reference range

\_\_\_\_\_

Second Propionyl-CoA Carboxylase (PCC) enzyme assay from lymphocytes

Unknown  Within normal limits  Abnormal

Second Propionyl-CoA Carboxylase (PCC) enzyme assay from lymphocytes significance

Diagnostic  Non-diagnostic

Second Propionyl-CoA Carboxylase (PCC) enzyme assay from lymphocytes value \_\_\_\_\_

Second Propionyl-CoA Carboxylase (PCC) enzyme assay from lymphocytes units

pmol/min/mg protein

Second Propionyl-CoA Carboxylase (PCC) enzyme assay from lymphocytes reference range

\_\_\_\_\_

Number of times Propionyl-CoA Carboxylase (PCC) enzyme assay from liver done  1  2

Propionyl-CoA Carboxylase (PCC) enzyme assay from liver

Unknown  Within normal limits  Abnormal

Propionyl-CoA Carboxylase (PCC) enzyme assay from liver significance

Diagnostic  Non-diagnostic

Propionyl-CoA Carboxylase (PCC) enzyme assay from liver value \_\_\_\_\_

Propionyl-CoA Carboxylase (PCC) enzyme assay from liver units  pmol/min/mg protein

Propionyl-CoA Carboxylase (PCC) enzyme assay from liver reference range

\_\_\_\_\_

Second Propionyl-CoA Carboxylase (PCC) enzyme assay from liver

Unknown  Within normal limits  Abnormal

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

Date      

Second Propionyl-CoA Carboxylase (PCC) enzyme assay from liver significance

 Diagnostic Non-diagnostic

Second Propionyl-CoA Carboxylase (PCC) enzyme assay from liver value \_\_\_\_\_

Second Propionyl-CoA Carboxylase (PCC) enzyme assay from liver units  pmol/min/mg proteinSecond Propionyl-CoA Carboxylase (PCC) enzyme assay from liver reference range  
\_\_\_\_\_

Pyruvate Carboxylase (PC) enzyme assay

 Unknown Pyruvate Carboxylase enzyme assay from fibroblasts Pyruvate Carboxylase enzyme assay from lymphocytesNumber of times Pyruvate Carboxylase (PC) enzyme assay from fibroblasts done  1  2

Pyruvate Carboxylase (PC) enzyme assay from fibroblasts

 Unknown Within normal limits Abnormal

Pyruvate Carboxylase (PC) enzyme assay from fibroblasts significance

 Diagnostic Non-diagnostic

Pyruvate Carboxylase (PC) enzyme assay from fibroblasts value \_\_\_\_\_

Pyruvate Carboxylase (PC) enzyme assay from fibroblasts units  pmol/min/mg proteinPyruvate Carboxylase (PC) enzyme assay from fibroblasts reference range  
\_\_\_\_\_

Second Pyruvate Carboxylase (PC) enzyme assay from fibroblasts

 Unknown Within normal limits Abnormal

Second Pyruvate Carboxylase (PC) enzyme assay from fibroblasts significance

 Diagnostic Non-diagnostic

Second Pyruvate Carboxylase (PC) enzyme assay from fibroblasts value \_\_\_\_\_

Second Pyruvate Carboxylase (PC) enzyme assay from fibroblasts units  pmol/min/mg proteinSecond Pyruvate Carboxylase (PC) enzyme assay from fibroblasts reference range  
\_\_\_\_\_Number of times Pyruvate Carboxylase (PC) enzyme assay from lymphocytes done  1  2

Pyruvate Carboxylase (PC) enzyme assay from lymphocytes

 Unknown Within normal limits Abnormal

Pyruvate Carboxylase (PC) enzyme assay from lymphocytes significance

 Diagnostic Non-diagnostic

Pyruvate Carboxylase (PC) enzyme assay from lymphocytes value \_\_\_\_\_

Pyruvate Carboxylase (PC) enzyme assay from lymphocytes units  pmol/min/mg protein

Pyruvate Carboxylase (PC) enzyme assay from lymphocytes reference range

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

Second Pyruvate Carboxylase (PC) enzyme assay from lymphocytes

Unknown  Within normal limits  Abnormal

Second Pyruvate Carboxylase (PC) enzyme assay from lymphocytes significance

Diagnostic  Non-diagnostic

Second Pyruvate Carboxylase (PC) enzyme assay from lymphocytes value \_\_\_\_\_

Second Pyruvate Carboxylase (PC) enzyme assay from lymphocytes units  pmol/min/mg protein

Second Pyruvate Carboxylase (PC) enzyme assay from lymphocytes reference range \_\_\_\_\_

Urine acylcarnitine

Unknown  Urine C4 acylcarnitine  Urine C5-OH acylcarnitine

Urine C5-DC acylcarnitine

Urine C4 acylcarnitine  Unknown  Within normal limits  Abnormal

Urine C4 acylcarnitine significance  Diagnostic  Non-diagnostic

Urine C4 acylcarnitine value \_\_\_\_\_

Urine C4 acylcarnitine units  mmol/mol Cr

Urine C4 acylcarnitine reference range \_\_\_\_\_

Urine C5-OH acylcarnitine  Unknown  Within normal limits  Abnormal

Urine C5-OH acylcarnitine significance  Diagnostic  Non-diagnostic

Urine C5-OH acylcarnitine value \_\_\_\_\_

Urine C5-OH acylcarnitine units  mmol/mol Cr

Urine C5-OH acylcarnitine reference range \_\_\_\_\_

Urine C5-DC acylcarnitine  Unknown  Within normal limits  Abnormal

Urine C5-DC acylcarnitine significance  Diagnostic  Non-diagnostic

Urine C5-DC acylcarnitine value \_\_\_\_\_

Urine C5-DC acylcarnitine units  mmol/mol Cr

Urine C5-DC acylcarnitine reference range \_\_\_\_\_

Urine organic acids  Unknown  Within normal limits  Abnormal

Urine organic acids significance  Diagnostic  Non-diagnostic

Urine organic acid comments

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

Other diagnostic lab  Unknown  Within normal limits  Abnormal

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

Date

Other 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

- Unknown                       Not done                       Done, not available
- Single gene                       Mutation panel                       Exome sequencing
- Full genome sequencing       Copy number variant               Deletion/duplication testing
- Other

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 MCD     HLCS     Other

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

HLCS: Specify allele 1 \_\_\_\_\_

HLCS: 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 \_\_\_\_\_



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

## Comments

---

Initial testing comments

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

Date      

## ***MCD - 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      

## ***MCD - 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  NoDocumented 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

### **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  
 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 \_\_\_\_\_

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"/>
--

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

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

Date of sick visit 2

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

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 \_\_\_\_\_

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

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

Date      

- 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  
 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 \_\_\_\_\_

Patient Name _____ Date <input type="text" value="MM"/> <input type="text" value="MM"/> <input type="text" value="DD"/> <input type="text" value="DD"/> <input type="text" value="YY"/> <input type="text" value="YY"/>
--

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 \_\_\_\_\_

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 unrelated

Sick visit 9 was a condition exacerbation    Unknown    Yes    No

Location for sick visit 9

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 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 unrelated

Sick 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    Other

Patient 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    No

Specific anesthesia precautions recommended due to metabolic condition    Unknown    Yes    No

Surgical precautions taken

Anesthesia complications    Unknown    Yes    No

Anesthesia complications-specify



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

Date

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 \_\_\_\_\_

**Comments**

---

Visit health history comments

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

# MCD - 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       Evidence of alopecia       Evidence of ataxia

Evidence of dermatitis     Other

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

## Comments

Visit findings comments

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

Date / /

## **MCD - 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 transplant

Transplant 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 <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

Developmental status  Typical  Atypical

Severity 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  
  Other

Developmental 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

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown             | <input type="checkbox"/> Developmental/behavioral pediatrician |
| <input type="checkbox"/> Neuropsychologist   | <input type="checkbox"/> Psychiatric APRN/CNP/CNS              |
| <input type="checkbox"/> Psychiatrist        | <input type="checkbox"/> Psychologist                          |
| <input type="checkbox"/> School psychologist | <input type="checkbox"/> Other                                 |

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

\_\_\_\_\_

Neuropsychometric evaluation performed since last visit  Unknown  Yes  No

Overall neuropsychometric impression  Above average  Average  Below average  In progress

Patient 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

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                       | <input type="checkbox"/> Developmental/behavioral pediatrician |
| <input type="checkbox"/> Marriage and family therapist | <input type="checkbox"/> Mental health counselor               |
| <input type="checkbox"/> Neuropsychologist             | <input type="checkbox"/> Pastoral counselor                    |
| <input type="checkbox"/> Psychiatric APRN/CNP/CNS      | <input type="checkbox"/> Psychiatrist                          |
| <input type="checkbox"/> Psychologist                  | <input type="checkbox"/> School psychologist                   |
| <input type="checkbox"/> Social worker                 | <input type="checkbox"/> 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      

## ***MCD - Visit Lab Studies***

### **Biochemical Labs**

Biochemical labs associated with this visit

- Unknown
- None
- 3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay
- Km for biotin
- Lactic acid
- Plasma acylcarnitine profile
- Propionyl-CoA Carboxylase (PCC) enzyme assay
- Pyruvate Carboxylase (PC) enzyme assay
- Urine acylcarnitine
- Urine organic acids
- Other

Please complete information about the 3-Methylcrotonyl CoA Carboxylase (3-MCC) enzyme assay on the Intake Initial Testing form.

Please complete information about Km for biotin on the Intake Initial Testing form.

Lactic acid    Within normal limits    Abnormal    In progress    Results unavailable

Lactic acid value \_\_\_\_\_

Lactic acid units    mmol/L    mg/dL

Lactic acid reference range \_\_\_\_\_

Plasma acylcarnitine profile    Within normal limits    Abnormal    In progress    Results unavailable

Plasma acylcarnitine profile comments

Please complete information about the Propionyl-CoA carboxylase (PCC) enzyme assay on the Intake Initial Testing form.

Please complete information about the Pyruvate Carboxylase (PC) enzyme assay on the Intake Initial Testing form.

Urine acylcarnitine

- Unknown                       Urine C4 acylcarnitine                       Urine C5-OH acylcarnitine
- Urine C5-DC acylcarnitine



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

Date

Urine C4 acylcarnitine  Within normal limits  Abnormal  In progress  Results unavailable

Urine C4 acylcarnitine value \_\_\_\_\_

Urine C4 acylcarnitine units  mmol/mol Cr

Urine C4 acylcarnitine reference range \_\_\_\_\_

Urine C5-OH acylcarnitine  Within normal limits  Abnormal  In progress  Results unavailable

Urine C5-OH acylcarnitine value \_\_\_\_\_

Urine C5-OH acylcarnitine units  mmol/mol Cr

Urine C5-OH acylcarnitine reference range \_\_\_\_\_

Urine C5-DC acylcarnitine  Within normal limits  Abnormal  In progress  Results unavailable

Urine C5-DC acylcarnitine value \_\_\_\_\_

Urine C5-DC acylcarnitine units  mmol/mol Cr

Urine C5-DC acylcarnitine reference range \_\_\_\_\_

Urine organic acids  Within normal limits  Abnormal  In progress  Results unavailable

Urine organic acid comments

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

Other visit lab  Within normal limits  Abnormal  In progress  Results unavailable

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) |

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

Date

Total cholesterol (random)  Uric acid  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 \_\_\_\_\_

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 \_\_\_\_\_

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

Date      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

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

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

Date      

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 \_\_\_\_\_

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 \_\_\_\_\_

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

Date    |    |

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<sup>9</sup>/L  10<sup>3</sup>/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<sup>6</sup>/uL  10<sup>12</sup>/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<sup>3</sup>/uL  10<sup>9</sup>/uL

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

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

Other hematology labs  Within normal limits  Abnormal  In progress  Results unavailable

Other hematology lab comments

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

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

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

### Liver Labs

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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 transpeptide | <input type="checkbox"/> Globulin                   |
| <input type="checkbox"/> Prealbumin           | <input type="checkbox"/> Total bilirubin             | <input type="checkbox"/> Total protein              |

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

Date       OtherAlbumin  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

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

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

Date      

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

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Renal labs associated with this visit

 Unknown None Blood urea nitrogen Serum creatinine 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

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

Date      

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

- |   |  |   |
|---|--|---|
| <input type="checkbox"/> Unknown                | <input type="checkbox"/> None                        | <input type="checkbox"/> B-type natriuretic peptide     |
| <input type="checkbox"/> Creatine phosphokinase | <input type="checkbox"/> C-reactive protein          | <input type="checkbox"/> Erythrocyte sedimentation rate |
| <input type="checkbox"/> Ferritin               | <input type="checkbox"/> Thyroid stimulating hormone | <input type="checkbox"/> Thyroxine (free)               |
| <input type="checkbox"/> Thyroxine (total)      | <input type="checkbox"/> Transferrin                 | <input type="checkbox"/> Troponin                       |
| <input type="checkbox"/> Urinalysis             | <input type="checkbox"/> Zinc                        | <input type="checkbox"/> Other                          |

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



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

Date

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

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

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"/>

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      

## ***MCD - Visit Studies Other***

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### **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

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

Musculoskeletal findings of note

Neurological imaging

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

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"/>
--

Positron emission tomography (PET) scan Abn

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 \_\_\_\_\_

Other imaging studies date   |   |

Other imaging studies explanation

**Comments**

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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"/>

Visit studies-other comments

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

Date |||||

## ***MCD - 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  Unknown  None  Biotin  Other

#### **Biotin**

Biotin formulation recommended/prescribed  Unknown  Capsule  Suspension  TabletBiotin route recommended/prescribed  Unknown  Feeding tube  Oral

Biotin dose recommended/prescribed \_\_\_\_\_

Biotin dose recommended/prescribed units  g  mg  ugBiotin frequency recommended/prescribed  Unknown  Once/day  Twice/day  PRN  Other

Biotin frequency recommended/prescribed-other, specify \_\_\_\_\_

Biotin taken as recommended/prescribed  Unknown  Yes  No

Actual biotin dose reported \_\_\_\_\_

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

Actual biotin dose reported units     g    mg    ug  
 Actual biotin frequency reported     Unknown    Once/day    Twice/day    PRN    Other  
 Actual biotin frequency reported-other, specify \_\_\_\_\_  
 Reason biotin is not taken as recommended/prescribed \_\_\_\_\_

Method of payment for biotin

- 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 \_\_\_\_\_  
 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



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"/>

- 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      ***MCD - 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 \_\_\_\_\_

**Comments**

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Nutrition comments

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

Date        ***MCD - Study Status***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 <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>

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

- |   |   |  |
|---|---|--|
| <input type="radio"/> Unknown                       | <input type="radio"/> Deceased          | <input type="radio"/> Lost to follow-up            |
| <input type="radio"/> Moved                         | <input type="radio"/> Refused follow-up | <input type="radio"/> Follow-up deemed unnecessary |
| <input type="radio"/> 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

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 <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"/>
--

- 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

- |   |   |  |
|---|---|--|
| <input type="radio"/> Unknown                       | <input type="radio"/> Deceased          | <input type="radio"/> Lost to follow-up            |
| <input type="radio"/> Moved                         | <input type="radio"/> Refused follow-up | <input type="radio"/> Follow-up deemed unnecessary |
| <input type="radio"/> 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      

## ***MCD - Pregnancy***

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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 <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

- |   |  |
|---|--|
| <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 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

- |   |  |
|---|--|
| <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 first 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 first pregnancy-other, specify \_\_\_\_\_

Partner tested for patient's disorder during first 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 first pregnancy    Unknown    Yes    No

Preimplantation genetic diagnosis for disorder done for first pregnancy    Unknown    Yes    No

Prenatal testing done for fetus for this disorder for first pregnancy    Unknown    Yes    No

Type of prenatal testing performed for first pregnancy    Unknown    Biochemical    Molecular

Method of prenatal testing for first 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 first 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                |   |

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      

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 |||||

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 <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>

- 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 \_\_\_\_\_

Number of documented episodes of hyperammonemia during first pregnancy

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Number of documented episodes of ketonuria during first pregnancy

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Number of documented episodes of metabolic acidosis during first pregnancy

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Treatment(s) for hyperammonemic episodes during first pregnancy

- Unknown                       No hyperammonemic episodes     Hospital admission
- Protein restriction               Ammonul                       Arginine

Maternal protein intake restricted during first pregnancy     Unknown     Yes     No

Number of grams of synthetic protein prescribed per day during first pregnancy (units = gm/day) \_\_\_\_\_

Number of grams of natural protein prescribed per day during first pregnancy (units = gm/day) \_\_\_\_\_

Ammonia scavenger drugs used during first pregnancy

- Unknown                       No scavengers used       Buphenyl                       Sodium benzoate

Other medications/supplements used during first 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

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

Date

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

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

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

Date |||||

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)

- 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

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 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

Unknown  Yes - normal for all screened disorders

Yes -abnormal  Yes - results pending

No

Describe abnormal newborn screen result for first newborn

Reason routine newborn screening was not done for first newborn

Unknown

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

Date      

- 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 \_\_\_\_\_

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

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

Date      

## 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

\_\_\_\_\_

## 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

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

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\_\_\_\_\_

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 Name \_\_\_\_\_

Date      Patient in good metabolic condition prior to second pregnancy  Unknown  Yes  NoPatient 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

 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



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

- 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

- |  |  |
|--|--|
| <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          |

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

Date

- Plasma acylcarnitine profile - Abn       Plasma acylcarnitine profile - WNL
- Plasma amino acids - Abn                 Plasma amino acids - WNL
- Plasma carnitine levels - Abn             Plasma carnitine levels - WNL
- Prealbumin - Abn low                     Prealbumin - WNL
- Transferrin - Abn                          Transferrin - WNL
- Urine acylcarnitines - Abn                Urine acylcarnitines - WNL
- Urine acylglycines - Abn                 Urine acylglycines - WNL
- Urine ketones - Abn high                 Urine ketones - WNL
- Urine organic acids - Abn                Urine organic acids - WNL
- 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

- Unknown                                       ED visits for hyperemesis/IV fluids
- Hospitalizations for hyperemesis/IV fluids     TPN
- Tube feedings                                 Additional medications
- Home lab monitoring                         Increased frequency of lab monitoring
- More intensive fetal monitoring             Bedrest
- Other

Additional interventions required during second pregnancy due to this metabolic condition-other, specify

\_\_\_\_\_

Total maternal weight gain (in kg) during second pregnancy \_\_\_\_\_

Number of documented episodes of hyperammonemia during second pregnancy

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Number of documented episodes of ketonuria during second pregnancy

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Number of documented episodes of metabolic acidosis during second pregnancy

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Treatment(s) for hyperammonemic episodes during second pregnancy

- Unknown                                       No hyperammonemic episodes     Hospital admission

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

Date

- Protein restriction
- Ammonul
- Arginine

Maternal protein intake restricted during second pregnancy     Unknown     Yes     No

Number of grams of synthetic protein prescribed per day during second pregnancy (units = gm/day) \_\_\_\_\_

Number of grams of natural protein prescribed per day during second pregnancy (units = gm/day) \_\_\_\_\_

Ammonia scavenger drugs used during second pregnancy

- Unknown
- No scavengers used
- Buphenyl
- Sodium benzoate

Other medications/supplements used during second 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
- 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

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

Date

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

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

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

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

Health concerns for second newborn not known to be related to disorder

[Empty text box for health concerns]

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

[Empty text box for additional interventions]

Birth measurements for second 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 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

[Empty text box for health concerns with APGAR < 8]

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

[Empty text box for health concerns with APGAR < 8]

Length of second newborn's stay in the hospital after birth

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

Date

- 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
- 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 - bichemical 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

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

Date      

## Abnormal newborn exam findings for second 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 second newborn-other, specify \_\_\_\_\_

Newborn death at or shortly after delivery for second newborn  Unknown  Yes  NoNewborn's death related to maternal disorder for second newborn  Unknown  Yes  NoSecond 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)

- |                               |                                 |                           |                         |                          |                          |                          |
|-------------------------------|---------------------------------|---------------------------|-------------------------|--------------------------|--------------------------|--------------------------|
| <input type="radio"/> Unknown | <input type="radio"/> Unplanned | <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"/> 11 | <input type="radio"/> 12 |
| <input type="radio"/> 13-24   | <input type="radio"/> 25-36     | <input type="radio"/> >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

- |  |                                      |  |
|--|--------------------------------------|--|
| <input type="checkbox"/> Unknown                     | <input type="checkbox"/> Male factor | <input type="checkbox"/> Female factor |
| <input type="checkbox"/> Combined male-female factor |                                      |  |

Cause(s) of male infertility for third pregnancy

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown       | <input type="checkbox"/> Abnormal sperm production or function |
| <input type="checkbox"/> Age           | <input type="checkbox"/> Cancer/cancer treatment related       |
| <input type="checkbox"/> Environmental | <input type="checkbox"/> Problems with sperm delivery          |
| <input type="checkbox"/> Tobacco use   | <input type="checkbox"/> Other                                 |

Cause(s) of male infertility for third pregnancy-other, specify \_\_\_\_\_

Cause(s) of female infertility for third 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 |

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"/>
---

- Over-exercise
- Thyroid problems
- Uterine fibroid
- Other
- Ovulation disorder
- Tobacco use
- Weight related

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
- Assisted reproductive technology
- Surgery
- None
- Fertility drugs

Type of assisted reproductive technologies used during third pregnancy

- Assisted hatching
- Surgical sperm aspiration
- ICSI
- Other
- IVF

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
- No

Partner also affected by disorder during third pregnancy  Unknown  Yes  No

Preimplantation genetic diagnosis for disorder done for third pregnancy  Unknown  Yes  No

Prenatal testing done for fetus for this disorder for third pregnancy  Unknown  Yes  No

Type 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
- Fetal echocardiogram
- Other
- None
- Increased frequency prenatal ultrasounds

Additional 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



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

Date

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

Treatment prescribed for patient's disorder during third pregnancy-other, specify

Patient in good metabolic condition prior to third pregnancy  Unknown  Yes  No

Patient in good metabolic condition during first trimester of third pregnancy  Unknown  Yes  No

Patient in good metabolic condition during second trimester of third pregnancy

Unknown

Pregnancy not sustained to second trimester

Yes

No

Patient in good metabolic condition during third trimester of third pregnancy

Unknown

Pregnancy not sustained to third trimester

Yes

No

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

5

6

7

8

9

10

>10

Number of hospitalizations for management of disorder during third pregnancy

Unknown  0

1

2

3

4

5

6

7

8

9

10

>10

Complications during third pregnancy

Unknown

Yes-related to disorder

Yes- not known to be related to disorder  No

Complications related to disorder during third pregnancy- specify

Unknown

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

- 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 third 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 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)

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

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

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"/> 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 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 |

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

- More intensive fetal monitoring
- Bedrest
- Other

Additional interventions required during third pregnancy due to this metabolic condition-other, specify

\_\_\_\_\_

Total maternal weight gain (in kg) during third pregnancy \_\_\_\_\_

Number of documented episodes of hyperammonemia during third pregnancy

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Number of documented episodes of ketonuria during third pregnancy

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Number of documented episodes of metabolic acidosis during third pregnancy

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Treatment(s) for hyperammonemic episodes during third pregnancy

- Unknown                       No hyperammonemic episodes     Hospital admission
- Protein restriction               Ammonul                       Arginine

Maternal protein intake restricted during third pregnancy     Unknown     Yes     No

Number of grams of synthetic protein prescribed per day during third pregnancy (units = gm/day) \_\_\_\_\_

Number of grams of natural protein prescribed per day during third pregnancy (units = gm/day) \_\_\_\_\_

Ammonia scavenger drugs used during third pregnancy

- Unknown                       No scavengers used       Buphenyl                       Sodium benzoate

Other medications/supplements used during third 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      

Cause(s) of male infertility for fourth pregnancy-other, specify \_\_\_\_\_

Cause(s) of female infertility for fourth 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 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

- |   |  |
|---|--|
| <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 fourth 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 fourth pregnancy-other, specify

\_\_\_\_\_

Partner tested for patient's disorder during fourth 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 fourth pregnancy  Unknown  Yes  NoPreimplantation genetic diagnosis for disorder done for fourth pregnancy  Unknown  Yes  NoPrenatal testing done for fetus for this disorder for fourth pregnancy  Unknown  Yes  NoType of prenatal testing performed for fourth pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for fourth 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 fourth 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 <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 <input type="text" value="MM"/> <input type="text" value="DD"/> <input type="text" value="YY"/>

- 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

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

- Unknown  ED visits for hyperemesis/IV fluids
- Hospitalizations for hyperemesis/IV fluids  TPN
- Tube feedings  Additional medications
- Home lab monitoring  Increased frequency of lab monitoring
- More intensive fetal monitoring  Bedrest
- Other

Additional interventions required during fourth pregnancy due to this metabolic condition-other, specify

\_\_\_\_\_

Total maternal weight gain (in kg) during fourth pregnancy \_\_\_\_\_

Number of documented episodes of hyperammonemia during fourth pregnancy

- 0
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10
- >10
- unknown

Number of documented episodes of ketonuria during fourth pregnancy

- 0
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10
- >10
- unknown

Number of documented episodes of metabolic acidosis during fourth pregnancy

- 0
- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10
- >10
- unknown

Treatment(s) for hyperammonemic episodes during fourth pregnancy

- Unknown  No hyperammonemic episodes  Hospital admission
- Protein restriction  Ammonul  Arginine

Maternal protein intake restricted during fourth pregnancy  Unknown  Yes  No

Number of grams of synthetic protein prescribed per day during fourth pregnancy (units = gm/day) \_\_\_\_\_

Number of grams of natural protein prescribed per day during fourth pregnancy (units = gm/day) \_\_\_\_\_

Ammonia scavenger drugs used during fourth pregnancy

- Unknown  No scavengers used  Buphenyl  Sodium benzoate

Other medications/supplements used during fourth pregnancy

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

Date

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

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

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

Date

- 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)

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

Patient Name _____ Date  M M   D D   Y Y
---

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

Health concern(s) with APGAR score at five minutes < 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



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

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

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

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                           | <input type="checkbox"/> Yes - biochemical normal                    |
| <input type="checkbox"/> Yes - biochemical abnormal        | <input type="checkbox"/> Yes - molecular normal                      |
| <input type="checkbox"/> Yes - molecular abnormal affected | <input type="checkbox"/> Yes - molecular abnormal unaffected carrier |
| <input type="checkbox"/> No                                |  |

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     NoNewborn's death related to maternal disorder for fourth newborn     Unknown     Yes     No

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

Date      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  
 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

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

Date      

- 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    NoPreimplantation genetic diagnosis for disorder done for fifth pregnancy    Unknown    Yes    NoPrenatal testing done for fetus for this disorder for fifth pregnancy    Unknown    Yes    NoType 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  
 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 \_\_\_\_\_

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

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

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  No

Patient 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

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)

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

Date

- 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
- 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

- Unknown                                       Ammonia - Abn high
- Ammonia - WNL                                 Blood glucose - Abn low
- Blood glucose - WNL                          Blood Glucose - Abn high

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

Date        

- |  |  |
|--|--|
| <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 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\_\_\_\_\_

Number of documented episodes of hyperammonemia during fifth pregnancy

- 0      1      2      3      4      5      6      7  
8      9      10      >10      unknown

Number of documented episodes of ketonuria during fifth pregnancy

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"/>

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Number of documented episodes of metabolic acidosis during fifth pregnancy

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Treatment(s) for hyperammonemic episodes during fifth pregnancy

- Unknown                               No hyperammonemic episodes     Hospital admission
- Protein restriction                       Ammonul                               Arginine

Maternal protein intake restricted during fifth pregnancy     Unknown     Yes     No

Number of grams of synthetic protein prescribed per day during fifth pregnancy (units = gm/day) \_\_\_\_\_

Number of grams of natural protein prescribed per day during fifth pregnancy (units = gm/day) \_\_\_\_\_

Ammonia scavenger drugs used during fifth pregnancy

- Unknown                       No scavengers used               Buphenyl                       Sodium benzoate

Other medications/supplements used during fifth 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

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

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

Actual interventions for labor/delivery related to patient's disorder for fifth pregnancy-other, specify

[Empty text box for interventions]

Acute health concerns experienced by the patient during delivery for fifth delivery

[Empty text box for health concerns]

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

[Empty text box for other lab results]

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

[Empty text box for other maternal interventions]

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



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

Date

Acute health concerns for fifth newborn related to maternal disorder (example: fetal distress secondary to maternal acute metabolic decompensation)

Additional lab tests and results done on fifth newborn specifically due to maternal disorder history

Health concerns for fifth newborn not known to be related to disorder

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

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

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

Date

Health concern(s) with APGAR score at one minute < 8 for fifth newborn

APGAR score at 5 minutes for fifth newborn

- Unknown  0       1       2       3       4       5
- 6       7       8       9       10

Health concern(s) with APGAR score at five minutes < 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

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

Date      

- Unknown  
 Yes - biochemical abnormal  
 Yes - molecular abnormal affected  
 No
- Yes - biochemical normal  
 Yes - molecular normal  
 Yes - molecular abnormal unaffected carrier

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

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

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

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown       | <input type="checkbox"/> Abnormal sperm production or function |
| <input type="checkbox"/> Age           | <input type="checkbox"/> Cancer/cancer treatment related       |
| <input type="checkbox"/> Environmental | <input type="checkbox"/> Problems with sperm delivery          |
| <input type="checkbox"/> Tobacco use   | <input type="checkbox"/> Other                                 |

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 |
|----------------------------------|--|--|

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

Date

Additional prenatal testing performed on fetus as a result of parent's disorder for sixth pregnancy

- Unknown  None
- Fetal echocardiogram  Increased frequency prenatal ultrasounds
- Other

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 \_\_\_\_\_

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

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

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

- 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

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

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

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

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"/> 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 sixth pregnancy: describe test(s) and result(s)

Additional interventions required during sixth pregnancy due to this metabolic condition

- Unknown  ED visits for hyperemesis/IV fluids
- Hospitalizations for hyperemesis/IV fluids  TPN
- Tube feedings  Additional medications
- Home lab monitoring  Increased frequency of lab monitoring
- More intensive fetal monitoring  Bedrest
- Other

Additional interventions required during sixth pregnancy due to this metabolic condition-other, specify

\_\_\_\_\_

Total maternal weight gain (in kg) during sixth pregnancy \_\_\_\_\_

Number of documented episodes of hyperammonemia during sixth pregnancy

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Number of documented episodes of ketonuria during sixth pregnancy

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Number of documented episodes of metabolic acidosis during sixth pregnancy

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Treatment(s) for hyperammonemic episodes during sixth pregnancy

- Unknown  No hyperammonemic episodes  Hospital admission
- Protein restriction  Ammonul  Arginine

Maternal protein intake restricted during sixth pregnancy  Unknown  Yes  No

Number of grams of synthetic protein prescribed per day during sixth pregnancy (units = gm/day) \_\_\_\_\_

Number of grams of natural protein prescribed per day during sixth pregnancy (units = gm/day) \_\_\_\_\_

Ammonia scavenger drugs used during sixth pregnancy

- Unknown  No scavengers used  Buphenyl  Sodium benzoate



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

Date

Other medications/supplements used during sixth 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

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

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

Date

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)

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)

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

Date

- 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

Health concern(s) with APGAR score at five minutes < 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

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

Date

- Unknown
- Yes - abnormal
- No
- Yes - normal for all screened disorders
- Yes - results pending

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

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 \_\_\_\_\_

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

Date      Newborn death at or shortly after delivery for sixth newborn  Unknown  Yes  NoNewborn's death related to maternal disorder for sixth newborn  Unknown  Yes  NoSixth 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

Cause(s) of male infertility for seventh pregnancy-other, specify \_\_\_\_\_

Cause(s) of female infertility for seventh 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 seventh pregnancy-other, specify \_\_\_\_\_

Cause(s) of infertility for seventh pregnancy, combined male-female factor- specify

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

Date      

Types of fertility treatment(s) received prior to seventh pregnancy

- Unknown  None  
 Assisted reproductive technology  Fertility drugs  
 Surgery

Type of assisted reproductive technologies used during seventh pregnancy

- Assisted hatching  ICSI  IVF  
 Surgical sperm aspiration  Other

Type of assisted reproductive technologies used during seventh pregnancy-other, specify \_\_\_\_\_

Partner tested for patient's disorder during seventh pregnancy

- Unknown  Yes-biochemical  Yes-molecular  No

Partner also affected by disorder during seventh pregnancy  Unknown  Yes  No

Preimplantation genetic diagnosis for disorder done for seventh pregnancy  Unknown  Yes  No

Prenatal testing done for fetus for this disorder for seventh pregnancy  Unknown  Yes  No

Type of prenatal testing performed for seventh pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for seventh pregnancy

- Unknown  Amniocentesis  Chorionic villus sampling

Additional prenatal testing performed on fetus as a result of parent's disorder for seventh pregnancy

- Unknown  None  
 Fetal echocardiogram  Increased frequency prenatal ultrasounds  
 Other

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 \_\_\_\_\_

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

Date      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 MedicationsBiochemical 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 NoPatient 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 trimesterYes No

Patient in good metabolic condition during third trimester of seventh pregnancy

Unknown Pregnancy not sustained to third trimesterYes 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 56 7 8 9 10  >10

Number of hospitalizations for management of disorder during seventh pregnancy

Unknown 0 1 2 3 4 56 7 8 9 10  >10

Complications during seventh pregnancy

Unknown Yes-related to disorderYes- not known to be related to disorder No

Complications related to disorder during seventh 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 seventh pregnancy-specify

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

Date

- 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

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

- Unknown
- Ammonia - Abn high



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

Date

- Ammonia - WNL
  - Blood glucose - WNL
  - BNP - Abn high
  - CBC - Abn
  - CO2 - Abn low
  - CK - Abn high
  - Glucose Tolerance Test (oral) - Abn
  - INR - Abn
  - Liver function tests - Abn high
  - Plasma acylcarnitine profile - Abn
  - Plasma amino acids - Abn
  - Plasma carnitine levels - Abn
  - Prealbumin - Abn low
  - Transferrin - Abn
  - Urine acylcarnitines - Abn
  - Urine acylglycines - Abn
  - Urine ketones - Abn high
  - Urine organic acids - Abn
  - Other
- Blood glucose - Abn low
  - Blood Glucose - Abn high
  - BNP - WNL
  - CBC - WNL
  - CO2 - WNL
  - CK - WNL
  - Glucose Tolerance Test (oral) - WNL
  - INR - WNL
  - Liver function tests - WNL
  - Plasma acylcarnitine profile - WNL
  - Plasma amino acids - WNL
  - Plasma carnitine levels - WNL
  - Prealbumin - WNL
  - Transferrin - WNL
  - Urine acylcarnitines - WNL
  - Urine acylglycines - WNL
  - Urine ketones - WNL
  - Urine organic acids - WNL

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

- Unknown
- Hospitalizations for hyperemesis/IV fluids
- Tube feedings
- Home lab monitoring
- More intensive fetal monitoring
- Other
- ED visits for hyperemesis/IV fluids
- TPN
- Additional medications
- Increased frequency of lab monitoring
- Bedrest

Additional interventions required during seventh pregnancy due to this metabolic condition-other, specify

\_\_\_\_\_

Total maternal weight gain (in kg) during seventh pregnancy \_\_\_\_\_

Number of documented episodes of hyperammonemia during seventh pregnancy

- 0
- 1
- 2
- 3
- 4
- 5
- 6
- 7

Patient Name _____
Date <input type="text" value="MM"/> <input type="text" value="DD"/> <input type="text" value="YY"/>

8       9       10       >10       unknown

Number of documented episodes of ketonuria during seventh pregnancy

0       1       2       3       4       5       6       7

8       9       10       >10       unknown

Number of documented episodes of metabolic acidosis during seventh pregnancy

0       1       2       3       4       5       6       7

8       9       10       >10       unknown

Treatment(s) for hyperammonemic episodes during seventh pregnancy

- Unknown                                       No hyperammonemic episodes     Hospital admission
- Protein restriction                               Ammonul                                       Arginine

Maternal protein intake restricted during seventh pregnancy     Unknown     Yes     No

Number of grams of synthetic protein prescribed per day during seventh pregnancy (units = gm/day) \_\_\_\_\_

Number of grams of natural protein prescribed per day during seventh pregnancy (units = gm/day) \_\_\_\_\_

Ammonia scavenger drugs used during seventh pregnancy

- Unknown                               No scavengers used       Buphenyl                                       Sodium benzoate

Other medications/supplements used during seventh 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

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

Date

- Altered anesthesia plan
- Other

Actual interventions for labor/delivery related to patient's disorder for seventh pregnancy-other, specify

Acute health concerns experienced by the patient during delivery for seventh delivery

Abnormal patient lab results during seventh delivery

- Unknown
- Elevated CK
- Hypoglycemia
- None
- Elevated liver function tests
- Metabolic decompensation
- Normal labs during delivery
- Hyperammonemia
- Other

Abnormal patient lab results during seventh delivery-other, specify

Additional maternal interventions during or after seventh delivery related to this disorder

- Unknown
- Dialysis
- Medications
- Other
- Ammonul
- ICU monitoring
- Resuscitation
- Blood/blood product transfusion
- Infusions
- TPN

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
- Casesarean section (emergent)
- Caesarean section (scheduled or non-urgent)
- Vaginal

Maternal inpatient days post- seventh delivery\_\_\_\_\_

Live delivery of seventh newborn     Unknown     Yes     No

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

Date

Acute health concerns for seventh newborn related to maternal disorder (example: fetal distress secondary to maternal acute metabolic decompensation)

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

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

Date

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

Health concern(s) with APGAR score at five minutes < 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

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

Date      

- Unknown  
 Yes - biochemical abnormal  
 Yes - molecular abnormal affected  
 No
- Yes - biochemical normal  
 Yes - molecular normal  
 Yes - molecular abnormal unaffected carrier

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

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

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

Cause(s) of male infertility for eighth pregnancy

- Unknown
- Age
- Environmental
- Tobacco use
- Abnormal sperm production or function
- Cancer/cancer treatment related
- Problems with sperm delivery
- Other

Cause(s) of male infertility for eighth pregnancy-other, specify \_\_\_\_\_

Cause(s) of female infertility for eighth 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 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
- Assisted reproductive technology
- Surgery
- None
- Fertility drugs

Type of assisted reproductive technologies used during eighth pregnancy

- Assisted hatching
- Surgical sperm aspiration
- ICSI
- Other
- IVF

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

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

Date       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 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  NoPatient 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



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

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

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

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

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 complications during eighth 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 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"/> 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                 |

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

Date      

- Urine organic acids - Abn                       Urine organic acids - WNL
- Other

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

- Unknown     ED visits for hyperemesis/IV fluids
- Hospitalizations for hyperemesis/IV fluids    TPN
- Tube feedings                                       Additional medications
- Home lab monitoring                               Increased frequency of lab monitoring
- More intensive fetal monitoring               Bedrest
- Other

Additional interventions required during eighth pregnancy due to this metabolic condition-other, specify

\_\_\_\_\_

Total maternal weight gain (in kg) during eighth pregnancy \_\_\_\_\_

Number of documented episodes of hyperammonemia during eighth pregnancy

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Number of documented episodes of ketonuria during eighth pregnancy

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Number of documented episodes of metabolic acidosis during eighth pregnancy

- 0       1       2       3       4       5       6       7
- 8       9       10       >10       unknown

Treatment(s) for hyperammonemic episodes during eighth pregnancy

- Unknown     No hyperammonemic episodes    Hospital admission
- Protein restriction                               Ammonul                                       Arginine

Maternal protein intake restricted during eighth pregnancy    Unknown    Yes    No

Number of grams of synthetic protein prescribed per day during eighth pregnancy (units = gm/day) \_\_\_\_\_

Number of grams of natural protein prescribed per day during eighth pregnancy (units = gm/day) \_\_\_\_\_

Ammonia scavenger drugs used during eighth pregnancy

- Unknown     No scavengers used       Buphenyl                                       Sodium benzoate

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

Date

Other medications/supplements used during eighth pregnancy

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

Acute health concerns experienced by the patient during delivery for eighth delivery

Abnormal patient lab results during eighth delivery

- Unknown  None  Normal labs during delivery
- Elevated CK  Elevated liver function tests  Hyperammonemia
- Hypoglycemia  Metabolic decompensation  Other

Abnormal patient lab results during eighth delivery-other, specify

Additional maternal interventions during or after eighth 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 eighth delivery related to this disorder-other, specify

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

Date

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

Unknown  Caesarean section (scheduled or non-urgent)

Casesarean section (emergent)  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)

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

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

Date [M][M] [D][D] [Y][Y]

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

Health concern(s) with APGAR score at five minutes < 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

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

Date       OtherReason 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

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

- 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 eighth newborn-other, specify \_\_\_\_\_

Newborn death at or shortly after delivery for eighth newborn  Unknown  Yes  NoNewborn's death related to maternal disorder for eighth newborn  Unknown  Yes  NoEighth 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

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

- 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
- Environmental  Problems with sperm delivery
- Tobacco use  Other

Cause(s) of male infertility for current pregnancy-other, specify \_\_\_\_\_

Cause(s) of female infertility for current 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 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  None
- Assisted reproductive technology  Fertility drugs
- Surgery

Type of assisted reproductive technologies used during current pregnancy

- Assisted hatching  ICSI  IVF
- Surgical sperm aspiration  Other

Type of assisted reproductive technologies used during current pregnancy-other, specify

\_\_\_\_\_

Partner tested for patient's disorder during current pregnancy



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

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

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

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

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

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

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

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

Date

- 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

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          |

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

- |   |   |
|---|---|
| <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 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\_\_\_\_\_

Number of documented episodes of hyperammonemia during current pregnancy

- |                         |                         |                          |                           |                               |                         |                         |                         |
|-------------------------|-------------------------|--------------------------|---------------------------|-------------------------------|-------------------------|-------------------------|-------------------------|
| <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 |                         |                         |                         |

Number of documented episodes of ketonuria during current pregnancy

- |                         |                         |                          |                           |                               |                         |                         |                         |
|-------------------------|-------------------------|--------------------------|---------------------------|-------------------------------|-------------------------|-------------------------|-------------------------|
| <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 |                         |                         |                         |

Number of documented episodes of metabolic acidosis during current pregnancy

- |                         |                         |                          |                           |                               |                         |                         |                         |
|-------------------------|-------------------------|--------------------------|---------------------------|-------------------------------|-------------------------|-------------------------|-------------------------|
| <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 |                         |                         |                         |

Treatment(s) for hyperammonemic episodes during current pregnancy

- |                                  |   |   |
|----------------------------------|---|---|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> No hyperammonemic episodes | <input type="checkbox"/> Hospital admission |
|----------------------------------|---|---|

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

Date

Protein restriction

Ammonul

Arginine

Maternal protein intake restricted during current pregnancy  Unknown  Yes  No

Number of grams of synthetic protein prescribed per day during current pregnancy (units = gm/day)

\_\_\_\_\_

Number of grams of natural protein prescribed per day during current pregnancy (units = gm/day) \_\_\_\_\_

Ammonia scavenger drugs used during current pregnancy

Unknown

No scavengers used

Buphenyl

Sodium benzoate

Other medications/supplements used during current pregnancy

Additional interventions planned for labor/delivery related to patient's disorder for current 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 current pregnancy-other, specify

\_\_\_\_\_

## Comments

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Pregnancy comments

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

Date      **MCD - 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

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 alioisoleucine  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 <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>

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

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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      **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

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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

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      ***MCD - 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 <input type="text" value="MM"/> <input type="text" value="DD"/> <input type="text" value="YY"/>
--

- 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**

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