

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

Date      

## PKU - 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  Amino acid disordersSpecify amino acid disorder diagnosis for the patient  Classic Phenylketonuria (PKU)PKU Diagnosis  Mild phenylketonuria  Moderate phenylketonuria  Classic phenylketonuria  Unknown

Patient disorder identification method

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

Family member with this condition

- Biological mother     Biological father     Full sibling                       Half sibling                       Other

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

## Care and Other Studies

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

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

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

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

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

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

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

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

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

## Education

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Maternal education

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

Paternal education

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

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

## Patient education

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

Special education services received prior to intake  Unknown  Yes  No

## Age patient qualified for special education services

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

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

## Ancestral Origin-Africa

- Unknown  Egypt  Eritrea  Ethiopia  Liberia  Somalia  South Africa
- Other

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

## Ancestral Origin-Asia

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

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

## Ancestral Origin-Europe

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

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

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

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

Ancestral Origin-North America

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

Ancestral Origin-North America-Canada     Unknown     French Canadian

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

Ancestral Origin-South America     Unknown     Colombia     Venezuela     Other

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

Ancestral Origin-Oceania     Unknown     Australia     Other

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

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

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

Race

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

Race-American Indian/Alaskan Native     Aleutian     Cherokee     Other

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

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

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

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

Mother's marital status at patient's birth

- Unknown       Married       Widowed       Divorced       Separated  
 Never married       Living with partner

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

State mother resides in at patient's birth

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

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

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

**Medical Coverage**

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Medical coverage at time of intake

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

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

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

**Language**

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Primary language spoken at home  Unknown  English  Non-English  Both

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

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

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

- Unknown  Yes  No

**Comments**

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Intake demographics comments

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## ***PKU - Intake Family History***

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### **Family History**

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Consanguinity Unknown Yes NoPatient was adopted Unknown Yes No

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

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

### **Siblings**

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

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

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

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

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

Sibling 1: Method of diagnosis

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

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

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

Sibling 1: Results of newborn screening for this condition

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

Sibling 1: Specify the type of diagnostic tests performed

Unknown Biochemical Molecular Imaging Other

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

Sibling 1: Deceased Unknown Yes No

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

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

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

Sibling 2: Affected with this condition Unknown Yes No

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

Date      Sibling 2: Enrolled in this study  Unknown  Yes  No

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

Sibling 2: Method of diagnosis

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

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

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

Sibling 2: Results of newborn screening for this condition

Unavailable  Presumptive positive  Negative/Normal  Borderline

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

Sibling 2: Specify the type of diagnostic tests performed

Unknown  Biochemical  Molecular  Imaging  Other

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

Sibling 2: Deceased  Unknown  Yes  No

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

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

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

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

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

Sibling 3: Method of diagnosis

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

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

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

Sibling 3: Results of newborn screening for this condition

Unavailable  Presumptive positive  Negative/Normal  Borderline

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

Sibling 3: Specify the type of diagnostic tests performed

Unknown  Biochemical  Molecular  Imaging  Other

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

Sibling 3: Deceased  Unknown  Yes  No

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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 \_\_\_\_\_

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Sibling 5: Results of newborn screening for this condition

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

Sibling 5: Specify the type of diagnostic tests performed

 Unknown       Biochemical       Molecular       Imaging       Other

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

Sibling 5: Deceased     Unknown     Yes     No

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

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

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

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

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

Sibling 6: Method of diagnosis

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

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

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

Sibling 6: Results of newborn screening for this condition

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

Sibling 6: Specify the type of diagnostic tests performed

 Unknown       Biochemical       Molecular       Imaging       Other

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

Sibling 6: Deceased     Unknown     Yes     No

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

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

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

Sibling 7: Affected with this condition     Unknown     Yes     No

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

Date      Sibling 7: Enrolled in this study  Unknown  Yes  No

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

Sibling 7: Method of diagnosis

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

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

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

Sibling 7: Results of newborn screening for this condition

 Unavailable  Presumptive positive  Negative/Normal  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

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

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

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

Biological mother: Method of diagnosis

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

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

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

Biological mother: Results of newborn screening for this condition

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

Biological mother: Specify the type of diagnostic tests performed

 Unknown  Biochemical  Molecular  Imaging  Other

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

Biological mother: Deceased  Unknown  Yes  No

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

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

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

Biological father: Method of diagnosis

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

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

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

Biological father: Results of newborn screening for this condition

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

Biological father: Specify the type of diagnostic tests performed

 Unknown  Biochemical  Molecular  Imaging  Other

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

Biological father: Deceased  Unknown  Yes  No

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

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

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

Maternal grandmother: Method of diagnosis

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

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

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

Maternal grandmother: Specify the type of diagnostic tests performed

 Unknown  Biochemical  Molecular  Imaging  Other

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

Maternal grandmother: Deceased  Unknown  Yes  No

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

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

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

Maternal grandfather: Method of diagnosis

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

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

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

Maternal grandfather: Specify the type of diagnostic tests performed

 Unknown  Biochemical  Molecular  Imaging  Other

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

Maternal grandfather: Deceased  Unknown  Yes  No

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

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

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

Paternal grandmother: Method of diagnosis

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

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

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

Paternal grandmother: Specify the type of diagnostic tests performed

- Unknown             Biochemical             Molecular             Imaging             Other

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

Paternal grandmother: Deceased     Unknown     Yes     No

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

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

Paternal grandfather: Affected with this condition     Unknown     Yes     No

Paternal grandfather: Enrolled in this study     Unknown     Yes     No

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

Paternal grandfather: Method of diagnosis

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

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

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

Paternal grandfather: Specify the type of diagnostic tests performed

- Unknown             Biochemical             Molecular             Imaging             Other

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

Paternal grandfather: Deceased     Unknown     Yes     No

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

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

**Other Family**

Other affected family members NOT listed above     Unknown     Yes     No

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

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

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

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

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

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

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

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

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

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

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

**Comments**

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Intake family history comments

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

Date      

## ***PKU - 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

--

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

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

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

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

**Pregnancy**Patient has biological children  Unknown  Yes  No

Please complete the pregnancy form.

Patient is pregnant  Unknown  Yes  No

Please complete the pregnancy form.

**Neonatal History**

Patient's birth was a result of multiple gestation pregnancy

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

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

Congenital anomalies  Unknown  Yes  No

Type of congenital anomalies

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

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

Date      

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

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

Neonatal complications    Unknown    Yes    No

Type of neonatal complications

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

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

Type of neonatal nutrition

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

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

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

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

Birth head circumference units    cm    in

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

Date      

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

Birth length units  cm  in

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

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

---

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

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

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

Full scale IQ obtained prior to intake  Unknown  Yes  NoMost recent full scale IQ date      

Most recent full scale IQ test name \_\_\_\_\_

Most recent full scale IQ number \_\_\_\_\_

Days of age at initiation of phenylalanine restriction \_\_\_\_\_

Days of age (after dietary phenylalanine restriction was initiated) at time first phenylalanine level was documented below 6 mg/dL \_\_\_\_\_

Days of age at time sapropterin dihydrochloride was initiated \_\_\_\_\_

**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

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"/>

Other

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

Comorbidities at time of intake

**Prior testing**

---

Echocardiogram prior to intake    Unknown    Yes    No

Echocardiogram date

Echocardiogram results    Unknown    Normal    Abnormal

Echocardiogram comments

Electrocardiogram prior to intake    Unknown    Yes    No

Electrocardiogram date

Electrocardiogram results    Unknown    Normal    Abnormal

Electrocardiogram comments

Electroencephalography prior to intake    Unknown    Yes    No

Electroencephalography date

Electroencephalography results    Unknown    Normal    Abnormal

Electroencephalography comments

Neurological imaging prior to intake    Unknown    Yes    No

Neurological imaging date

Neurological imaging results    Unknown    Normal    Abnormal

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

Date      

## 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 \_\_\_\_\_

Age (in years) at time of first Dexa scan \_\_\_\_\_

**Phenylalanine Level History**

Average blood phenylalanine level age 0 &lt; 1 year \_\_\_\_\_

Average blood phenylalanine level units age 0 < 1 year   umol/dL   mg/dL   umol/L

Number of phenylalanine levels measured age 0 &lt; 1 year \_\_\_\_\_

Average tyrosine level age 0 &lt; 1 year \_\_\_\_\_

Average tyrosine level units age 0 < 1 year   umol/dL   mg/dL   umol/L

Number of tyrosine levels measured age 0 &lt; 1 year \_\_\_\_\_

Average phenylalanine intake age 0 &lt; 1 year \_\_\_\_\_

Average phenylalanine intake units age 0 < 1 year   mg/day

Average blood phenylalanine level age 1 &lt; 2 years \_\_\_\_\_

Average blood phenylalanine level units age 1 < 2 years   umol/dL   mg/dL   umol/L

Number of phenylalanine levels measured age 1 &lt; 2 years \_\_\_\_\_

Average tyrosine level age 1 &lt; 2 years \_\_\_\_\_

Average tyrosine level units age 1 < 2 years   umol/dL   mg/dL   umol/L

Number of tyrosine levels measured age 1 &lt; 2 years \_\_\_\_\_

Average phenylalanine intake age 1 &lt; 2 years \_\_\_\_\_

Average phenylalanine intake units age 1 < 2 years   mg/day

Average blood phenylalanine level age 2 &lt; 3 years \_\_\_\_\_

Average blood phenylalanine level units age 2 < 3 years   umol/dL   mg/dL   umol/L

Number of phenylalanine levels measured age 2 &lt; 3 years \_\_\_\_\_

Average tyrosine level age 2 &lt; 3 years \_\_\_\_\_

Average tyrosine level units age 2 < 3 years   umol/dL   mg/dL   umol/L

Number of tyrosine levels measured age 2 &lt; 3 years \_\_\_\_\_

Average phenylalanine intake age 2 &lt; 3 years \_\_\_\_\_

Average phenylalanine intake units age 2 < 3 years   mg/day

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

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Average blood phenylalanine level age 3 &lt; 4 years \_\_\_\_\_

Average blood phenylalanine level units age 3 < 4 years    umol/dL    mg/dL    umol/L

Number of phenylalanine levels measured age 3 &lt; 4 years \_\_\_\_\_

Average tyrosine level age 3 &lt; 4 years \_\_\_\_\_

Average tyrosine level units age 3 < 4 years    umol/dL    mg/dL    umol/L

Number of tyrosine levels measured age 3 &lt; 4 years \_\_\_\_\_

Average phenylalanine intake age 3 &lt; 4 years \_\_\_\_\_

Average phenylalanine intake units age 3 < 4 years    mg/day

Average blood phenylalanine level age 4 &lt; 5 years \_\_\_\_\_

Average blood phenylalanine level units age 4 < 5 years    umol/dL    mg/dL    umol/L

Number of phenylalanine levels measured age 4 &lt; 5 years \_\_\_\_\_

Average tyrosine level age 4 &lt; 5 years \_\_\_\_\_

Average tyrosine level units age 4 < 5 years    umol/dL    mg/dL    umol/L

Number of tyrosine levels measured age 4 &lt; 5 years \_\_\_\_\_

Average phenylalanine intake age 4 &lt; 5 years \_\_\_\_\_

Average phenylalanine intake units age 4 < 5 years    mg/day

Average blood phenylalanine level age 5 &lt; 6 years \_\_\_\_\_

Average blood phenylalanine level units age 5 < 6 years    umol/dL    mg/dL    umol/L

Number of phenylalanine levels measured age 5 &lt; 6 years \_\_\_\_\_

Average tyrosine level age 5 &lt; 6 years \_\_\_\_\_

Average tyrosine level units age 5 < 6 years    umol/dL    mg/dL    umol/L

Number of tyrosine levels measured age 5 &lt; 6 years \_\_\_\_\_

Average phenylalanine intake age 5 &lt; 6 years \_\_\_\_\_

Average phenylalanine intake units age 5 < 6 years    mg/day

Average blood phenylalanine level age 6 &lt; 7 years \_\_\_\_\_

Average blood phenylalanine level units age 6 < 7 years    umol/dL    mg/dL    umol/L

Number of phenylalanine levels measured age 6 &lt; 7 years \_\_\_\_\_

Average tyrosine level age 6 &lt; 7 years \_\_\_\_\_

Average tyrosine level units age 6 < 7 years    umol/dL    mg/dL    umol/L

Number of tyrosine levels measured age 6 &lt; 7 years \_\_\_\_\_

Average phenylalanine intake age 6 &lt; 7 years \_\_\_\_\_

Average phenylalanine intake units age 6 < 7 years    mg/day

Average blood phenylalanine level age 7 &lt; 8 years \_\_\_\_\_

Average blood phenylalanine level units age 7 < 8 years    umol/dL    mg/dL    umol/L

Number of phenylalanine levels measured age 7 &lt; 8 years \_\_\_\_\_

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

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Average tyrosine level age 7 &lt; 8 years \_\_\_\_\_

Average tyrosine level units age 7 < 8 years   umol/dL   mg/dL   umol/L

Number of tyrosine levels measured age 7 &lt; 8 years \_\_\_\_\_

Average phenylalanine intake age 7 &lt; 8 years \_\_\_\_\_

Average phenylalanine intake units age 7 < 8 years   mg/day

Average blood phenylalanine level age 8 &lt; 9 years \_\_\_\_\_

Average blood phenylalanine level units age 8 < 9 years   umol/dL   mg/dL   umol/L

Number of phenylalanine levels measured age 8 &lt; 9 years \_\_\_\_\_

Average tyrosine level age 8 &lt; 9 years \_\_\_\_\_

Average tyrosine level units age 8 < 9 years   umol/dL   mg/dL   umol/L

Number of tyrosine levels measured age 8 &lt; 9 years \_\_\_\_\_

Average phenylalanine intake age 8 &lt; 9 years \_\_\_\_\_

Average phenylalanine intake units age 8 < 9 years   mg/day

Average blood phenylalanine level age 9 &lt; 10 years \_\_\_\_\_

Average blood phenylalanine level units age 9 < 10 years   umol/dL   mg/dL   umol/L

Number of phenylalanine levels measured age 9 &lt; 10 years \_\_\_\_\_

Average tyrosine level age 9 &lt; 10 years \_\_\_\_\_

Average tyrosine level units age 9 < 10 years   umol/dL   mg/dL   umol/L

Number of tyrosine levels measured age 9 &lt; 10 years \_\_\_\_\_

Average phenylalanine intake age 9 &lt; 10 years \_\_\_\_\_

Average phenylalanine intake units age 9 < 10 years   mg/day

Average blood phenylalanine level age 10 &lt; 12 years \_\_\_\_\_

Average blood phenylalanine level units age 10 < 12 years   umol/dL   mg/dL   umol/L

Number of phenylalanine levels measured age 10 &lt; 12 years \_\_\_\_\_

Average tyrosine level age 10 &lt; 12 years \_\_\_\_\_

Average tyrosine level units age 10 < 12 years   umol/dL   mg/dL   umol/L

Number of tyrosine levels measured age 10 &lt; 12 years \_\_\_\_\_

Average phenylalanine intake age 10 &lt; 12 years \_\_\_\_\_

Average phenylalanine intake units age 10 < 12 years   mg/day

Average blood phenylalanine level age 12 &lt; 14 years \_\_\_\_\_

Average blood phenylalanine level units age 12 < 14 years   umol/dL   mg/dL   umol/L

Number of phenylalanine levels measured age 12 &lt; 14 years \_\_\_\_\_

Average tyrosine level age 12 &lt; 14 years \_\_\_\_\_

Average tyrosine level units age 12 < 14 years   umol/dL   mg/dL   umol/L

Number of tyrosine levels measured age 12 &lt; 14 years \_\_\_\_\_

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

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Average phenylalanine intake age 12 &lt; 14 years \_\_\_\_\_

Average phenylalanine intake units age 12 < 14 years  mg/day

Average blood phenylalanine level age 14 &lt; 16 years \_\_\_\_\_

Average blood phenylalanine level units age 14 < 16 years  umol/dL  mg/dL  umol/L

Number of phenylalanine levels measured age 14 &lt; 16 years \_\_\_\_\_

Average tyrosine level age 14 &lt; 16 years \_\_\_\_\_

Average tyrosine level units age 14 < 16 years  umol/dL  mg/dL  umol/L

Number of tyrosine levels measured age 14 &lt; 16 years \_\_\_\_\_

Average phenylalanine intake age 14 &lt; 16 years \_\_\_\_\_

Average phenylalanine intake units age 14 < 16 years  mg/day

Average blood phenylalanine level age 16 &lt; 18 years \_\_\_\_\_

Average blood phenylalanine level units age 16 < 18 years  umol/dL  mg/dL  umol/L

Number of phenylalanine levels measured age 16 &lt; 18 years \_\_\_\_\_

Average tyrosine level age 16 &lt; 18 years \_\_\_\_\_

Average tyrosine level units age 16 < 18 years  umol/dL  mg/dL  umol/L

Number of tyrosine levels measured age 16 &lt; 18 years \_\_\_\_\_

Average phenylalanine intake age 16 &lt; 18 years \_\_\_\_\_

Average phenylalanine intake units age 16 < 18 years  mg/day

Average blood phenylalanine level age 18 &lt; 20 years \_\_\_\_\_

Average blood phenylalanine level units age 18 < 20 years  umol/dL  mg/dL  umol/L

Number of phenylalanine levels measured age 18 &lt; 20 years \_\_\_\_\_

Average tyrosine level age 18 &lt; 20 years \_\_\_\_\_

Average tyrosine level units age 18 < 20 years  umol/dL  mg/dL  umol/L

Number of tyrosine levels measured age 18 &lt; 20 years \_\_\_\_\_

Average phenylalanine intake age 18 &lt; 20 years \_\_\_\_\_

Average phenylalanine intake units age 18 < 20 years  mg/day

Average blood phenylalanine level age 20 &lt; 25 years \_\_\_\_\_

Average blood phenylalanine level units age 20 < 25 years  umol/dL  mg/dL  umol/L

Number of phenylalanine levels measured age 20 &lt; 25 years \_\_\_\_\_

Average tyrosine level age 20 &lt; 25 years \_\_\_\_\_

Average tyrosine level units age 20 < 25 years  umol/dL  mg/dL  umol/L

Number of tyrosine levels measured age 20 &lt; 25 years \_\_\_\_\_

Average phenylalanine intake age 20 &lt; 25 years \_\_\_\_\_

Average phenylalanine intake units age 20 < 25 years  mg/day

Average blood phenylalanine level age 25 &lt; 30 years \_\_\_\_\_

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

Date      Average blood phenylalanine level units age 25 < 30 years  umol/dL  mg/dL  umol/L

Number of phenylalanine levels measured age 25 &lt; 30 years \_\_\_\_\_

Average tyrosine level age 25 &lt; 30 years \_\_\_\_\_

Average tyrosine level units age 25 < 30 years  umol/dL  mg/dL  umol/L

Number of tyrosine levels measured age 25 &lt; 30 years \_\_\_\_\_

Average phenylalanine intake age 25 &lt; 30 years \_\_\_\_\_

Average phenylalanine intake units age 25 < 30 years  mg/day

Average blood phenylalanine level age 30 &lt; 35 years \_\_\_\_\_

Average blood phenylalanine level units age 30 < 35 years  umol/dL  mg/dL  umol/L

Number of phenylalanine levels measured age 30 &lt; 35 years \_\_\_\_\_

Average tyrosine level age 30 &lt; 35 years \_\_\_\_\_

Average tyrosine level units age 30 < 35 years  umol/dL  mg/dL  umol/L

Number of tyrosine levels measured age 30 &lt; 35 years \_\_\_\_\_

Average phenylalanine intake age 30 &lt; 35 years \_\_\_\_\_

Average phenylalanine intake units age 30 < 35 years  mg/day

Average blood phenylalanine level age 35 &lt; 40 years \_\_\_\_\_

Average blood phenylalanine level units age 35 < 40 years  umol/dL  mg/dL  umol/L

Number of phenylalanine levels measured age 35 &lt; 40 years \_\_\_\_\_

Average tyrosine level age 35 &lt; 40 years \_\_\_\_\_

Average tyrosine level units age 35 < 40 years  umol/dL  mg/dL  umol/L

Number of tyrosine levels measured age 35 &lt; 40 years \_\_\_\_\_

Average phenylalanine intake age 35 &lt; 40 years \_\_\_\_\_

Average phenylalanine intake units age 35 < 40 years  mg/day

Average blood phenylalanine level age 40 &lt; 50 years \_\_\_\_\_

Average blood phenylalanine level units age 40 < 50 years  umol/dL  mg/dL  umol/L

Number of phenylalanine levels measured age 40 &lt; 50 years \_\_\_\_\_

Average tyrosine level age 40 &lt; 50 years \_\_\_\_\_

Average tyrosine level units age 40 < 50 years  umol/dL  mg/dL  umol/L

Number of tyrosine levels measured age 40 &lt; 50 years \_\_\_\_\_

Average phenylalanine intake age 40 &lt; 50 years \_\_\_\_\_

Average phenylalanine intake units age 40 < 50 years  mg/day

Average blood phenylalanine level age &gt; 50 years \_\_\_\_\_

Average blood phenylalanine level units age > 50 years  umol/dL  mg/dL  umol/L

Number of phenylalanine levels measured age &gt; 50 years \_\_\_\_\_

Average tyrosine level age &gt; 50 years \_\_\_\_\_

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

Date

Average tyrosine level units age > 50 years umol/dL mg/dL umol/L

Number of tyrosine levels measured age > 50 years\_\_\_\_\_

Average phenylalanine intake age > 50 years\_\_\_\_\_

Average phenylalanine intake units age > 50 years mg/day

Average blood phenylalanine level for current year of life through intake\_\_\_\_\_

Average blood phenylalanine level units for current year of life through intake

umol/dL mg/dL umol/L

Number of phenylalanine levels measured for current year of life through intake\_\_\_\_\_

Average tyrosine level for current year of life through intake\_\_\_\_\_

Average tyrosine level units for current year of life through intake umol/dL mg/dL umol/L

Number of tyrosine levels measured for current year of life through intake\_\_\_\_\_

Average phenylalanine intake for current year of life through intake\_\_\_\_\_

Average phenylalanine intake units for current year of life through intake mg/day

**Emergency Management**

---

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

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

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

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

Patient/primary caregiver was given the 24 hour on-call contact information for a specialty provider

Unknown Yes No

**Comments**

---

Intake past health history comments

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

Date      

## PKU - 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

Phenylalanine on first newborn screen \_\_\_\_\_

Phenylalanine on first newborn screen units  umol/L

Phenylalanine/Tyrosine on first newborn screen \_\_\_\_\_

Tyrosine on first newborn screen \_\_\_\_\_

Tyrosine on first newborn screen units  umol/L

Other result/report on first newborn screen

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

Date second newborn screen collected      

Reason for second newborn screen

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

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

Phenylalanine on second newborn screen \_\_\_\_\_

Phenylalanine on second newborn screen units  umol/L

Phenylalanine/Tyrosine on second newborn screen \_\_\_\_\_

Tyrosine on second newborn screen \_\_\_\_\_

Tyrosine on second newborn screen units  umol/L

Other result/report on second newborn screen



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

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

Left ear: Diagnosis: Type of hearing loss

- |  |   |
|--|---|
| <input type="checkbox"/> Sensorineural | <input type="checkbox"/> Conductive                                 |
| <input type="checkbox"/> Mixed         | <input type="checkbox"/> Auditory neuropathy/Auditory dys-synchrony |
| <input type="checkbox"/> Other         |   |

**Comments**

---

Intake newborn screening comments

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

Date      

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

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

### Diagnostic Testing

---

Diagnostic labs performed

- |  |   |
|--|---|
| <input type="checkbox"/> Unknown                   | <input type="checkbox"/> None   |
| <input type="checkbox"/> Blood spot phenylalanine  | <input type="checkbox"/> Blood spot tyrosine                            |
| <input type="checkbox"/> Plasma amino acid profile | <input type="checkbox"/> Plasma phenylalanine                           |
| <input type="checkbox"/> Plasma tyrosine           | <input type="checkbox"/> RBC dihydropteridine reductase (DHPR) activity |
| <input type="checkbox"/> Urine organic acids       | <input type="checkbox"/> Urine pterins                                  |
| <input type="checkbox"/> Other                     |   |

Blood spot phenylalanine     Unknown     Within normal limits     Abnormal

Blood spot phenylalanine significance     Diagnostic     Non-diagnostic

Blood spot phenylalanine value \_\_\_\_\_

Blood spot phenylalanine units

umol/L     umol/dL     umol/mL     nmol/L     nmol/dL     nmol/mL     mmol/L

mmol/dL     mmol/mL     mg/dL     g/dL     ug/dL

Blood spot phenylalanine reference range \_\_\_\_\_

Blood spot tyrosine     Unknown     Within normal limits     Abnormal

Blood spot tyrosine significance     Diagnostic     Non-diagnostic

Blood spot tyrosine value \_\_\_\_\_

Blood spot tyrosine units     umol/dL     umol/L     mg/dL

Blood spot tyrosine reference range \_\_\_\_\_

Plasma amino acid profile     Unknown     Within normal limits     Abnormal

Plasma amino acid profile significance     Diagnostic     Non-diagnostic

Plasma amino acid comments

Plasma phenylalanine     Unknown     Within normal limits     Abnormal

Plasma phenylalanine significance     Diagnostic     Non-diagnostic

Plasma phenylalanine value \_\_\_\_\_

Plasma phenylalanine units     umol/dL     umol/L     mg/dL

Plasma phenylalanine reference range \_\_\_\_\_

Plasma tyrosine     Unknown     Within normal limits     Abnormal

Plasma tyrosine significance     Diagnostic     Non-diagnostic

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

Date      

Plasma tyrosine value \_\_\_\_\_

Plasma tyrosine units  umol/dL  umol/L  mg/dL

Plasma tyrosine reference range \_\_\_\_\_

RBC dihydropteridine reductase (DHPR) activity  Unknown  Within normal limits  AbnormalRBC dihydropteridine reductase (DHPR) activity significance  Diagnostic  Non-diagnostic

RBC dihydropteridine reductase (DHPR) activity value \_\_\_\_\_

RBC dihydropteridine reductase (DHPR) activity units  nmol/min/mg Hb  uU/g Hb

RBC dihydropteridine reductase (DHPR) activity reference range \_\_\_\_\_

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

Urine organic acid comments

--

Urine pterins  Biopterin  Neopterin  PrimapterinUrine biopterin-number of samples  1  2  3

Urine biopterin percentage \_\_\_\_\_

Urine biopterin value \_\_\_\_\_

Urine biopterin units  % biopterin  mmol/mol Cr  nmol/L  ug/L

Urine biopterin reference range \_\_\_\_\_

Urine biopterin percentage \_\_\_\_\_

Urine biopterin value \_\_\_\_\_

Urine biopterin units  % biopterin  mmol/mol Cr  nmol/L  ug/L

Urine biopterin reference range \_\_\_\_\_

Urine biopterin percentage \_\_\_\_\_

Urine biopterin value \_\_\_\_\_

Urine biopterin units  % biopterin  mmol/mol Cr  nmol/L  ug/L

Urine biopterin reference range \_\_\_\_\_

Urine neopterin-number of samples  1  2  3

Urine neopterin percentage \_\_\_\_\_

Urine neopterin value \_\_\_\_\_

Urine neopterin units  nmol/mol Cr  nmol/L

Urine neopterin reference range \_\_\_\_\_

Urine neopterin percentage \_\_\_\_\_

Urine neopterin value \_\_\_\_\_

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

Urine neopterin units  nmol/mol Cr  nmol/L

Urine neopterin reference range \_\_\_\_\_

Urine neopterin percentage \_\_\_\_\_

Urine neopterin value \_\_\_\_\_

Urine neopterin units  nmol/mol Cr  nmol/L

Urine neopterin reference range \_\_\_\_\_

Urine primapterin-number of samples  1  2  3

Urine primapterin percentage \_\_\_\_\_

Urine primapterin value \_\_\_\_\_

Urine primapterin units  nmol/L

Urine primapterin reference range \_\_\_\_\_

Urine primapterin percentage \_\_\_\_\_

Urine primapterin value \_\_\_\_\_

Urine primapterin units  nmol/L

Urine primapterin reference range \_\_\_\_\_

Urine primapterin percentage \_\_\_\_\_

Urine primapterin value \_\_\_\_\_

Urine primapterin units  nmol/L

Urine primapterin reference range \_\_\_\_\_

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

Other diagnostic lab  Unknown  Within normal limits  Abnormal

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

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

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

Reason genotyping was done

- Unknown                     
  Diagnosis confirmation  
  Modifier genes                     
  Pharmacogenetics  
 Other

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

Reason genotyping was not done

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

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

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

Gene(s) associated with PKU     PAH     Other

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

PAH: Specify allele 1\_\_\_\_\_

PAH: Specify allele 2\_\_\_\_\_

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

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

Maternal genetic testing done     Unknown     Yes     No     Genotyping in progress

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

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

Paternal genetic testing done     Unknown     Yes     No     Genotyping in progress

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

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

**Comments**

---

Initial testing comments

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

Date      

## PKU - 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      

## PKU - Visit Health History

### Health Status

#### Immunization status

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

Reason immunization status not up to date

#### Current comorbidities

Patient has had seizures since last visit  Unknown  Yes  No

### 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 \_\_\_\_\_

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

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

Date of sick visit 2

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

Date

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

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

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"/>

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 \_\_\_\_\_

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

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

Date of sick visit 7

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

Date      Reason for sick visit 7  Unknown  Condition related  Condition unrelatedSick 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  OtherPatient 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 unrelatedSick 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 OtherPatient 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 unrelatedSick visit 9 was a condition exacerbation  Unknown  Yes  No

Location for sick visit 9

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

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"/>
--

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

Surgeries since last visit     Unknown     Yes     No

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

Surgery complications     Unknown     Yes     No

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

Date

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" 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"/>

# PKU - 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 atopic dermatitis     Evidence of tremor     Other

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

## Comments

Visit findings comments

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

Date      ***PKU - Visit Ancillary Care*****Care Coordination**Missed subspecialty visits since last visit  Unknown  Yes  No

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

Other health services currently received  Unknown  Yes  No

Specify other current health services

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

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

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

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

Preventive care status

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

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

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

Community resources currently received  Unknown  Yes  No

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

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    No

Standardized developmental screening tool(s) used    Unknown    Yes    No

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

Date

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

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

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

\_\_\_\_\_

Neuropsychometric evaluation performed since last visit  Unknown  Yes  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

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

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

\_\_\_\_\_

Behavioral concerns  Unknown  Yes  No

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

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      

## PKU - Visit Lab Studies

### Biochemical Labs

Biochemical labs associated with this visit

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> None                            |
| <input type="checkbox"/> Dihydropteridine reductase (DHPR)   | <input type="checkbox"/> Folic acid                      |
| <input type="checkbox"/> Isoleucine                          | <input type="checkbox"/> Leucine                         |
| <input type="checkbox"/> Phenylalanine                       | <input type="checkbox"/> Plasma amino acid profile       |
| <input type="checkbox"/> Plasma essential fatty acid profile | <input type="checkbox"/> Plasma total homocysteine level |
| <input type="checkbox"/> Selenium                            | <input type="checkbox"/> Serum vitamin B12 level         |
| <input type="checkbox"/> Total 25-hydroxyvitamin D           | <input type="checkbox"/> Tyrosine                        |
| <input type="checkbox"/> Urine methylmalonic acid (MMA)      | <input type="checkbox"/> Urine organic acids             |
| <input type="checkbox"/> Urine pterins                       | <input type="checkbox"/> Valine                          |
| <input type="checkbox"/> Other                               |  |

Dihydropteridine reductase (DHPR)

- Within normal limits    Abnormal    In progress    Results unavailable

Dihydropteridine reductase (DHPR) value \_\_\_\_\_

Dihydropteridine reductase (DHPR) units    nmol/min/mg Hb    uU/g Hb

Dihydropteridine reductase (DHPR) reference range \_\_\_\_\_

Folic acid    Within normal limits    Abnormal    In progress    Results unavailable

Folic acid value \_\_\_\_\_

Folic acid units    nmol/L    ng/mL

Folic acid reference range \_\_\_\_\_

Isoleucine    Within normal limits    Abnormal    In progress    Results unavailable

Isoleucine value \_\_\_\_\_

Isoleucine units    umol/dL    umol/L    mg/dL

Isoleucine reference range \_\_\_\_\_

Leucine    Within normal limits    Abnormal    In progress    Results unavailable

Leucine value \_\_\_\_\_

Leucine units    umol/dL    umol/L    mg/dL

Leucine reference range \_\_\_\_\_

Phenylalanine    Within normal limits    Abnormal    In progress    Results unavailable

Phenylalanine value \_\_\_\_\_

Phenylalanine units    umol/dL    umol/L    mg/dL

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

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Phenylalanine reference range \_\_\_\_\_

Plasma amino acid profile  Within normal limits  Abnormal  In progress  Results unavailable

Plasma amino acid comments

Plasma essential fatty acid profile

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

Alpha linolenic acid  Within normal limits  Abnormal  In progress  Results unavailable

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

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

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

Docosahexaenoic acid (DHA) acid

 Within normal limits  Abnormal  In progress  Results unavailable

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

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

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

EPA  Within normal limits  Abnormal  In progress  Results unavailable

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

EPA units  nmol/L  ng/mL  nmol/mL

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

Linoleic acid  Within normal limits  Abnormal  In progress  Results unavailable

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

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

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

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

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

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

Total fatty acids  Within normal limits  Abnormal  In progress  Results unavailable

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

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

Other plasma essential fatty acid result

 Within normal limits  Abnormal  In progress  Results unavailable

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

Date      

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

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

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

Other plasma essential fatty acid comments

Plasma total homocysteine level

 Within normal limits    Abnormal    In progress    Results unavailable

Plasma total homocysteine level value \_\_\_\_\_

Plasma total homocysteine level units    umol/L    mg/L

Plasma total homocysteine level reference range \_\_\_\_\_

Selenium    Within normal limits    Abnormal    In progress    Results unavailable

Selenium value \_\_\_\_\_

Selenium units    umol/L    ug/L

Selenium reference range \_\_\_\_\_

Serum vitamin B12 level    Within normal limits    Abnormal    In progress    Results unavailable

Serum vitamin B12 level value \_\_\_\_\_

Serum vitamin B12 level units    pmol/L    pg/mL    ng/mL

Serum vitamin B12 level reference range \_\_\_\_\_

Total 25-hydroxyvitamin D    Within normal limits    Abnormal    In progress    Results unavailable

Total 25-hydroxyvitamin D value \_\_\_\_\_

Total 25-hydroxyvitamin D units    nmol/L    ug/L    ng/mL

Total 25-hydroxyvitamin D reference range \_\_\_\_\_

Tyrosine    Within normal limits    Abnormal    In progress    Results unavailable

Tyrosine value \_\_\_\_\_

Tyrosine units    umol/dL    umol/L    mg/dL

Tyrosine reference range \_\_\_\_\_

Urine methylmalonic acid level

 Within normal limits    Abnormal    In progress    Results unavailable

Urine methylmalonic acid level value \_\_\_\_\_

Urine methylmalonic acid level units    mmol/mol Cr    ug/mg Cr

Urine methylmalonic acid level reference range \_\_\_\_\_

Urine organic acids    Within normal limits    Abnormal    In progress    Results unavailable

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

Date

Urine organic acid comments

Urine pterins   Biopterin   Neopterin   Primapterin

Urine biopterin-number of samples    1    2    3

Urine biopterin percentage\_\_\_\_\_

Urine biopterin value\_\_\_\_\_

Urine biopterin units   % biopterin   mmol/mol Cr   nmol/L   ug/L

Urine biopterin percentage\_\_\_\_\_

Urine biopterin value\_\_\_\_\_

Urine biopterin units   % biopterin   mmol/mol Cr   nmol/L   ug/L

Urine biopterin percentage\_\_\_\_\_

Urine biopterin value\_\_\_\_\_

Urine biopterin units   % biopterin   mmol/mol Cr   nmol/L   ug/L

Urine neopterin-number of samples    1    2    3

Urine neopterin percentage\_\_\_\_\_

Urine neopterin value\_\_\_\_\_

Urine neopterin units   nmol/mol Cr   nmol/L

Urine neopterin percentage\_\_\_\_\_

Urine neopterin value\_\_\_\_\_

Urine neopterin units   nmol/mol Cr   nmol/L

Urine neopterin percentage\_\_\_\_\_

Urine neopterin value\_\_\_\_\_

Urine neopterin units   nmol/mol Cr   nmol/L

Urine primapterin-number of samples    1    2    3

Urine primapterin percentage\_\_\_\_\_

Urine primapterin value\_\_\_\_\_

Urine primapterin units   nmol/L

Urine primapterin percentage\_\_\_\_\_

Urine primapterin value\_\_\_\_\_

Urine primapterin units   nmol/L

Urine primapterin percentage\_\_\_\_\_

Urine primapterin value\_\_\_\_\_

Urine primapterin units   nmol/L

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

Date

Valine  Within normal limits  Abnormal  In progress  Results unavailable

Valine value \_\_\_\_\_

Valine units  umol/dL  umol/L  mg/dL

Valine reference range \_\_\_\_\_

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

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Chemistry labs associated with this visit

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

Arterial blood gas  Within normal limits  Abnormal  In progress  Results unavailable

Arterial blood gas comments

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

Arterial blood gas units  mmHg  mEq/L

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

Anion gap  Within normal limits  Abnormal  In progress  Results unavailable

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

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

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

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

Date      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 \_\_\_\_\_

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

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

Date

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

Other chemistry lab comments

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

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

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

### Hematology Labs

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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 \_\_\_\_\_

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

Date      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 \_\_\_\_\_

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 \_\_\_\_\_

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

Date      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 transpeptidase | <input type="checkbox"/> Globulin                   |
| <input type="checkbox"/> Prealbumin           | <input type="checkbox"/> Total bilirubin               | <input type="checkbox"/> Total protein              |
| <input type="checkbox"/> Other                |  |   |

Albumin  Within normal limits  Abnormal  In progress  Results unavailable

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

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

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

Alkaline phosphatase  Within normal limits  Abnormal  In progress  Results unavailable

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

Alkaline phosphatase units  U/L  ukat/L

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

Alanine aminotransferase  Within normal limits  Abnormal  In progress  Results unavailable

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

Alanine aminotransferase units  U/L  ukat/L

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

Aspartate aminotransferase  Within normal limits  Abnormal  In progress  Results unavailable

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

Aspartate aminotransferase units  U/L  ukat/L

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

Date

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

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 \_\_\_\_\_

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

Date

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

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

### Renal Labs

---

Renal labs associated with this visit

- Unknown  None
- Blood urea nitrogen  Serum creatinine
- 24-hour creatinine clearance  Nuclear medicine glomerular filtration rate
- Other

Blood urea nitrogen     Within normal limits     Abnormal     In progress     Results unavailable

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

Blood urea nitrogen units     mmol/L     mg/dL

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

Serum creatinine     Within normal limits     Abnormal     In progress     Results unavailable

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

Serum creatinine units     umol/L     mg/dL

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

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

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

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

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

Nuclear medicine glomerular filtration rate

Within normal limits     Abnormal     In progress     Results unavailable

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

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

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

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

Other renal labs     Within normal limits     Abnormal     In progress     Results unavailable

Other renal lab comments

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

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

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

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

Date      **Miscellaneous Labs**

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

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 \_\_\_\_\_

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

Date

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

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      

## PKU - Visit Studies Other

### Home Monitoring

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

Type of home monitoring

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

#### Glucose

Frequency of glucose home monitoring  Unknown  Only when symptomatic  Routinely

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

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

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

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

 mmol/L  mg/dL

#### Branched Chain Amino Acids

Type of branched chain amino acid home monitoring

- Unknown  Alloisoleucine  Isoleucine  Leucine  Valine

Sample type of branched chain amino acid home monitoring

- Unknown  Filter paper blood  Plasma

Frequency of branched chain amino acid home monitoring

- Unknown  Only when symptomatic  Routinely

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

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

#### Alloisoleucine

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

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

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

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

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

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

Date      

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

**Isoleucine**

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

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

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

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

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

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

**Leucine**

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

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

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

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

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

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

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

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

**Valine**

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Date      

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

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

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

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

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

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

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

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

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

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

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

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

**Dinitrophenylhydrazine**

Frequency of dinitrophenylhydrazine home monitoring

 Unknown  Only when symptomatic  Routinely

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

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

Result of dinitrophenylhydrazine home monitoring samples

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

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

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

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

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

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

**Other**

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

Frequency of other home monitoring  Unknown  Only when symptomatic  Routinely

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

Date      

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

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

**Laboratory Monitoring**

---

Laboratory monitoring other than home monitoring done since last visit

 Unknown                       No                                       Phenylalanine                       Tyrosine**Phenylalanine**Sample type of phenylalanine laboratory monitoring     Unknown     Filter paper blood     Plasma

Frequency of phenylalanine laboratory monitoring

 Unknown                       Only when symptomatic                       Routinely

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

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

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

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

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

Average phenylalanine on laboratory monitoring samples \_\_\_\_\_

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

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

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

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

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

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

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

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

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

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

Average tyrosine on laboratory monitoring samples \_\_\_\_\_

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

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

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

**Other Studies**

---

Physiological tests associated with this visit

 Unknown                       None                                       Electrocardiogram (ECG) Electroencephalography (EEG)     Electromyography (EMG)                       Holter monitoring

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

Date

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

Cardiac stress test findings of note

**Imaging Studies**

Imaging studies associated with this visit

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

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

Date    |    |

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
- 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

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"/>

- 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

### Dexa Scan

---

Dexa scan(s) associated with this visit    Unknown    Yes    No

Number of Dexa scans performed    1    2

First Dexa scan sites

- |                                  |                                |                                     |  |
|----------------------------------|--------------------------------|-------------------------------------|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Heel  | <input type="checkbox"/> Hip        | <input type="checkbox"/> Pelvis                |
| <input type="checkbox"/> Spine   | <input type="checkbox"/> Wrist | <input type="checkbox"/> Total body | <input type="checkbox"/> Total body minus head |
| <input type="checkbox"/> Other   |                                |                                     |  |

First Dexa scan: Heel result    Unknown    Abnormal    Normal

First Dexa scan: Heel Zscore \_\_\_\_\_

First Dexa scan: Hip result    Unknown    Abnormal    Normal

First Dexa scan: Hip Zscore \_\_\_\_\_

First Dexa scan: Pelvis result    Unknown    Abnormal    Normal

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

First Dexa scan: Pelvis Zscore\_\_\_\_\_

First Dexa scan: Spine result    Unknown    Abnormal    Normal

First Dexa scan: Spine Zscore\_\_\_\_\_

First Dexa scan: Wrist result    Unknown    Abnormal    Normal

First Dexa scan: Wrist Zscore\_\_\_\_\_

First Dexa scan: Total body result    Unknown    Abnormal    Normal

First Dexa scan: Total body Zscore\_\_\_\_\_

First Dexa scan: Total body minus head result    Unknown    Abnormal    Normal

First Dexa scan: Total body minus head Zscore\_\_\_\_\_

First Dexa scan site-other, specify\_\_\_\_\_

First Dexa scan: Other site result    Unknown    Abnormal    Normal

First Dexa scan: Other site Zscore\_\_\_\_\_

Second Dexa scan sites

- |                                  |                                |                                     |  |
|----------------------------------|--------------------------------|-------------------------------------|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Heel  | <input type="checkbox"/> Hip        | <input type="checkbox"/> Pelvis                |
| <input type="checkbox"/> Spine   | <input type="checkbox"/> Wrist | <input type="checkbox"/> Total body | <input type="checkbox"/> Total body minus head |
| <input type="checkbox"/> Other   |                                |                                     |  |

Second Dexa scan: Heel result    Unknown    Abnormal    Normal

Second Dexa scan: Heel Zscore\_\_\_\_\_

Second Dexa scan: Hip result    Unknown    Abnormal    Normal

Second Dexa scan: Hip Zscore\_\_\_\_\_

Second Dexa scan: Pelvis result    Unknown    Abnormal    Normal

Second Dexa scan: Pelvis Zscore\_\_\_\_\_

Second Dexa scan: Spine result    Unknown    Abnormal    Normal

Second Dexa scan: Spine Zscore\_\_\_\_\_

Second Dexa scan: Wrist result    Unknown    Abnormal    Normal

Second Dexa scan: Wrist Zscore\_\_\_\_\_

Second Dexa scan: Total body result    Unknown    Abnormal    Normal

Second Dexa scan: Total body Zscore\_\_\_\_\_

Second Dexa scan: Total body minus head result    Unknown    Abnormal    Normal

Second Dexa scan: Total body minus head Zscore\_\_\_\_\_

Second Dexa scan site-other, specify\_\_\_\_\_

Second Dexa scan: Other site result    Unknown    Abnormal    Normal

Second Dexa scan: Other site Zscore\_\_\_\_\_

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

**Comments**

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Visit studies-other comments

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

Date      

## ***PKU - Visit Management And Treatment Pharmacotherapy***

### **Pharmacotherapy**

#### Medications

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

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

Homeopathic therapies  Unknown  Yes  No

Specify homeopathic therapies

#### **Disease Treatment**

Treatment recommended/prescribed

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                          | <input type="checkbox"/> None                        |
| <input type="checkbox"/> Large neutral amino acids (LNAA) | <input type="checkbox"/> Sapropterin dihydrochloride |
| <input type="checkbox"/> Tyrosine                         | <input type="checkbox"/> Other                       |

#### **Large Neutral Amino Acids**

LNAA route recommended/prescribed  Unknown  Feeding tube  IV  Oral

LNAA dose recommended/prescribed \_\_\_\_\_

LNAA dose recommended/prescribed units  g  mg

LNAA frequency recommended/prescribed

 Unknown  Once/day  Twice/day  Three times/day  Four times/day

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

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

 Other

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

LNAA taken as recommended/prescribed  Unknown  Yes  No

Actual LNAA dose reported \_\_\_\_\_

Actual LNAA dose reported units  g  mg

Actual LNAA frequency reported

 Unknown  Once/day  Twice/day  Three times/day  Four times/day Other

Actual LNAA frequency reported-other, specify \_\_\_\_\_

Reason LNAA is not taken as recommended/prescribed \_\_\_\_\_

Method of payment for LNAA

 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**Sapropterin dihydrochloride**

Neuropsychological evaluation performed before prescribing sapropterin dihydrochloride

 Unknown  Yes  No

Neuropsychological evaluation performed while taking sapropterin dihydrochloride

 Unknown  Yes  No

Sapropterin dihydrochloride route recommended/prescribed

 Unknown  Feeding tube  Oral-dissolved  Oral-tablet

Sapropterin dihydrochloride dose recommended/prescribed \_\_\_\_\_

Sapropterin dihydrochloride dose recommended/prescribed units  g  mgSapropterin dihydrochloride frequency recommended/prescribed  Unknown  Once/day  Other

Sapropterin dihydrochloride frequency recommended/prescribed-other, specify

\_\_\_\_\_

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

Sapropterin dihydrochloride taken as recommended/prescribed  Unknown  Yes  No

Actual sapropterin dihydrochloride dose reported \_\_\_\_\_

Actual sapropterin dihydrochloride dose reported units  g  mg

Actual sapropterin dihydrochloride frequency reported  Unknown  Once/day  Other

Actual sapropterin dihydrochloride frequency reported-other, specify \_\_\_\_\_

Reason sapropterin dihydrochloride is not taken as recommended/prescribed \_\_\_\_\_

Criterion used to determine sapropterin dihydrochloride responsiveness

- Unknown
- > 20% decrease in phenylalanine level post-medication
- > 30% decrease in phenylalanine level post-medication
- Improved academic performance
- Improved attention
- Improved behavior
- Improved mood stability
- Increased dietary phenylalanine tolerance
- Other

Criterion used to determine sapropterin dihydrochloride responsiveness-Other, specify

\_\_\_\_\_

Method of payment for sapropterin dihydrochloride

- 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

**Tyrosine**

Tyrosine route recommended/prescribed  Unknown  Feeding tube  Oral

Tyrosine dose recommended/prescribed \_\_\_\_\_

Tyrosine dose recommended/prescribed units  g  mg

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

Tyrosine frequency recommended/prescribed

- Unknown
- Once/day
- Twice/day
- Three times/day
- Four times/day
- Other

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

Tyrosine taken as recommended/prescribed  Unknown  Yes  No

Actual tyrosine dose reported \_\_\_\_\_

Actual tyrosine dose reported units  g  mg

Actual tyrosine frequency reported

- Unknown
- Once/day
- Twice/day
- Three times/day
- Four times/day
- Other

Actual tyrosine frequency reported-other, specify \_\_\_\_\_

Reason tyrosine is not taken as recommended/prescribed \_\_\_\_\_

Method of payment for tyrosine

- 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 \_\_\_\_\_

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"/>

Reason other treatment is not taken as recommended/prescribed \_\_\_\_\_

Method of payment for other treatment

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

**Comments**

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Medication and supplement comments

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

Date      

## ***PKU - 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 <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"/>
---

- 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 \_\_\_\_\_

**Phenylalanine Restricted Diet**

Phenylalanine restricted diet recommended/prescribed  Unknown  Yes  No

Amount of phenylalanine recommended/prescribed per day (not including metabolic formula)  
\_\_\_\_\_

Amount of phenylalanine recommended/prescribed per day (not including metabolic formula) units  
 mg

Average phenylalanine intake per day reported since last outpatient visit \_\_\_\_\_

Average phenylalanine intake per day reported since last outpatient visit units  mg

**Other**

Average protein intake (grams per day) reported since last outpatient metabolic visit \_\_\_\_\_

Other amino acid supplements recommended/prescribed  Unknown  Yes  No

Name(s) of other amino acid supplement(s) recommended/prescribed \_\_\_\_\_

Formulations of other amino acid supplement(s) recommended/prescribed

- Unknown       Bar               Capsules               Powder/Mix-in       Tablets
- Other

Formulations of other amino acid supplement(s) recommended/prescribed-other, specify  
\_\_\_\_\_

Amount of other amino acid supplement(s) recommended/prescribed \_\_\_\_\_

Amount of other amino acid supplement(s) recommended/prescribed units \_\_\_\_\_

Other amino acid supplement(s) frequency recommended/prescribed \_\_\_\_\_

**Comments**

Nutrition comments

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

Date        ***PKU - 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

Specify other non-IBEMC participating center \_\_\_\_\_

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

- Declined consent at age of majority or emancipation
- Subject initiated withdrawal
- Investigator initiated withdrawal

Specify reason subject initiated withdrawal, if known

Specify reason investigator initiated withdrawal, if known

Second date of study status change

Condition follow-up status    Active    Inactive

Reason for inactive status

- Unknown
- Moved
- Subject withdrawal from study
- Deceased
- Refused follow-up
- Lost to follow-up
- Follow-up deemed unnecessary

Date of death

Age of death (in years) \_\_\_\_\_

Moved-specify

- Moved to another IBEMC participating center
- Moved to another non-IBEMC participating center
- Moved-condition follow-up status unknown

Specify other IBEMC participating center

- IL-Ann & Robert H. Lurie Children's Hospital of Chicago

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

- IL-University of Illinois
- IN- Riley Hospital for Children Indiana University Health
- KY-University of Louisville
- MI-University of Michigan
- MI-Wayne State University Children's Hospital of Michigan
- MN-University of Minnesota
- MO-University of Missouri
- NE-University of Nebraska
- NJ-Hackensack University
- NY-University of Rochester
- NY-Women's & Children's Hospital of Buffalo
- OH-Cincinnati Children's Hospital
- OH-Nationwide Children's Hospital
- OK-Saint Francis Hospital
- OK-University of Oklahoma
- PA-Children's Hospital of Pittsburgh
- SD-Sanford Children's Specialty Clinic
- WI-University of Wisconsin
- WI-Medical College of Wisconsin

Specify other non-IBEMC participating center \_\_\_\_\_

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

- Declined consent at age of majority or emancipation
- Subject initiated withdrawal
- Investigator initiated withdrawal



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

Date

- OK-Saint Francis Hospital
- OK-University of Oklahoma
- PA-Children's Hospital of Pittsburgh
- SD-Sanford Children's Specialty Clinic
- WI-University of Wisconsin
- WI-Medical College of Wisconsin

Specify other non-IBEMC participating center \_\_\_\_\_

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

- Declined consent at age of majority or emancipation
- Subject initiated withdrawal
- Investigator initiated withdrawal

Specify reason subject initiated withdrawal, if known

Specify reason investigator initiated withdrawal, if known

Fourth date of study status change

Condition follow-up status    Active    Inactive

Reason for inactive status

- Unknown
- Deceased
- Lost to follow-up
- Moved
- Refused follow-up
- Follow-up deemed unnecessary
- Subject withdrawal from study

Date of death

Age of death (in years) \_\_\_\_\_

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

Moved-specify

- Moved to another IBEMC participating center
- Moved to another non-IBEMC participating center
- Moved-condition follow-up status unknown

Specify other IBEMC participating center

- IL-Ann & Robert H. Lurie Children's Hospital of Chicago
- IL-University of Illinois
- IN- Riley Hospital for Children Indiana University Health
- KY-University of Louisville
- MI-University of Michigan
- MI-Wayne State University Children's Hospital of Michigan
- MN-University of Minnesota
- MO-University of Missouri
- NE-University of Nebraska
- NJ-Hackensack University
- NY-University of Rochester
- NY-Women's & Children's Hospital of Buffalo
- OH-Cincinnati Children's Hospital
- OH-Nationwide Children's Hospital
- OK-Saint Francis Hospital
- OK-University of Oklahoma
- PA-Children's Hospital of Pittsburgh
- SD-Sanford Children's Specialty Clinic
- WI-University of Wisconsin
- WI-Medical College of Wisconsin

Specify other non-IBEMC participating center \_\_\_\_\_

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

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

Date

Declined consent at age of majority or emancipation

Subject initiated withdrawal

Investigator initiated withdrawal

Specify reason subject initiated withdrawal, if known

Specify reason investigator initiated withdrawal, if known

### Comments

---

Study status comments

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

Date      

## PKU - Pregnancy

---

Patient has had one or more pregnancies  Unknown  Yes  No

Number of pregnancies \_\_\_\_\_

Number of term pregnancies \_\_\_\_\_

Number of preterm pregnancies \_\_\_\_\_

Number of pregnancies ending in abortion/miscarriage \_\_\_\_\_

Number of pregnancies resulting in live birth \_\_\_\_\_

Patient has biological children  Unknown  Yes  No

Number of biological children \_\_\_\_\_

### First Pregnancy

---

Patient's age at time of first pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before first pregnancy (in months)

- Unknown  Unplanned  1  2  3  4  5  
 6  7  8  9  10  11  12  
 13-24  25-36  >36

History of infertility for first pregnancy

- Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for first pregnancy

- Unknown  Male factor  Female factor  
 Combined male-female factor

Cause(s) of male infertility for first pregnancy

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

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

Cause(s) of female infertility for first pregnancy

- Unknown  Age  
 Alcohol use  Cancer/cancer treatment related  
 Early menopause  Fallopian tube damage/blockage

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

- Hyperprolactinemia
- Polycystic ovary syndrome (PCOS)
- Over-exercise
- Thyroid problems
- Uterine fibroid
- Other
- Pelvic adhesions
- Premature ovarian insufficiency
- Ovulation disorder
- Tobacco use
- Weight related

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

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

\_\_\_\_\_

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

- Unknown
- None
- Assisted reproductive technology
- Fertility drugs
- Surgery

Type of assisted reproductive technologies used during first pregnancy

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

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

\_\_\_\_\_

Partner tested for patient's disorder during first pregnancy

- Unknown
- Yes-biochemical
- Yes-molecular
- No

Partner also affected by disorder during first pregnancy    Unknown    Yes    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

- Unknown
- Amniocentesis
- Chorionic villus sampling

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

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

Additional prenatal testing performed on fetus as a result of parent's disorder for first pregnancy-other, specify \_\_\_\_\_

First pregnancy terminated    Unknown    Yes    No

Reason first pregnancy terminated

- Elective due to fetus affected with disorder

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

Date      

- Elective due to other fetal well-being unrelated to disorder
- Elective for other reason
- Elective due to maternal well-being
- Spontaneous

Gestational age (in weeks) at time of first pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for first pregnancy \_\_\_\_\_

Prenatal care received during first pregnancy  Unknown  Yes  No

Weeks gestation prenatal care started during first pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during first pregnancy

- Unknown  Metabolic diet  Medications
- Biochemical lab monitoring  Avoidance of fasting  Other

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

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

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

Patient in good metabolic condition during second trimester of first pregnancy

- Unknown  Pregnancy not sustained to second trimester
- Yes  No

Patient in good metabolic condition during third trimester of first pregnancy

- Unknown  Pregnancy not sustained to third trimester
- Yes  No

Number of outpatient metabolic visits for patient during first pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during first pregnancy

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

Number of hospitalizations for management of disorder during first pregnancy

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

Complications during first pregnancy

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

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

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

Complications related to disorder during first pregnancy- specify

- Unknown
- Acute fatty liver of pregnancy (AFLP)
- Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)
- Intrauterine growth restriction (AUGR)
- Mother affected with this condition
- Other

Complications not known to be related to disorder during first pregnancy-specify

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

Complications during first pregnancy-other, specify\_\_\_\_\_

Number of ED visits for complications during first pregnancy, unrelated to management of disorder

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

Number of hospitalizations for complications during first pregnancy, unrelated to management of disorder

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

Duration of longest inpatient hospitalization (in days) for any reason during first pregnancy\_\_\_\_\_

Highest value of primary metabolite of concern during first pregnancy (specify metabolite, value, and units of measure)

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

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

Lowest value of primary metabolite of concern during first pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during first pregnancy

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> Ammonia - Abn high                  |
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> C02 - Abn low                       | <input type="checkbox"/> C02 - WNL                           |
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

Other laboratory studies done on patient during first pregnancy: describe test(s) and result(s)

Additional interventions required during first pregnancy due to this metabolic condition

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                                    | <input type="checkbox"/> ED visits for hyperemesis/IV fluids   |
| <input type="checkbox"/> Hospitalizations for hyperemesis/IV fluids | <input type="checkbox"/> TPN                                   |
| <input type="checkbox"/> Tube feedings                              | <input type="checkbox"/> Additional medications                |
| <input type="checkbox"/> Home lab monitoring                        | <input type="checkbox"/> Increased frequency of lab monitoring |

Patient Name _____ Date <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 \_\_\_\_\_

Additional interventions planned for labor/delivery related to patient's disorder for first pregnancy

- Unknown
- None
- Additional maternal lab monitoring
- Altered anesthesia plan
- Change in delivery site
- IV fluids
- Letter to OB/MFM specialist
- Planned C-section
- Referral for high risk OB management
- Other

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

Actual interventions for labor/delivery related to patient's disorder for first pregnancy

- Unknown
- No
- Letter to OB/MFM specialist
- IV fluids
- Planned C-section
- Additional maternal lab monitoring
- Change in delivery site
- Referral for high risk OB management
- Altered anesthesia plan
- Other

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

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

Abnormal patient lab results during first delivery

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

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

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

Abnormal patient lab results during first delivery-other, specify

[Empty text box for abnormal patient lab results]

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

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

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

[Empty text box for additional maternal interventions]

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)

[Empty text box for acute health concerns]

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

[Empty text box for additional lab tests]

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

[Empty text box for health concerns]

Additional interventions for first newborn during or shortly after delivery (other than labs)

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

Date

- Unknown
- Blood/blood product transfusion
- Glucose infusion
- Medications
- NICU/special care nursery monitoring
- Resuscitation
- TPN
- Other

Additional interventions for first newborn during or shortly after delivery-other, specify

Birth measurements for first newborn  Unknown  Head circumference  Length  Weight

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

Birth head circumference units  cm  in

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

Birth length units  cm  in

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

Birth weight units  lbs  kg  gm  oz

APGAR score at 1 minute for first newborn

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

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

APGAR score at 5 minutes for first newborn

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

Health concern(s) with APGAR score at five minutes < 8 for first newborn

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

- Unknown
- <24 hours
- 24-28 hours
- 3-5 days
- 6-14 days
- >14 days
- N/A - baby was not born in a hospital
- baby is still in the hospital

Routine newborn screening done for first newborn

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

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

Describe abnormal newborn screen result for first newborn

Reason routine newborn screening was not done for first newborn

- Unknown
- Died prior to collection of NBS
- Refused
- Transferred to another facility prior to NBS collection
- Transfused prior to collection of NBS
- Other

Reason routine newborn screening was not done for first newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the first baby after birth to rule out the mother's disorder

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

Additional studies and results (examples: brain MRI, echocardiogram) done on first newborn to assess for effects of maternal disorder

First newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

- Unknown
- Yes
- No

Abnormal newborn exam findings for first newborn

- Unknown
- None
- Congenital heart disease
- Dysmorphism
- Lethargy
- Microcephaly
- Other congenital anomalies
- Poor feeding
- Respiratory distress
- Seizure(s)
- Small for gestational age
- Other

Abnormal newborn exam findings for first newborn-other, specify \_\_\_\_\_

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

Date      Newborn death at or shortly after delivery for first newborn  Unknown  Yes  NoNewborn's death related to maternal disorder for first newborn  Unknown  Yes  NoFirst newborn currently alive  Unknown  Yes  No**Second Pregnancy**

---

Patient's age at time of second pregnancy (in years)\_\_\_\_\_

Length of time patient tried to become pregnant before second pregnancy (in months)

- Unknown  Unplanned  1  2  3  4  5  
 6  7  8  9  10  11  12  
 13-24  25-36  >36

History of infertility for second pregnancy

- Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for second pregnancy

- Unknown  Male factor  Female factor  
 Combined male-female factor

Cause(s) of male infertility for second pregnancy

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

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

Cause(s) of female infertility for second pregnancy

- Unknown  Age  
 Alcohol use  Cancer/cancer treatment related  
 Early menopause  Fallopian tube damage/blockage  
 Hyperprolactinemia  Pelvic adhesions  
 Polycystic ovary syndrome (PCOS)  Premature ovarian insufficiency  
 Over-exercise  Ovulation disorder  
 Thyroid problems  Tobacco use  
 Uterine fibroid  Weight related  
 Other

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

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

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

Date      

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

- Unknown  None  
 Assisted reproductive technology  Fertility drugs  
 Surgery

Type of assisted reproductive technologies used during second pregnancy

- Assisted hatching  ICSI  IVF  
 Surgical sperm aspiration  Other

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

Partner tested for patient's disorder during second pregnancy

- Unknown  Yes-biochemical  Yes-molecular  No

Partner also affected by disorder during second pregnancy  Unknown  Yes  No

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

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

Type of prenatal testing performed for second pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for second pregnancy

- Unknown  Amniocentesis  Chorionic villus sampling

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

- Unknown  None  
 Fetal echocardiogram  Increased frequency prenatal ultrasounds  
 Other

Additional prenatal testing performed on fetus as a result of parent's disorder for second pregnancy-other, specify \_\_\_\_\_

Second pregnancy terminated  Unknown  Yes  No

Reason second pregnancy terminated

- Elective due to fetus affected with disorder  
 Elective due to other fetal well-being unrelated to disorder  
 Elective for other reason  
 Elective due to maternal well-being  
 Spontaneous

Gestational age (in weeks) at time of second pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for second pregnancy \_\_\_\_\_

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

Date      Prenatal care received during second pregnancy  Unknown  Yes  No

Weeks gestation prenatal care started during second pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during second pregnancy

 Unknown  Metabolic diet  Medications Biochemical lab monitoring  Avoidance of fasting  Other

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

Patient in good metabolic condition prior to second pregnancy  Unknown  Yes  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

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

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

Complications during second pregnancy-other, specify \_\_\_\_\_

Number of ED visits for complications during second pregnancy, unrelated to management of disorder

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

Number of hospitalizations for complications during second pregnancy, unrelated to management of disorder

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

Duration of longest inpatient hospitalization (in days) for any reason during second pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during second pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during second pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during second pregnancy

- Unknown                                       Ammonia - Abn high

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

Date      

- |  |  |
|--|--|
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> 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                               |  |

Other laboratory studies done on patient during second pregnancy: describe test(s) and result(s)

Additional interventions required during second pregnancy due to this metabolic condition

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                                    | <input type="checkbox"/> ED visits for hyperemesis/IV fluids   |
| <input type="checkbox"/> Hospitalizations for hyperemesis/IV fluids | <input type="checkbox"/> TPN                                   |
| <input type="checkbox"/> Tube feedings                              | <input type="checkbox"/> Additional medications                |
| <input type="checkbox"/> Home lab monitoring                        | <input type="checkbox"/> Increased frequency of lab monitoring |
| <input type="checkbox"/> More intensive fetal monitoring            | <input type="checkbox"/> Bedrest                               |
| <input type="checkbox"/> Other                                      |  |

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

\_\_\_\_\_

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

Additional interventions planned for labor/delivery related to patient's disorder for second pregnancy

- |                                  |                               |
|----------------------------------|-------------------------------|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> None |
|----------------------------------|-------------------------------|

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

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

- 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

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

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

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

- |   |   |
|---|---|
| <input type="radio"/> Unknown                       | <input type="radio"/> Caesarean section (scheduled or non-urgent) |
| <input type="radio"/> Casesarean section (emergent) | <input type="radio"/> Vaginal                                     |

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

Live delivery of second newborn     Unknown     Yes     No

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

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

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

Additional interventions for second newborn during or shortly after delivery (other than labs)

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                              | <input type="checkbox"/> Blood/blood product transfusion |
| <input type="checkbox"/> Glucose infusion                     | <input type="checkbox"/> Medications                     |
| <input type="checkbox"/> NICU/special care nursery monitoring | <input type="checkbox"/> Resuscitation                   |
| <input type="checkbox"/> TPN                                  | <input type="checkbox"/> Other                           |

Additional interventions for second newborn during or shortly after delivery-other, specify

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

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

APGAR score at 5 minutes for second newborn

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

Health concern(s) with APGAR score at five minutes < 8 for second newborn

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

- Unknown  <24 hours
- 24-28 hours 3-5 days
- 6-14 days  >14 days
- N/A - baby was not born in a hospital baby is still in the hospital

Routine newborn screening done for second newborn

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

Describe abnormal newborn screen result for second newborn

Reason routine newborn screening was not done for second newborn

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

Date

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

Additional studies and results (examples: brain MRI, echocardiogram) done on second newborn to assess for effects of maternal disorder

Second newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

- Unknown  Yes  No

Abnormal newborn exam findings for second newborn

- Unknown  None  Congenital heart disease
- Dysmorphism  Lethargy  Microcephaly
- Other congenital anomalies  Poor feeding  Respiratory distress
- Seizure(s)  Small for gestational age  Other

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

Newborn death at or shortly after delivery for second newborn  Unknown  Yes  No

Newborn's death related to maternal disorder for second newborn  Unknown  Yes  No

Second newborn currently alive  Unknown  Yes  No

### Third Pregnancy

Patient's age at time of third pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before third pregnancy (in months)

- Unknown  Unplanned  1  2  3  4  5

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

- 6       7       8       9       10       11       12  
 13-24       25-36       >36

History of infertility for third pregnancy

- Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for third pregnancy

- Unknown                       Male factor                       Female factor  
 Combined male-female factor

Cause(s) of male infertility for third pregnancy

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

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

Cause(s) of female infertility for third pregnancy

- Unknown                       Age  
 Alcohol use                       Cancer/cancer treatment related  
 Early menopause               Fallopian tube damage/blockage  
 Hyperprolactinemia               Pelvic adhesions  
 Polycystic ovary syndrome (PCOS)       Premature ovarian insufficiency  
 Over-exercise                       Ovulation disorder  
 Thyroid problems               Tobacco use  
 Uterine fibroid                       Weight related  
 Other

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

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

\_\_\_\_\_

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

- Unknown                       None  
 Assisted reproductive technology       Fertility drugs  
 Surgery

Type of assisted reproductive technologies used during third pregnancy

- Assisted hatching               ICSI                               IVF

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

Date      Surgical sperm aspiration OtherType of assisted reproductive technologies used during third pregnancy-other, specify  
\_\_\_\_\_

Partner tested for patient's disorder during third pregnancy

Unknown Yes-biochemical Yes-molecular NoPartner also affected by disorder during third pregnancy Unknown Yes NoPreimplantation genetic diagnosis for disorder done for third pregnancy Unknown Yes NoPrenatal testing done for fetus for this disorder for third pregnancy Unknown Yes NoType of prenatal testing performed for third pregnancy Unknown Biochemical Molecular

Method of prenatal testing for third pregnancy

Unknown Amniocentesis Chorionic villus sampling

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

Unknown NoneFetal echocardiogram Increased frequency prenatal ultrasoundsOther

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
- Elective due to maternal well-being
- Spontaneous

Gestational age (in weeks) at time of third pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for third pregnancy \_\_\_\_\_

Prenatal care received during third pregnancy Unknown Yes No

Weeks gestation prenatal care started during third pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during third pregnancy

Unknown Metabolic diet Medications

Biochemical lab monitoring Avoidance of fasting Other

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

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

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

--

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

Patient in good metabolic condition during second trimester of third pregnancy

UnknownPregnancy not sustained to second trimesterYesNo

Patient in good metabolic condition during third trimester of third pregnancy

UnknownPregnancy not sustained to third trimesterYesNo

Number of outpatient metabolic visits for patient during third pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during third pregnancy

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

Number of hospitalizations for management of disorder during third pregnancy

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

Complications during third pregnancy

UnknownYes-related to disorderYes- not known to be related to disorder No

Complications related to disorder during third pregnancy- specify

UnknownAcute fatty liver of pregnancy (AFLP)Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)Intrauterine growth restriction (AUGR)Mother affected with this conditionOther

Complications not known to be related to disorder during third pregnancy-specify

UnknownAdvanced maternal age (35+ years of age)Ectopic pregnancyGestational diabetesGroup B strep

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

Date

- Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)
- Intrauterine growth restriction (AUGR)
- Inadequate prenatal care
- Maternal prenatal substance exposure
- Preeclampsia
- Rh isoimmunization
- Toxemia
- Young maternal age (15 years of age + under)
- Preterm labor
- Other

Complications during third pregnancy-other, specify \_\_\_\_\_

Number of ED visits for complications during third pregnancy, unrelated to management of disorder

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

Number of hospitalizations for complications during third pregnancy, unrelated to management of disorder

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

Duration of longest inpatient hospitalization (in days) for any reason during third pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during third pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during third pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during third pregnancy

- |  |   |
|--|---|
| <input type="checkbox"/> Unknown             | <input type="checkbox"/> Ammonia - Abn high       |
| <input type="checkbox"/> Ammonia - WNL       | <input type="checkbox"/> Blood glucose - Abn low  |
| <input type="checkbox"/> Blood glucose - WNL | <input type="checkbox"/> Blood Glucose - Abn high |
| <input type="checkbox"/> BNP - Abn high      | <input type="checkbox"/> BNP - WNL                |
| <input type="checkbox"/> CBC - Abn           | <input type="checkbox"/> CBC - WNL                |
| <input type="checkbox"/> C02 - Abn low       | <input type="checkbox"/> C02 - WNL                |

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

Date      

- |  |  |
|--|--|
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

Other laboratory studies done on patient during third pregnancy: describe test(s) and result(s)

Additional interventions required during third pregnancy due to this metabolic condition

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                                    | <input type="checkbox"/> ED visits for hyperemesis/IV fluids   |
| <input type="checkbox"/> Hospitalizations for hyperemesis/IV fluids | <input type="checkbox"/> TPN                                   |
| <input type="checkbox"/> Tube feedings                              | <input type="checkbox"/> Additional medications                |
| <input type="checkbox"/> Home lab monitoring                        | <input type="checkbox"/> Increased frequency of lab monitoring |
| <input type="checkbox"/> More intensive fetal monitoring            | <input type="checkbox"/> Bedrest                               |
| <input type="checkbox"/> Other                                      |  |

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

\_\_\_\_\_

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

Additional interventions planned for labor/delivery related to patient's disorder for third pregnancy

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                              | <input type="checkbox"/> None                    |
| <input type="checkbox"/> Additional maternal lab monitoring   | <input type="checkbox"/> Altered anesthesia plan |
| <input type="checkbox"/> Change in delivery site              | <input type="checkbox"/> IV fluids               |
| <input type="checkbox"/> Letter to OB/MFM specialist          | <input type="checkbox"/> Planned C-section       |
| <input type="checkbox"/> Referral for high risk OB management | <input type="checkbox"/> Other                   |

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

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      

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

## Complications during fourth pregnancy

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

## Complications related to disorder during fourth pregnancy- specify

- Unknown  
 Acute fatty liver of pregnancy (AFLP)  
 Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)  
 Intrauterine growth restriction (AUGR)  
 Mother affected with this condition  
 Other

## Complications not known to be related to disorder during fourth pregnancy-specify

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

Complications during fourth pregnancy-other, specify \_\_\_\_\_

## Number of ED visits for complications during fourth pregnancy, unrelated to management of disorder

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

## Number of hospitalizations for complications during fourth pregnancy, unrelated to management of disorder

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

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

Date    |    |   

Duration of longest inpatient hospitalization (in days) for any reason during fourth pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during fourth pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during fourth pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during fourth pregnancy

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> Ammonia - Abn high                  |
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> CO <sub>2</sub> - Abn low           | <input type="checkbox"/> CO <sub>2</sub> - WNL               |
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

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

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

Other laboratory studies done on patient during fourth pregnancy: describe test(s) and result(s)

[Empty text box for laboratory studies]

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

\_\_\_\_\_

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

Additional interventions planned for labor/delivery related to patient's disorder for fourth pregnancy

- Unknown
- Additional maternal lab monitoring
- Change in delivery site
- Letter to OB/MFM specialist
- Referral for high risk OB management
- None
- Altered anesthesia plan
- IV fluids
- Planned C-section
- Other

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

[Empty text box for additional interventions]

Actual interventions for labor/delivery related to patient's disorder for fourth pregnancy

- Unknown
- Letter to OB/MFM specialist
- Planned C-section
- Change in delivery site
- Altered anesthesia plan
- No
- IV fluids
- Additional maternal lab monitoring
- Referral for high risk OB management
- Other

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

[Empty text box for actual interventions]

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

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

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

[Empty text box for acute health concerns]

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

[Empty text box for abnormal lab results other]

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

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

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

[Empty text box for additional maternal interventions other]

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)

[Empty text box for acute health concerns for newborn]

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

Date

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

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

Additional interventions for fourth newborn during or shortly after delivery (other than labs)

- Unknown  Blood/blood product transfusion
- Glucose infusion  Medications
- NICU/special care nursery monitoring  Resuscitation
- TPN  Other

Additional interventions for fourth newborn during or shortly after delivery-other, specify

Birth measurements for fourth newborn  Unknown  Head circumference  Length  Weight

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

Birth head circumference units  cm  in

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

Birth length units  cm  in

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

Birth weight units  lbs  kg  gm  oz

APGAR score at 1 minute for fourth newborn

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

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

APGAR score at 5 minutes for fourth newborn

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

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

Date      

Health concern(s) with APGAR score at five minutes &lt; 8 for fourth newborn

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

- Unknown  <24 hours  
 24-28 hours  3-5 days  
 6-14 days  >14 days  
 N/A - baby was not born in a hospital  baby is still in the hospital

Routine newborn screening done for fourth newborn

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

Describe abnormal newborn screen result for fourth newborn

Reason routine newborn screening was not done for fourth newborn

- Unknown  
 Died prior to collection of NBS  
 Refused  
 Transferred to another facility prior to NBS collection  
 Transfused prior to collection of NBS  
 Other

Reason routine newborn screening was not done for fourth newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the fourth baby after birth to rule out the mother's disorder

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

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

Date      

Additional studies and results (examples: brain MRI, echocardiogram) done on fourth newborn to assess for effects of maternal disorder

Fourth newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

Unknown                       Yes                       No

Abnormal newborn exam findings for fourth newborn

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 fourth newborn-other, specify \_\_\_\_\_

Newborn death at or shortly after delivery for fourth newborn    Unknown    Yes    No

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

Fourth newborn currently alive    Unknown    Yes    No

## **Fifth Pregnancy**

---

Patient's age at time of fifth pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before fifth pregnancy (in months)

Unknown    Unplanned    1                       2                       3                       4                       5  
 6                       7                       8                       9                       10                       11                       12  
 13-24                       25-36                       >36

History of infertility for fifth pregnancy

Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for fifth pregnancy

Unknown                       Male factor                       Female factor  
 Combined male-female factor

Cause(s) of male infertility for fifth pregnancy

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

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

- Tobacco use  Other

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

Cause(s) of female infertility for fifth pregnancy

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

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

\_\_\_\_\_

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

- Unknown  None  
 Assisted reproductive technology  Fertility drugs  
 Surgery

Type of assisted reproductive technologies used during fifth pregnancy

- Assisted hatching  ICSI  IVF  
 Surgical sperm aspiration  Other

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

\_\_\_\_\_

Partner tested for patient's disorder during fifth pregnancy

- Unknown  Yes-biochemical  Yes-molecular  No

Partner also affected by disorder during fifth pregnancy  Unknown  Yes  No

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

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

Type of prenatal testing performed for fifth pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for fifth pregnancy

- Unknown  Amniocentesis  Chorionic villus sampling

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

- Unknown  None  
 Fetal echocardiogram  Increased frequency prenatal ultrasounds

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

Date

Other

Additional prenatal testing performed on fetus as a result of parent's disorder for fifth pregnancy-other, specify \_\_\_\_\_

Fifth pregnancy terminated  Unknown  Yes  No

Reason fifth pregnancy terminated

- Elective due to fetus affected with disorder
- Elective due to other fetal well-being unrelated to disorder
- Elective for other reason
- Elective due to maternal well-being
- Spontaneous

Gestational age (in weeks) at time of fifth pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for fifth pregnancy \_\_\_\_\_

Prenatal care received during fifth pregnancy  Unknown  Yes  No

Weeks gestation prenatal care started during fifth pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during fifth pregnancy

- Unknown  Metabolic diet  Medications
- Biochemical lab monitoring  Avoidance of fasting  Other

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

Patient in good metabolic condition prior to fifth pregnancy  Unknown  Yes  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

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

Date      

Number of hospitalizations for management of disorder during fifth pregnancy

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

Complications during fifth pregnancy

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

Complications related to disorder during fifth pregnancy- specify

- Unknown  
 Acute fatty liver of pregnancy (AFLP)  
 Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)  
 Intrauterine growth restriction (AUGR)  
 Mother affected with this condition  
 Other

Complications not known to be related to disorder during fifth pregnancy-specify

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

Complications during fifth pregnancy-other, specify \_\_\_\_\_

Number of ED visits for complications during fifth pregnancy, unrelated to management of disorder

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

Number of hospitalizations for complications during fifth pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5

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

Date      

6     
  7     
  8     
  9     
  10     
  >10

Duration of longest inpatient hospitalization (in days) for any reason during fifth pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during fifth pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during fifth pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during fifth pregnancy

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> Ammonia - Abn high                  |
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> CO <sub>2</sub> - Abn low           | <input type="checkbox"/> CO <sub>2</sub> - WNL               |
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

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

Date |||||

Other laboratory studies done on patient during fifth pregnancy: describe test(s) and result(s)

Additional interventions required during fifth pregnancy due to this metabolic condition

- 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 fifth pregnancy due to this metabolic condition-other, specify

\_\_\_\_\_

Total maternal weight gain (in kg) 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

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

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

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

Abnormal patient lab results during fifth delivery

- |                                       |  |  |
|---------------------------------------|--|--|
| <input type="checkbox"/> Unknown      | <input type="checkbox"/> None                          | <input type="checkbox"/> Normal labs during delivery |
| <input type="checkbox"/> Elevated CK  | <input type="checkbox"/> Elevated liver function tests | <input type="checkbox"/> Hyperammonemia              |
| <input type="checkbox"/> Hypoglycemia | <input type="checkbox"/> Metabolic decompensation      | <input type="checkbox"/> Other                       |

Abnormal patient lab results during fifth delivery-other, specify

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

- |                                      |   |  |
|--------------------------------------|---|--|
| <input type="checkbox"/> Unknown     | <input type="checkbox"/> Ammonul        | <input type="checkbox"/> Blood/blood product transfusion |
| <input type="checkbox"/> Dialysis    | <input type="checkbox"/> ICU monitoring | <input type="checkbox"/> Infusions                       |
| <input type="checkbox"/> Medications | <input type="checkbox"/> Resuscitation  | <input type="checkbox"/> TPN                             |
| <input type="checkbox"/> Other       |   |  |

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

Patient death during or shortly after fifth delivery    Unknown    Yes    No

Disorder contributed to death of mother during fifth delivery    Unknown    Yes    No

Weeks gestation at time of fifth delivery - round to nearest week\_\_\_\_\_

Method of fifth delivery

- |   |   |
|---|---|
| <input type="radio"/> Unknown                       | <input type="radio"/> Caesarean section (scheduled or non-urgent) |
| <input type="radio"/> Casesarean section (emergent) | <input type="radio"/> Vaginal                                     |

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

Live delivery of fifth newborn    Unknown    Yes    No

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

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

Date

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

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

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

Date      

Health concern(s) with APGAR score at five minutes &lt; 8 for fifth newborn

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

- Unknown  <24 hours  
 24-28 hours  3-5 days  
 6-14 days  >14 days  
 N/A - baby was not born in a hospital  baby is still in the hospital

Routine newborn screening done for fifth newborn

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

Describe abnormal newborn screen result for fifth newborn

Reason routine newborn screening was not done for fifth newborn

- Unknown  
 Died prior to collection of NBS  
 Refused  
 Transferred to another facility prior to NBS collection  
 Transfused prior to collection of NBS  
 Other

Reason routine newborn screening was not done for fifth newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the fifth baby after birth to rule out the mother's disorder

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

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

Date      

Additional studies and results (examples: brain MRI, echocardiogram) done on fifth newborn to assess for effects of maternal disorder

Fifth newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

Unknown  Yes  No

Abnormal newborn exam findings for fifth newborn

Unknown  None  Congenital heart disease  
 Dysmorphism  Lethargy  Microcephaly  
 Other congenital anomalies  Poor feeding  Respiratory distress  
 Seizure(s)  Small for gestational age  Other

Abnormal newborn exam findings for fifth newborn-other, specify \_\_\_\_\_

Newborn death at or shortly after delivery for fifth newborn  Unknown  Yes  No

Newborn's death related to maternal disorder for fifth newborn  Unknown  Yes  No

Fifth newborn currently alive  Unknown  Yes  No

## Sixth Pregnancy

Patient's age at time of sixth pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before sixth pregnancy (in months)

Unknown  Unplanned  1  2  3  4  5  
 6  7  8  9  10  11  12  
 13-24  25-36  >36

History of infertility for sixth pregnancy

Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for sixth pregnancy

Unknown  Male factor  Female factor  
 Combined male-female factor

Cause(s) of male infertility for sixth pregnancy

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

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

Date      

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

Cause(s) of female infertility for sixth pregnancy

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                          | <input type="checkbox"/> Age                             |
| <input type="checkbox"/> Alcohol use                      | <input type="checkbox"/> Cancer/cancer treatment related |
| <input type="checkbox"/> Early menopause                  | <input type="checkbox"/> Fallopian tube damage/blockage  |
| <input type="checkbox"/> Hyperprolactinemia               | <input type="checkbox"/> Pelvic adhesions                |
| <input type="checkbox"/> Polycystic ovary syndrome (PCOS) | <input type="checkbox"/> Premature ovarian insufficiency |
| <input type="checkbox"/> Over-exercise                    | <input type="checkbox"/> Ovulation disorder              |
| <input type="checkbox"/> Thyroid problems                 | <input type="checkbox"/> Tobacco use                     |
| <input type="checkbox"/> Uterine fibroid                  | <input type="checkbox"/> Weight related                  |
| <input type="checkbox"/> Other                            |  |

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

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

\_\_\_\_\_

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

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                          | <input type="checkbox"/> None            |
| <input type="checkbox"/> Assisted reproductive technology | <input type="checkbox"/> Fertility drugs |
| <input type="checkbox"/> Surgery                          |  |

Type of assisted reproductive technologies used during sixth pregnancy

- |  |                                |                              |
|--|--------------------------------|------------------------------|
| <input type="checkbox"/> Assisted hatching         | <input type="checkbox"/> ICSI  | <input type="checkbox"/> IVF |
| <input type="checkbox"/> Surgical sperm aspiration | <input type="checkbox"/> Other |                              |

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

\_\_\_\_\_

Partner tested for patient's disorder during sixth pregnancy

- |                                  |  |  |                             |
|----------------------------------|--|--|-----------------------------|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Yes-biochemical | <input type="checkbox"/> Yes-molecular | <input type="checkbox"/> No |
|----------------------------------|--|--|-----------------------------|

Partner also affected by disorder during sixth pregnancy  Unknown  Yes  NoPreimplantation genetic diagnosis for disorder done for sixth pregnancy  Unknown  Yes  NoPrenatal testing done for fetus for this disorder for sixth pregnancy  Unknown  Yes  NoType of prenatal testing performed for sixth pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for sixth pregnancy

- |                                  |  |  |
|----------------------------------|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Amniocentesis | <input type="checkbox"/> Chorionic villus sampling |
|----------------------------------|--|--|

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

- |   |   |
|---|---|
| <input type="checkbox"/> Unknown              | <input type="checkbox"/> None                                     |
| <input type="checkbox"/> Fetal echocardiogram | <input type="checkbox"/> Increased frequency prenatal ultrasounds |
| <input type="checkbox"/> Other                |   |

Patient Name _____
Date <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 sixth pregnancy-other, specify \_\_\_\_\_

Sixth pregnancy terminated Unknown Yes No

Reason sixth pregnancy terminated

- Elective due to fetus affected with disorder
- Elective due to other fetal well-being unrelated to disorder
- Elective for other reason
- Elective due to maternal well-being
- Spontaneous

Gestational age (in weeks) at time of sixth pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for sixth pregnancy \_\_\_\_\_

Prenatal care received during sixth pregnancy Unknown Yes No

Weeks gestation prenatal care started during sixth pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during sixth pregnancy

- Unknown  Metabolic diet  Medications
- Biochemical lab monitoring  Avoidance of fasting  Other

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

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

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

Patient in good metabolic condition during second trimester of sixth pregnancy

- Unknown  Pregnancy not sustained to second trimester
- Yes  No

Patient in good metabolic condition during third trimester of sixth pregnancy

- Unknown  Pregnancy not sustained to third trimester
- Yes  No

Number of outpatient metabolic visits for patient during sixth pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during sixth pregnancy

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

Number of hospitalizations for management of disorder during sixth pregnancy

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

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

Complications during sixth pregnancy

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

Complications related to disorder during sixth pregnancy- specify

- Unknown
- Acute fatty liver of pregnancy (AFLP)
- Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)
- Intrauterine growth restriction (AUGR)
- Mother affected with this condition
- Other

Complications not known to be related to disorder during sixth pregnancy-specify

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

Complications during sixth pregnancy-other, specify\_\_\_\_\_

Number of ED visits for complications during sixth pregnancy, unrelated to management of disorder

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

Number of hospitalizations for complications during sixth pregnancy, unrelated to management of disorder

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

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

Date |||||

Duration of longest inpatient hospitalization (in days) for any reason during sixth pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during sixth pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during sixth pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during sixth pregnancy

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> Ammonia - Abn high                  |
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> CO2 - Abn low                       | <input type="checkbox"/> CO2 - WNL                           |
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

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

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

Other laboratory studies done on patient during sixth pregnancy: describe test(s) and result(s)

[Empty text box for laboratory studies]

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

\_\_\_\_\_

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

Additional interventions planned for labor/delivery related to patient's disorder for sixth pregnancy

- Unknown
- Additional maternal lab monitoring
- Change in delivery site
- Letter to OB/MFM specialist
- Referral for high risk OB management
- None
- Altered anesthesia plan
- IV fluids
- Planned C-section
- Other

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

[Empty text box for additional interventions]

Actual interventions for labor/delivery related to patient's disorder for sixth pregnancy

- Unknown
- Letter to OB/MFM specialist
- Planned C-section
- Change in delivery site
- Altered anesthesia plan
- No
- IV fluids
- Additional maternal lab monitoring
- Referral for high risk OB management
- Other

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

[Empty text box for actual interventions]

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

Date

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

Abnormal patient lab results during sixth delivery

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

Abnormal patient lab results during sixth delivery-other, specify

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

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

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

Patient death during or shortly after sixth delivery    Unknown    Yes    No

Disorder contributed to death of mother during sixth delivery    Unknown    Yes    No

Weeks gestation at time of sixth delivery - round to nearest week\_\_\_\_\_

Method of sixth delivery

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

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

Live delivery of sixth newborn    Unknown    Yes    No

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

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

Date

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

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

Additional interventions for sixth newborn during or shortly after delivery (other than labs)

- Unknown  Blood/blood product transfusion
- Glucose infusion  Medications
- NICU/special care nursery monitoring  Resuscitation
- TPN  Other

Additional interventions for sixth newborn during or shortly after delivery-other, specify

Birth measurements for sixth newborn  Unknown  Head circumference  Length  Weight

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

Birth head circumference units  cm  in

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

Birth length units  cm  in

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

Birth weight units  lbs  kg  gm  oz

APGAR score at 1 minute for sixth newborn

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

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

APGAR score at 5 minutes for sixth newborn

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

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

Date |||||

Health concern(s) with APGAR score at five minutes &lt; 8 for sixth newborn

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

- Unknown  <24 hours  
 24-28 hours  3-5 days  
 6-14 days  >14 days  
 N/A - baby was not born in a hospital  baby is still in the hospital

Routine newborn screening done for sixth newborn

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

Describe abnormal newborn screen result for sixth newborn

Reason routine newborn screening was not done for sixth newborn

- Unknown  
 Died prior to collection of NBS  
 Refused  
 Transferred to another facility prior to NBS collection  
 Transfused prior to collection of NBS  
 Other

Reason routine newborn screening was not done for sixth newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the sixth baby after birth to rule out the mother's disorder

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

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

Date      

Additional studies and results (examples: brain MRI, echocardiogram) done on sixth newborn to assess for effects of maternal disorder

Sixth newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

Unknown  Yes  No

Abnormal newborn exam findings for sixth newborn

Unknown  None  Congenital heart disease  
 Dysmorphism  Lethargy  Microcephaly  
 Other congenital anomalies  Poor feeding  Respiratory distress  
 Seizure(s)  Small for gestational age  Other

Abnormal newborn exam findings for sixth newborn-other, specify \_\_\_\_\_

Newborn death at or shortly after delivery for sixth newborn  Unknown  Yes  No

Newborn's death related to maternal disorder for sixth newborn  Unknown  Yes  No

Sixth newborn currently alive  Unknown  Yes  No

## Seventh Pregnancy

Patient's age at time of seventh pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before seventh pregnancy (in months)

Unknown  Unplanned  1  2  3  4  5  
 6  7  8  9  10  11  12  
 13-24  25-36  >36

History of infertility for seventh pregnancy

Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for seventh pregnancy

Unknown  Male factor  Female factor  
 Combined male-female factor

Cause(s) of male infertility for seventh pregnancy

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

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

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

\_\_\_\_\_

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

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

Date      

Additional prenatal testing performed on fetus as a result of parent's disorder for seventh pregnancy-other, specify \_\_\_\_\_

Seventh pregnancy terminated  Unknown  Yes  No

Reason seventh pregnancy terminated

- Elective due to fetus affected with disorder  
 Elective due to other fetal well-being unrelated to disorder  
 Elective for other reason  
 Elective due to maternal well-being  
 Spontaneous

Gestational age (in weeks) at time of seventh pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for seventh pregnancy \_\_\_\_\_

Prenatal care received during seventh pregnancy  Unknown  Yes  No

Weeks gestation prenatal care started during seventh pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during seventh pregnancy

- Unknown  Metabolic diet  Medications  
 Biochemical lab monitoring  Avoidance of fasting  Other

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

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

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

Patient in good metabolic condition during second trimester of seventh pregnancy

- Unknown  Pregnancy not sustained to second trimester  
 Yes  No

Patient in good metabolic condition during third trimester of seventh pregnancy

- Unknown  Pregnancy not sustained to third trimester  
 Yes  No

Number of outpatient metabolic visits for patient during seventh pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during seventh pregnancy

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

Number of hospitalizations for management of disorder during seventh pregnancy

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

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

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

Complications during seventh pregnancy

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

Complications related to disorder during seventh pregnancy- specify

- Unknown
- Acute fatty liver of pregnancy (AFLP)
- Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)
- Intrauterine growth restriction (AUGR)
- Mother affected with this condition
- Other

Complications not known to be related to disorder during seventh pregnancy-specify

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

Complications during seventh pregnancy-other, specify \_\_\_\_\_

Number of ED visits for complications during seventh pregnancy, unrelated to management of disorder

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

Number of hospitalizations for complications during seventh pregnancy, unrelated to management of disorder

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

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

Date |||||

Duration of longest inpatient hospitalization (in days) for any reason during seventh pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during seventh pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during seventh pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during seventh pregnancy

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> Ammonia - Abn high                  |
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> CO <sub>2</sub> - Abn low           | <input type="checkbox"/> CO <sub>2</sub> - WNL               |
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

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

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

Other laboratory studies done on patient during seventh pregnancy: describe test(s) and result(s)

[Empty text box for laboratory studies]

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\_\_\_\_\_

Additional interventions planned for labor/delivery related to patient's disorder for seventh pregnancy

- Unknown
- Additional maternal lab monitoring
- Change in delivery site
- Letter to OB/MFM specialist
- Referral for high risk OB management
- None
- Altered anesthesia plan
- IV fluids
- Planned C-section
- Other

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

[Empty text box for additional interventions]

Actual interventions for labor/delivery related to patient's disorder for seventh pregnancy

- Unknown
- Letter to OB/MFM specialist
- Planned C-section
- Change in delivery site
- Altered anesthesia plan
- No
- IV fluids
- Additional maternal lab monitoring
- Referral for high risk OB management
- Other

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

[Empty text box for actual interventions]

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

Date

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

Abnormal patient lab results during seventh delivery

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

Abnormal patient lab results during seventh delivery-other, specify

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

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

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

Patient death during or shortly after seventh delivery     Unknown     Yes     No

Disorder contributed to death of mother during seventh delivery     Unknown     Yes     No

Weeks gestation at time of seventh delivery - round to nearest week\_\_\_\_\_

Method of seventh delivery

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

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

Live delivery of seventh newborn     Unknown     Yes     No

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

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

Date

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

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

Additional interventions for seventh newborn during or shortly after delivery (other than labs)

- Unknown  Blood/blood product transfusion
- Glucose infusion  Medications
- NICU/special care nursery monitoring  Resuscitation
- TPN  Other

Additional interventions for seventh newborn during or shortly after delivery-other, specify

Birth measurements for seventh newborn  Unknown  Head circumference  Length  Weight

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

Birth head circumference units  cm  in

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

Birth length units  cm  in

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

Birth weight units  lbs  kg  gm  oz

APGAR score at 1 minute for seventh newborn

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

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

APGAR score at 5 minutes for seventh newborn

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

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

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

Health concern(s) with APGAR score at five minutes &lt; 8 for seventh newborn

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

- Unknown                                       <24 hours  
 24-28 hours                                       3-5 days  
 6-14 days     >14 days  
 N/A - baby was not born in a hospital     baby is still in the hospital

Routine newborn screening done for seventh newborn

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

Describe abnormal newborn screen result for seventh newborn

Reason routine newborn screening was not done for seventh newborn

- Unknown  
 Died prior to collection of NBS  
 Refused  
 Transferred to another facility prior to NBS collection  
 Transfused prior to collection of NBS  
 Other

Reason routine newborn screening was not done for seventh newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the seventh baby after birth to rule out the mother's disorder

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

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

Date      

Additional studies and results (examples: brain MRI, echocardiogram) done on seventh newborn to assess for effects of maternal disorder

Seventh newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

Unknown                       Yes                       No

Abnormal newborn exam findings for seventh newborn

Unknown                       None                       Congenital heart disease  
 Dysmorphism                       Lethargy                       Microcephaly  
 Other congenital anomalies    Poor feeding                       Respiratory distress  
 Seizure(s)                       Small for gestational age    Other

Abnormal newborn exam findings for seventh newborn-other, specify \_\_\_\_\_

Newborn death at or shortly after delivery for seventh newborn    Unknown    Yes    No

Newborn's death related to maternal disorder for seventh newborn    Unknown    Yes    No

Seventh newborn currently alive    Unknown    Yes    No

## **Eighth Pregnancy**

---

Patient's age at time of eighth pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before eighth pregnancy (in months)

Unknown    Unplanned    1                       2                       3                       4                       5  
 6                       7                       8                       9                       10                       11                       12  
 13-24                       25-36                       >36

History of infertility for eighth pregnancy

Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for eighth pregnancy

Unknown                       Male factor                       Female factor  
 Combined male-female factor

Cause(s) of male infertility for eighth pregnancy

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

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

- Tobacco use  Other

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

Cause(s) of female infertility for eighth pregnancy

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

Cause(s) of infertility for eighth pregnancy, combined male-female factor- specify \_\_\_\_\_

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

- Unknown  None  
 Assisted reproductive technology  Fertility drugs  
 Surgery

Type of assisted reproductive technologies used during eighth pregnancy

- Assisted hatching  ICSI  IVF  
 Surgical sperm aspiration  Other

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

Partner tested for patient's disorder during eighth pregnancy

- Unknown  Yes-biochemical  Yes-molecular  No

Partner also affected by disorder during eighth pregnancy  Unknown  Yes  No

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

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

Type of prenatal testing performed for eighth pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for eighth pregnancy

- Unknown  Amniocentesis  Chorionic villus sampling

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

- Unknown  None  
 Fetal echocardiogram  Increased frequency prenatal ultrasounds

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

Date

Other

Additional prenatal testing performed on fetus as a result of parent's disorder for eighth pregnancy-other, specify \_\_\_\_\_

Eighth pregnancy terminated  Unknown  Yes  No

Reason eighth pregnancy terminated

- Elective due to fetus affected with disorder
- Elective due to other fetal well-being unrelated to disorder
- Elective for other reason
- Elective due to maternal well-being
- Spontaneous

Gestational age (in weeks) at time of eighth pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for eighth pregnancy \_\_\_\_\_

Prenatal care received during eighth pregnancy  Unknown  Yes  No

Weeks gestation prenatal care started during eighth pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during eighth pregnancy

- Unknown  Metabolic diet  Medications
- Biochemical lab monitoring  Avoidance of fasting  Other

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

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

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

Patient in good metabolic condition during second trimester of eighth pregnancy

- Unknown  Pregnancy not sustained to second trimester
- Yes  No

Patient in good metabolic condition during third trimester of eighth pregnancy

- Unknown  Pregnancy not sustained to third trimester
- Yes  No

Number of outpatient metabolic visits for patient during eighth pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during eighth pregnancy

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

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

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

Number of hospitalizations for management of disorder during eighth pregnancy

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

Complications during eighth pregnancy

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

Complications related to disorder during eighth pregnancy- specify

- Unknown  
 Acute fatty liver of pregnancy (AFLP)  
 Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)  
 Intrauterine growth restriction (AUGR)  
 Mother affected with this condition  
 Other

Complications not known to be related to disorder during eighth pregnancy-specify

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

Complications during eighth pregnancy-other, specify \_\_\_\_\_

Number of ED visits for complications during eighth pregnancy, unrelated to management of disorder

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

Number of hospitalizations for complications during eighth pregnancy, unrelated to management of disorder

- Unknown  0       1       2       3       4       5

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

Date      

6     
  7     
  8     
  9     
  10     
  >10

Duration of longest inpatient hospitalization (in days) for any reason during eighth pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during eighth pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during eighth pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during eighth pregnancy

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> Ammonia - Abn high                  |
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> CO <sub>2</sub> - Abn low           | <input type="checkbox"/> CO <sub>2</sub> - WNL               |
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

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

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

Other laboratory studies done on patient during eighth pregnancy: describe test(s) and result(s)

[Empty text box for laboratory studies]

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

\_\_\_\_\_

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

Actual interventions for labor/delivery related to patient's disorder for eighth pregnancy

- Unknown
- Letter to OB/MFM specialist
- Planned C-section
- Change in delivery site
- Altered anesthesia plan
- No
- IV fluids
- Additional maternal lab monitoring
- Referral for high risk OB management
- Other

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

[Empty text box for additional interventions]

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

[Empty text box for acute health concerns]

Abnormal patient lab results during eighth delivery

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

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

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

Abnormal patient lab results during eighth delivery-other, specify

[Empty text box for abnormal patient lab results]

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

[Empty text box for additional maternal interventions]

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)

[Empty text box for acute health concerns]

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

[Empty text box for additional lab tests]

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

[Empty text box for health concerns]

Additional interventions for eighth 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 eighth newborn during or shortly after delivery-other, specify

Birth measurements for eighth newborn  Unknown  Head circumference  Length  Weight

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

Birth head circumference units  cm  in

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

Birth length units  cm  in

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

Birth weight units  lbs  kg  gm  oz

APGAR score at 1 minute for eighth newborn

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

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

APGAR score at 5 minutes for eighth newborn

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

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

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

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

Describe abnormal newborn screen result for eighth newborn

Reason routine newborn screening was not done for eighth newborn

- Unknown
- Died prior to collection of NBS
- Refused
- Transferred to another facility prior to NBS collection
- Transfused prior to collection of NBS
- Other

Reason routine newborn screening was not done for eighth newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the eighth baby after birth to rule out the mother's disorder

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

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

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

Date      

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  
 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

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

Date      

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

- Unknown  Yes-biochemical  Yes-molecular  No

Partner also affected by disorder during current pregnancy  Unknown  Yes  NoPreimplantation genetic diagnosis for disorder done for current pregnancy  Unknown  Yes  NoPrenatal testing done for fetus for this disorder for current pregnancy  Unknown  Yes  NoType 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) \_\_\_\_\_

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

Date      

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for current pregnancy \_\_\_\_\_

Prenatal care received during current pregnancy  Unknown  Yes  No

Weeks gestation prenatal care started during current pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during current pregnancy

- Unknown  Metabolic diet  Medications  
 Biochemical lab monitoring  Avoidance of fasting  Other

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

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

Patient in good metabolic condition during first trimester of current pregnancy

- Unknown  Yes  No

Patient in good metabolic condition during second trimester of current pregnancy

- Unknown  Pregnancy not sustained to second trimester  
 Yes  No

Patient in good metabolic condition during third trimester of current pregnancy

- Unknown  Pregnancy not sustained to third trimester  
 Yes  No

Number of outpatient metabolic visits for patient during current pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during current pregnancy

- 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)

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

Date

Mother affected with this condition

Other

Complications not known to be related to disorder during current pregnancy-specify

Unknown

Advanced maternal age (35+ years of age)

Ectopic pregnancy

Gestational diabetes

Group B strep

Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)

Intrauterine growth restriction (AUGR)

Inadequate prenatal care

Maternal prenatal substance exposure

Preeclampsia

Rh isoimmunization

Toxemia

Young maternal age (15 years of age + under)

Preterm labor

Other

Complications during current pregnancy-other, specify \_\_\_\_\_

Number of ED visits for complications during current pregnancy, unrelated to management of disorder

Unknown  0  1  2  3  4  5

6  7  8  9  10  >10

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)

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

Date      

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"/> 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 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 |

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"/>

- More intensive fetal monitoring
- Bedrest
- 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 \_\_\_\_\_

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      **PKU - Dialysis**

Number of different episodes during which dialysis (any type) was used

 1     2     3     4     5     6     7     8     9     10     >10
**First Dialysis Treatment**

Type(s) of dialysis received during first episode

- Unknown             APD/CCPD             ECMO             CAPD  
 CVVH             CVVD             CVVHDF             Hemodialysis  
 Peritoneal dialysis

Reason for first episode of dialysis

- Unknown             Hyperammonemia     MSUD             Organ failure             Sepsis  
 Other

Reason for first episode of dialysis-other, specify

Start date of first episode of dialysis treatment      

Duration (in days) of first episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during first episode of dialysis

- Unknown             None             Ammonia             Blood urea nitrogen  
 Plasma alloisoleucine     Plasma isoleucine     Plasma leucine             Plasma valine  
 Serum creatinine             Other

Metabolite(s) of concern during first episode of dialysis-other,specify \_\_\_\_\_

Peak value of ammonia during first episode of dialysis \_\_\_\_\_

Peak value of ammonia during first episode of dialysis units

 umol/L             ug/dL             ug/L             ug/mL             g/dL

Peak value of blood urea nitrogen during first episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during first episode of dialysis units     mg/dL     mmol/L

Peak value of plasma alloisoleucine during first episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during first episode of dialysis units

 umol/dL             umol/L             mg/dL

Peak value of plasma isoleucine during first episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during first episode of dialysis units     umol/dL     umol/L     mg/dL

Peak value of plasma leucine during first episode of dialysis \_\_\_\_\_

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

Peak value of plasma leucine during first episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of plasma valine during first episode of dialysis \_\_\_\_\_

Peak value of plasma valine during first episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of serum creatinine during first episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during first episode of dialysis units  umol/L  mg/dL

Peak value of other metabolite during first episode of dialysis \_\_\_\_\_

Peak value of other metabolite during first episode of dialysis units \_\_\_\_\_

Reason first episode of dialysis treatment was stopped

- Unknown  Acute episode resolved  Treatment withdrawn  Death
- Other

Reason first episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

### Second Dialysis Treatment

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Type(s) of dialysis received during second episode

- Unknown  APD/CCPD  ECMO  CAPD
- CVVH  CVVD  CVVHDF  Hemodialysis
- Peritoneal dialysis

Reason for second episode of dialysis

- Unknown  Hyperammonemia  MSUD  Organ failure  Sepsis
- Other

Reason for second episode of dialysis-other, specify

Start date of second episode of dialysis treatment    |    |

Duration (in days) of second episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during second episode of dialysis

- Unknown  None  Ammonia  Blood urea nitrogen
- Plasma allosioleucine  Plasma isoleucine  Plasma leucine  Plasma valine
- Serum creatinine  Other

Metabolite(s) of concern during second episode of dialysis- other, specify \_\_\_\_\_

Peak value of ammonia during second episode of dialysis \_\_\_\_\_

Peak value of ammonia during second episode of dialysis units

- umol/L  ug/dL  ug/L  ug/mL  g/dL

Peak value of blood urea nitrogen during second episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during second episode of dialysis units  mg/dL  mmol/L

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

Date      

Peak value of plasma alloisoleucine during second episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during second episode of dialysis units

 umol/dL umol/L mg/dL

Peak value of plasma isoleucine during second episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during second episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of plasma leucine during second episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during second episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of plasma valine during second episode of dialysis \_\_\_\_\_

Peak value of plasma valine during second episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of serum creatinine during second episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during second episode of dialysis units  umol/L  mg/dL

Peak value of other metabolite during second episode of dialysis \_\_\_\_\_

Peak value of other metabolite during second episode of dialysis units \_\_\_\_\_

Reason second episode of dialysis treatment was stopped

 Unknown Acute episode resolved  Treatment withdrawn  Death Other

Reason second episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

**Third Dialysis Treatment**

Type(s) of dialysis received during third episode

 Unknown APD/CCPD ECMO CAPD CVVH CVVD CVVHDF Hemodialysis Peritoneal dialysis

Reason for third episode of dialysis

 Unknown Hyperammonemia  MSUD Organ failure Sepsis Other

Reason for third episode of dialysis-other, specify

Start date of third episode of dialysis treatment      

Duration (in days) of third episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during third episode of dialysis

 Unknown None Ammonia Blood urea nitrogen Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine Serum creatinine Other

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

Date      

Metabolite(s) of concern during third episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during third episode of dialysis \_\_\_\_\_

Peak value of ammonia during third episode of dialysis units

 umol/L       ug/dL       ug/L       ug/mL       g/dL

Peak value of blood urea nitrogen during third episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during third episode of dialysis units     mg/dL     mmol/L

Peak value of plasma alloisoleucine during third episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during third episode of dialysis units

 umol/dL                       umol/L                       mg/dL

Peak value of plasma isoleucine during third episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during third episode of dialysis units     umol/dL     umol/L     mg/dL

Peak value of plasma leucine during third episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during third episode of dialysis units     umol/dL     umol/L     mg/dL

Peak value of plasma valine during third episode of dialysis \_\_\_\_\_

Peak value of plasma valine during third episode of dialysis units     umol/dL     umol/L     mg/dL

Peak value of serum creatinine during third episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during third episode of dialysis units     umol/L     mg/dL

Peak value of other metabolite during third episode of dialysis \_\_\_\_\_

Peak value of other metabolite during third episode of dialysis units \_\_\_\_\_

Reason third episode of dialysis treatment was stopped

 Unknown                       Acute episode resolved     Treatment withdrawn     Death Other

Reason third episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

**Fourth Dialysis Treatment**

Type(s) of dialysis received during fourth episode

 Unknown                       APD/CCPD                       ECMO                       CAPD  
 CVVH                       CVVD                       CVVHDF                       Hemodialysis  
 Peritoneal dialysis

Reason for fourth episode of dialysis

 Unknown                       Hyperammonemia     MSUD                       Organ failure                       Sepsis Other

Reason for fourth episode of dialysis-other, specify

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

Date        Start date of fourth episode of dialysis treatment      

Duration (in days) of fourth episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during fourth episode of dialysis

- Unknown                       None                       Ammonia                       Blood urea nitrogen  
 Plasma alloisoleucine    Plasma isoleucine         Plasma leucine               Plasma valine  
 Serum creatinine         Other

Metabolite(s) of concern during fourth episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during fourth episode of dialysis \_\_\_\_\_

Peak value of ammonia during fourth episode of dialysis units

- umol/L                       ug/dL                       ug/L                       ug/mL                       g/dL

Peak value of blood urea nitrogen during fourth episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during fourth episode of dialysis units    mg/dL    mmol/L

Peak value of plasma alloisoleucine during fourth episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during fourth episode of dialysis units

- umol/dL                       umol/L                       mg/dL

Peak value of plasma isoleucine during fourth episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during fourth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of plasma leucine during fourth episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during fourth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of plasma valine during fourth episode of dialysis \_\_\_\_\_

Peak value of plasma valine during fourth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of serum creatinine during fourth episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during fourth episode of dialysis units    umol/L    mg/dL

Peak value of other metabolite during fourth episode of dialysis \_\_\_\_\_

Peak value of other metabolite during fourth episode of dialysis units \_\_\_\_\_

Reason fourth episode of dialysis treatment was stopped

- Unknown                       Acute episode resolved    Treatment withdrawn    Death  
 Other

Reason fourth episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

## Fifth Dialysis Treatment

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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

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      **PKU - Transplant**Number of organ transplants received  1  2  3  4  5  >5**First Transplant**

First organ received by transplant

- Unknown     Bone marrow     Heart     Kidney     Liver     Lung  
 Pancreas     Stem Cell     Other

First organ received by transplant, other- specify \_\_\_\_\_

Age (in days) at first transplant \_\_\_\_\_

Reason for first transplant

- Unknown     Treatment of disorder     Renal failure     Liver failure  
 Heart failure     Other

Reason for first transplant, other- specify \_\_\_\_\_

Patient followed by Metabolism on an outpatient basis post first transplant  Unknown  Yes  No

Number of outpatient metabolic visits in the last year post first transplant

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

Metabolic labs monitored post first transplant  Unknown  Yes  No

Metabolic lab monitoring post first transplant: Note date(s), test(s), and normal or abnormal result(s)

Known complications during the first transplant procedure  Unknown  Yes  No

Known complications during the first transplant procedure- specify

- Clotting     Death     Major bleeding     Other

Known complications during the first transplant procedure, other- specify \_\_\_\_\_

Known complications post first transplant  Unknown  Yes  No

Known complications post first transplant- specify

- |  |   |
|--|---|
| <input type="checkbox"/> Unknown                       | <input type="checkbox"/> Cancer                         |
| <input type="checkbox"/> Cataract                      | <input type="checkbox"/> Clotting                       |
| <input type="checkbox"/> Death                         | <input type="checkbox"/> Delayed graft function         |
| <input type="checkbox"/> Diabetes mellitus             | <input type="checkbox"/> High cholesterol               |
| <input type="checkbox"/> Hypertension                  | <input type="checkbox"/> Infection                      |
| <input type="checkbox"/> Major bleeding                | <input type="checkbox"/> Osteoporosis                   |
| <input type="checkbox"/> Reappearance of heart disease | <input type="checkbox"/> Reappearance of kidney disease |

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

Date

Reappearance of liver disease       Rejection

Transplant related renal impairment     Other

Known complications post first transplant, other- specify \_\_\_\_\_

### Second Transplant

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Second organ received by transplant

Unknown     Bone marrow     Heart       Kidney       Liver       Lung

Pancreas     Stem Cell     Other

Second organ received by transplant, other- specify \_\_\_\_\_

Age (in days) at second transplant \_\_\_\_\_

Reason for second transplant

Unknown       Treatment of disorder     Renal failure       Liver failure

Heart failure       Other

Reason for second transplant, other- specify \_\_\_\_\_

Patient followed by Metabolism on an outpatient basis post second transplant     Unknown     Yes     No

Number of outpatient metabolic visits in the last year post transplant

1     2     3     4     5     6     7     8     9     10     >10

Metabolic labs monitored post second transplant     Unknown     Yes     No

Metabolic lab monitoring post second transplant: Note date(s), test(s), and normal or abnormal result(s)

Complications during the second transplant procedure     Unknown     Yes     No

Complications during the second transplant procedure- specify

Clotting       Death       Major bleeding       Other

Complications during the second transplant procedure, other- specify \_\_\_\_\_

Complications post second transplant     Unknown     Yes     No

Complications post second transplant- specify

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

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

- 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 fifth transplant, other- specify \_\_\_\_\_

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

Date

Provide details for any other transplants not listed above

**Comments**

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Transplant comments