

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

Date      **BIOPT-REG - Intake Demographics**

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

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**Patient condition category  Amino acid disordersSpecify amino acid disorder diagnosis for the patient  Biopterin defect in cofactor regeneration (BIOPT REG)

BIOPT-REG Diagnosis

- Dihydropteridine reductase (DHPR) deficiency  Carbinolamine-4a-dehydratase (PCD) deficiency

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## Patient disorder identification method

- 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)

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- 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)

## 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

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- Lithuania      Malta      Netherlands      Norway      Poland
- Romania      Serbia      Slovakia      Slovenia      Spain
- Sweden      Switzerland      Ukraine      United Kingdom      Other

Ancestral Origin-Europe-Italy    Unknown    Sicily

Ancestral Origin-Europe-Romania    Unknown    Transylvania

Ancestral Origin-Europe-United Kingdom    Unknown    England    Nothern Ireland    Scotland    Wales

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

Ancestral Origin-North America

- Unknown      Aleutian Islands      Canada      Dominican Republic    Honduas
- 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    Other

Ancestral 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**

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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

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- |                          |                          |                          |                          |                          |                          |
|--------------------------|--------------------------|--------------------------|--------------------------|--------------------------|--------------------------|
| <input type="radio"/> CA | <input type="radio"/> CO | <input type="radio"/> CT | <input type="radio"/> DE | <input type="radio"/> DC | <input type="radio"/> FL |
| <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

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

Date      ***BIOPT-REG - Intake Family History*****Family History**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 |M|M| |D|D| |Y|Y|

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

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

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

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

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

Sibling 4: Method of diagnosis

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

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

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

Sibling 4: Results of newborn screening for this condition

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

Sibling 4: Specify the type of diagnostic tests performed

 Unknown  Biochemical  Molecular  Imaging  Other

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

Sibling 4: Deceased  Unknown  Yes  No

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

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

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

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

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

Sibling 5: Method of diagnosis

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

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

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



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

Date      

Sibling 5: Results of newborn screening for this condition

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

Sibling 5: Specify the type of diagnostic tests performed

 Unknown       Biochemical       Molecular       Imaging       Other

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

Sibling 5: Deceased     Unknown     Yes     No

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

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

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

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

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

Sibling 6: Method of diagnosis

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

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

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

Sibling 6: Results of newborn screening for this condition

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

Sibling 6: Specify the type of diagnostic tests performed

 Unknown       Biochemical       Molecular       Imaging       Other

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

Sibling 6: Deceased     Unknown     Yes     No

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

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

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

Sibling 7: Affected with this condition     Unknown     Yes     No

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

Date      Sibling 7: Enrolled in this study  Unknown  Yes  No

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

Sibling 7: Method of diagnosis

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

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

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

Sibling 7: Results of newborn screening for this condition

Unavailable  Presumptive positive  Negative/Normal  Borderline

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

Sibling 7: Specify the type of diagnostic tests performed

Unknown  Biochemical  Molecular  Imaging  Other

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

Sibling 7: Deceased  Unknown  Yes  No

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

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

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

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

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

Sibling 8: Method of diagnosis

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

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

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

Sibling 8: Results of newborn screening for this condition

Unavailable  Presumptive positive  Negative/Normal  Borderline

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

Sibling 8: Specify the type of diagnostic tests performed

Unknown  Biochemical  Molecular  Imaging  Other

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

Sibling 8: Deceased  Unknown  Yes  No

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

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

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

Biological mother: Method of diagnosis

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

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

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

Biological mother: Results of newborn screening for this condition

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

Biological mother: Specify the type of diagnostic tests performed

 Unknown  Biochemical  Molecular  Imaging  Other

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

Biological mother: Deceased  Unknown  Yes  No

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

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

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

Biological father: Method of diagnosis

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

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

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

Biological father: Results of newborn screening for this condition

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

Biological father: Specify the type of diagnostic tests performed

 Unknown  Biochemical  Molecular  Imaging  Other

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

Biological father: Deceased  Unknown  Yes  No

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

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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

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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      

## ***BIOPT-REG - Intake Past Health History***

### **Prenatal History**

Prenatal diagnosis done for this condition    Unknown    Yes    No

Form of prenatal diagnosis

- Unknown                       Amniocentesis                       Chorionic villus sampling (CVS)  
 Fetal blood

Amniocentesis diagnosis    Biochemical/enzyme    DNA

Chorionic villus sampling (CVS) diagnosis    Biochemical/enzyme    DNA

Issues concerning mother's pregnancy with this patient

- Unknown                       None                       Pregnancy complications  
 Assisted reproduction

Pregnancy complications/risk factors

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

Type of prenatal substance exposure

- Unknown                       Alcohol                       Tobacco                       Illicit drugs  
 Harmful chemicals    Known teratogens

Maternal treatment for affected fetus    Unknown    None    Yes    No

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Date      

Type of maternal treatment for affected fetus

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

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

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

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

## Pregnancy

---

Patient has biological children  Unknown  Yes  No

Please complete the pregnancy form.

Patient is pregnant  Unknown  Yes  No

Please complete the pregnancy form.

## Neonatal History

---

Patient's birth was a result of multiple gestation pregnancy

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

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

Congenital anomalies  Unknown  Yes  No

Type of congenital anomalies

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



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

Date      

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

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

Neonatal complications    Unknown    Yes    No

Type of neonatal complications

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

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

Type of neonatal nutrition

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

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

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

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

Birth head circumference units    cm    in

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

Date      

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

Birth length units  cm  in

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

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

---

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

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

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

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

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

Brain MRI prior to intake  Unknown  Yes  No

Brain MRI date

Brain MRI results  Unknown  Normal  Abnormal

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

Date

Brain MRI comments

[Empty text box for Brain MRI comments]

Neurological imaging prior to intake Unknown Yes No

Neurological imaging date

Neurological imaging results Unknown Normal Abnormal

Neurological imaging comments

[Empty text box for Neurological imaging comments]

History of a seizure disorder Unknown Yes No

Patient has biospecimen stored Unknown Yes No

Type of patient biospecimen Blood Urine Other tissue

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

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

**Phenylalanine Level History**

---

Average blood phenylalanine level age 0 < 1 year \_\_\_\_\_

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

Number of phenylalanine levels measured age 0 < 1 year \_\_\_\_\_

Average tyrosine level age 0 < 1 year \_\_\_\_\_

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

Number of tyrosine levels measured age 0 < 1 year \_\_\_\_\_

Average phenylalanine intake age 0 < 1 year \_\_\_\_\_

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

Average blood phenylalanine level age 1 < 2 years \_\_\_\_\_

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

Number of phenylalanine levels measured age 1 < 2 years \_\_\_\_\_

Average tyrosine level age 1 < 2 years \_\_\_\_\_

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

Number of tyrosine levels measured age 1 < 2 years \_\_\_\_\_

Average phenylalanine intake age 1 < 2 years \_\_\_\_\_

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

Average blood phenylalanine level age 2 < 3 years \_\_\_\_\_

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

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

Date      

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

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

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

Date      

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 \_\_\_\_\_

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

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

Date      

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 \_\_\_\_\_

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 \_\_\_\_\_

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

Date      

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 \_\_\_\_\_

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 \_\_\_\_\_



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

Date      

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 \_\_\_\_\_

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

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

Average phenylalanine intake age &gt; 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  NoPatient/primary caregiver was given a sick day plan specific to this condition  Unknown  Yes  No

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

 Unknown Yes No**Comments**

---

Intake past health history comments

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

Date      

## ***BIOPT-REG - 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      

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

Date third newborn screen collected      

Reason for third newborn screen

- Unsatisfactory     Borderline Result     NICU Protocol     TPN     Transfused  
 State Mandate     Other

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

Phenylalanine on third newborn screen \_\_\_\_\_

Phenylalanine on third newborn screen units     umol/L

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

Tyrosine on third newborn screen \_\_\_\_\_

Tyrosine on third newborn screen units     umol/L

Other result/report on third newborn screen

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

## Newborn Hearing Screen

---

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

Recommendation

- Repeat hearing screen     Referral for diagnostic testing     Risk factor monitoring only  
 Refused further action     No further action required

Date of Audiological Diagnostic Evaluation      Right ear: Diagnosis: Hearing loss?     Yes     No

Right ear: Diagnosis: Degree of hearing loss

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

Right ear: Diagnosis: Type of hearing loss

- Sensorineural     Conductive  
 Mixed     Auditory neuropathy/Auditory dys-synchrony  
 Other

Left ear: Diagnosis: Hearing loss?     Yes     No

Left ear: Diagnosis: Degree of hearing loss

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"/>

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

Left ear: Diagnosis: Type of hearing loss

- Sensorineural       Conductive  
 Mixed       Auditory neuropathy/Auditory dys-synchrony  
 Other

### Comments

---

Intake newborn screening comments

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

Date      ***BIOPT-REG - Intake Initial Testing***

Patient status at time of NBS reporting to specialty center

 Unknown                       Well                                       Symptomatic                               Deceased

Patient symptoms at time of initial contact

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

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

Date      

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

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

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

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

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

Date      

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

## Diagnostic Testing

---

### Diagnostic labs performed

- |   |   |
|---|---|
| <input type="checkbox"/> Unknown  | <input type="checkbox"/> None   |
| <input type="checkbox"/> Blood spot phenylalanine                         | <input type="checkbox"/> Blood spot tyrosine                            |
| <input type="checkbox"/> CSF 5-hydroxyindoleacetic acid (5-HIAA)          | <input type="checkbox"/> CSF 5-methyltetrahydrofolic (5-MTHF) acid      |
| <input type="checkbox"/> CSF homovanillic acid                            | <input type="checkbox"/> CSF pterins                                    |
| <input type="checkbox"/> Dihydropteridine reductase enzyme assay          | <input type="checkbox"/> Plasma amino acid profile                      |
| <input type="checkbox"/> Plasma phenylalanine                             | <input type="checkbox"/> Plasma tyrosine                                |
| <input type="checkbox"/> Pterin-4a-carbinolamine dehydratase enzyme assay | <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 \_\_\_\_\_

CSF 5-hydroxyindoleacetic acid (5-HIAA)     Unknown     Within normal limits     Abnormal

CSF 5-hydroxyindoleacetic acid (5-HIAA) significance     Diagnostic     Non-diagnostic

CSF 5-hydroxyindoleacetic acid (5-HIAA) value \_\_\_\_\_

CSF 5-hydroxyindoleacetic acid (5-HIAA) units     umol/L     nmol/L     pmol/mL     ng/mL

CSF 5-hydroxyindoleacetic acid (5-HIAA) reference range \_\_\_\_\_

CSF 5-methyltetrahydrofolic acid (5-MTHF)     Unknown     Within normal limits     Abnormal

CSF 5-methyltetrahydrofolic acid (5-MTHF) significance     Diagnostic     Non-diagnostic

CSF 5-methyltetrahydrofolic acid (5-MTHF) value \_\_\_\_\_

CSF 5-methyltetrahydrofolic acid (5-MTHF) units     umol/L     nmol/L     pmol/mL     ng/mL

CSF 5-methyltetrahydrofolic acid (5-MTHF) reference range \_\_\_\_\_

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

Date      CSF homovanillic acid  Unknown  Within normal limits  AbnormalCSF homovanillic acid significance  Diagnostic  Non-diagnostic

CSF homovanillic acid value \_\_\_\_\_

CSF homovanillic acid units  umol/L  nmol/L  pmol/mL  ng/mL

CSF homovanillic acid reference range \_\_\_\_\_

CSF pterins  Biopterin  Dihydrobiopterin  NeopterinCSF biopterin-number of samples  1  2  3

CSF biopterin percentage \_\_\_\_\_

CSF biopterin value \_\_\_\_\_

CSF biopterin units  umol/L  nmol/L  pmol/mL  ng/mL

CSF biopterin test date \_\_\_\_\_

CSF biopterin percentage \_\_\_\_\_

CSF biopterin value \_\_\_\_\_

CSF biopterin units  umol/L  nmol/L  pmol/mL  ng/mL

CSF biopterin test date \_\_\_\_\_

CSF biopterin percentage \_\_\_\_\_

CSF biopterin value \_\_\_\_\_

CSF biopterin units  umol/L  nmol/L  pmol/mL  ng/mL

CSF biopterin test date \_\_\_\_\_

CSF dihydrobiopterin-number of samples  1  2  3

CSF dihydrobiopterin percentage \_\_\_\_\_

CSF dihydrobiopterin value \_\_\_\_\_

CSF dihydrobiopterin units  umol/L  nmol/L  pmol/mL  ng/mL

CSF dihydrobiopterin test date \_\_\_\_\_

CSF dihydrobiopterin percentage \_\_\_\_\_

CSF dihydrobiopterin value \_\_\_\_\_

CSF dihydrobiopterin units  umol/L  nmol/L  pmol/mL  ng/mL

CSF dihydrobiopterin test date \_\_\_\_\_

CSF dihydrobiopterin percentage \_\_\_\_\_

CSF dihydrobiopterin value \_\_\_\_\_

CSF dihydrobiopterin units  umol/L  nmol/L  pmol/mL  ng/mL

CSF dihydrobiopterin test date \_\_\_\_\_

CSF neopterin-number of samples  1  2  3

CSF neopterin percentage \_\_\_\_\_

CSF neopterin value \_\_\_\_\_



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

Date

CSF neopterin units umol/L nmol/L pmol/mL ng/mL

CSF neopterin test date\_\_\_\_\_

CSF neopterin percentage\_\_\_\_\_

CSF neopterin value\_\_\_\_\_

CSF neopterin units umol/L nmol/L pmol/mL ng/mL

CSF neopterin test date\_\_\_\_\_

CSF neopterin percentage\_\_\_\_\_

CSF neopterin value\_\_\_\_\_

CSF neopterin units umol/L nmol/L pmol/mL ng/mL

CSF neopterin test date\_\_\_\_\_

Number of times dihydropteridine reductase in RBCs enzyme assay done 1 2

Dihydropteridine reductase in RBCs enzyme assay Unknown Within normal limits Abnormal

Dihydropteridine reductase in RBCs enzyme assay significance Diagnostic Non-diagnostic

Dihydropteridine reductase in RBCs enzyme assay value\_\_\_\_\_

Dihydropteridine reductase in RBCs enzyme assay units nmol/min/mg Hb

Dihydropteridine reductase in RBCs enzyme assay reference range\_\_\_\_\_

Second dihydropteridine reductase in RBCs enzyme assay

Unknown Within normal limits Abnormal

Second dihydropteridine reductase in RBCs enzyme assay significance

Diagnostic Non-diagnostic

Second dihydropteridine reductase in RBCs enzyme assay value\_\_\_\_\_

Second dihydropteridine reductase in RBCs enzyme assay units nmol/min/mg Hb

Second dihydropteridine reductase in RBCs enzyme assay 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

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

Date      Plasma tyrosine significance  Diagnostic  Non-diagnostic

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

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

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

Number of times Pterin-4a-carbinolamine dehydratase enzyme assay done  1  2Pterin-4a-carbinolamine dehydratase enzyme assay  Unknown  Within normal limits  AbnormalPterin-4a-carbinolamine dehydratase enzyme assay significance  Diagnostic  Non-diagnostic

Pterin-4a-carbinolamine dehydratase enzyme assay value \_\_\_\_\_

Pterin-4a-carbinolamine dehydratase enzyme assay units \_\_\_\_\_

Pterin-4a-carbinolamine dehydratase enzyme assay reference range \_\_\_\_\_

Second Pterin-4a-carbinolamine dehydratase enzyme assay

 Unknown  Within normal limits  Abnormal

Second Pterin-4a-carbinolamine dehydratase enzyme assay significance

 Diagnostic  Non-diagnostic

Second Pterin-4a-carbinolamine dehydratase enzyme assay value \_\_\_\_\_

Second Pterin-4a-carbinolamine dehydratase enzyme assay units \_\_\_\_\_

Second Pterin-4a-carbinolamine dehydratase enzyme assay 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 \_\_\_\_\_

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"/>

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 \_\_\_\_\_

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 \_\_\_\_\_

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

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

Other diagnostic lab comments

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

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

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

BH4 loading test done  Unknown  Yes  No

BH4 loading test dose \_\_\_\_\_

BH4 loading test dose units  mg  mg/kgBH4 loading test results  Unknown  Within normal limits  AbnormalBH4 loading test significance  Diagnostic  Non-diagnosticCombined BH4/phenylalanine loading test result  Unknown  Within normal limits  AbnormalCombined BH4/phenylalanine loading test significance  Diagnostic  Non-diagnostic

## 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                  |  |   |

Reason genotyping was done

- |                                  |   |   |   |
|----------------------------------|---|---|---|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Diagnosis confirmation | <input type="checkbox"/> Modifier genes | <input type="checkbox"/> Pharmacogenetics |
| <input type="checkbox"/> Other   |   |   |   |

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

Reason genotyping was not done

- |   |   |   |
|---|---|---|
| <input type="checkbox"/> Unknown        | <input type="checkbox"/> Clinician deemed unnecessary | <input type="checkbox"/> Not covered by insurance |
| <input type="checkbox"/> Parent refusal | <input type="checkbox"/> Test not available           | <input type="checkbox"/> Other                    |

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

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

Gene(s) associated with BIOPT (REG)  PCBD1  QDPR  Other

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

PCBD1: Specify allele 1 \_\_\_\_\_

PCBD1: Specify allele 2 \_\_\_\_\_

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"/>

QDPR: Specify allele 1 \_\_\_\_\_

QDPR: 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

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Initial testing comments

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

Date      **BIOPT-REG - 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**

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Patient has new biological sibling since last visit     Unknown     Yes     No

Complete sibling history questions on the Family History form.

**Comments**

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Visit demographics and history comments

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

Date      ***BIOPT-REG - 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 unrelatedSick 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    OtherPatient 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 unrelatedSick 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    OtherPatient 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 unrelatedSick 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    OtherPatient was admitted to the hospital as a result of sick visit 4    Unknown    Yes    No

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

Date      

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 unrelatedSick 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  OtherPatient 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 unrelatedSick 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  OtherPatient 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      

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

ICD-9 codes for sick visit 9 known  Yes  No

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

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

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

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

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

Location for sick visit 10

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

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

ICD-9 codes for sick visit 10 known  Yes  No

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

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

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

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

New symptoms developed since last visit

- |   |   |  |                                       |
|---|---|--|---------------------------------------|
| <input type="checkbox"/> Unknown              | <input type="checkbox"/> None             | <input type="checkbox"/> Athetosis       | <input type="checkbox"/> Chorea       |
| <input type="checkbox"/> Dystonia             | <input type="checkbox"/> Eczema           | <input type="checkbox"/> Hypersalivation | <input type="checkbox"/> Hypertonia   |
| <input type="checkbox"/> Hypotonia            | <input type="checkbox"/> Extremity tremor | <input type="checkbox"/> Lethargy        | <input type="checkbox"/> Opisthotonus |
| <input type="checkbox"/> Orofacial dyskinesia | <input type="checkbox"/> Pneumonia        | <input type="checkbox"/> Seizures        |                                       |

New symptoms developed since last visit-Type of seizures  Myoclonic  Tonic clonic  Other

New symptoms developed since last visit-Type of seizures-other, specify \_\_\_\_\_

Brain abnormalities noted since last visit  Unknown  Yes  No

Type of brain abnormalities noted since last visit

- |  |  |
|--|--|
| <input type="checkbox"/> Abnormal EEG              | <input type="checkbox"/> Abnormal vascular proliferation in cortex |
| <input type="checkbox"/> Basal ganglia abnormality | <input type="checkbox"/> Brain atrophy                             |
| <input type="checkbox"/> Calcification             | <input type="checkbox"/> Thalamus abnormality                      |
| <input type="checkbox"/> White matter abnormality  | <input type="checkbox"/> Other                                     |

Type of brain abnormalities noted since last visit-Other, specify \_\_\_\_\_

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

Date

**Procedures**

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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

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

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"/>

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      

## ***BLOPT-REG - 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     inHow height/length measured     Unknown     Supine     Standing

Weight \_\_\_\_\_

Weight units     lbs     kg     gm     oz

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

Visit findings

- Unknown       None       Evidence of athetosis  
 Evidence of chorea       Evidence of dystonia       Evidence of eczema  
 Evidence of hypersalivation       Evidence of hypertonia       Evidence of hypotonia  
 Evidence of extremity tremor       Evidence of lethargy       Evidence of opisthotonus  
 Evidence of orofacial dyskinesia     Other

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

### **Comments**

Visit findings comments





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  AtypicalSeverity of atypical development  Unknown  Mild delay  Moderate delay  Severe delay

Developmental milestones that were achieved in a typical order and timeframe

 Unknown  None  Cognitive  Fine motor  Gross motor Social-emotional  Speech-language  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

\_\_\_\_\_

Progressive loss of developmental milestones since last visit  Unknown  Yes  NoNeuropsychometric evaluation performed since last visit  Unknown  Yes  NoOverall neuropsychometric impression  Above average  Average  Below average  In progressPatient has mental health concerns  Unknown  Yes  No

Referred for further mental health assessment

 Unknown  Yes  
 No  Previously referred Family declined further assessments

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

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

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

\_\_\_\_\_

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

Behavioral concerns    Unknown    Yes    No

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

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

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

### Education

Special education assessment recommended

Unknown    Yes

No    Special education services already received

Reason special education services received

<input type="checkbox"/> Unknown	<input type="checkbox"/> Cognitive disability	<input type="checkbox"/> Developmental delay
<input type="checkbox"/> Fine motor disability	<input type="checkbox"/> Gross motor disability	<input type="checkbox"/> Learning disability
<input type="checkbox"/> Social-emotional disability	<input type="checkbox"/> Speech/Language disability	<input type="checkbox"/> Other health impairment (OHI)
<input type="checkbox"/> 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

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

Date

- Specific learning disabilities
- Speech or language impairments
- Traumatic brain injury
- Other health disabilities

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

**Comments**

---

Visit ancillary care comments

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

Date      ***BIOPT-REG - 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"/> Phenylalanine                       | <input type="checkbox"/> Plasma amino acid profile                        |
| <input type="checkbox"/> Plasma essential fatty acid profile | <input type="checkbox"/> Plasma total homocysteine                        |
| <input type="checkbox"/> Prolactin                           | <input type="checkbox"/> Pterin-4a-carbinolamine dehydratase enzyme assay |
| <input type="checkbox"/> Selenium                            | <input type="checkbox"/> Serum vitamin B12                                |
| <input type="checkbox"/> Total 25-hydroxyvitamin D           | <input type="checkbox"/> Tyrosine   |
| <input type="checkbox"/> Urine methylmalonic acid            | <input type="checkbox"/> Urine organic acids                              |
| <input type="checkbox"/> Urine pterins                       | <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 \_\_\_\_\_

Phenylalanine    Within normal limits    Abnormal    In progress    Results unavailable

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

Phenylalanine units    umol/dL    umol/L    mg/dL

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

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

Plasma amino acid comments

Plasma essential fatty acid profile

- |  |   |   |
|--|---|---|
| <input type="checkbox"/> Unknown       | <input type="checkbox"/> Alpha linolenic acid (C18:3w3) | <input type="checkbox"/> Docosahexaenoic acid (DHA) |
| <input type="checkbox"/> EPA (C20:5w3) | <input type="checkbox"/> Linoleic acid (C18:2w6)        | <input type="checkbox"/> Triene/tetraene ratio      |

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

Date

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

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

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

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

Plasma total homocysteine level units  umol/L  mg/L

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

Prolactin  Within normal limits  Abnormal  In progress  Results unavailable

Prolactin value \_\_\_\_\_

Prolactin units  pmol/L  ng/mL  ug/L

Prolactin reference range \_\_\_\_\_

Please complete information about the Pterin-4a-carbinolamine dehydratase enzyme assay on the Intake Initial Testing form.

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

Urine organic acid comments

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

Date

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

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

Other visit lab    Within normal limits    Abnormal    In progress    Results unavailable



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

Date

Other visit lab comments

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

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

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

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

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

### Chemistry Labs

---

Chemistry labs associated with this visit

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

Arterial blood gas    Within normal limits    Abnormal    In progress    Results unavailable

Arterial blood gas comments

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

Arterial blood gas units    mmHg    mEq/L

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

Anion gap    Within normal limits    Abnormal    In progress    Results unavailable

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

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

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

Ammonia    Within normal limits    Abnormal    In progress    Results unavailable

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

Ammonia units    umol/L    ug/dL

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

Calcium    Within normal limits    Abnormal    In progress    Results unavailable

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

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

Date      Calcium units  mmol/L  mg/dL

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

Chloride  Within normal limits  Abnormal  In progress  Results unavailable

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

Chloride units  mmol/L  mg/L

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

Carbon dioxide  Within normal limits  Abnormal  In progress  Results unavailable

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

Carbon dioxide units  mEq/L  mmol/L

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

Glucose  Within normal limits  Abnormal  In progress  Results unavailable

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

Glucose units  mmol/L  mg/dL

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

Potassium  Within normal limits  Abnormal  In progress  Results unavailable

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

Potassium units  mEq/L  mmol/L

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

Lactate dehydrogenase  Within normal limits  Abnormal  In progress  Results unavailable

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

Lactate dehydrogenase units  U/L

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

Magnesium  Within normal limits  Abnormal  In progress  Results unavailable

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

Magnesium units  mmol/L  mg/dL

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

Sodium  Within normal limits  Abnormal  In progress  Results unavailable

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

Sodium units  mEq/L  mmol/L

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

Phosphorous  Within normal limits  Abnormal  In progress  Results unavailable

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

Phosphorous units  mmol/L  mg/dL

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

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

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

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

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

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

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

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

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

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

Uric acid  Within normal limits  Abnormal  In progress  Results unavailable

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

Uric acid units  umol/L  mg/dL

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

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

Other chemistry labs  Within normal limits  Abnormal  In progress  Results unavailable

Other chemistry lab comments

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

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

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

### Hematology Labs

---

Hematology labs associated with this visit

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

Fibrinogen  Within normal limits  Abnormal  In progress  Results unavailable

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

Fibrinogen units  umol/L  mg/dL

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

Hematocrit  Within normal limits  Abnormal  In progress  Results unavailable

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

Hematocrit units  %  Proportion of total hemoglobin

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

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

Date      Hemoglobin  Within normal limits  Abnormal  In progress  Results unavailable

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

Hemoglobin units  g/dL  g/L

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

International Normalized Ratio (INR)

 Within normal limits  Abnormal  In progress  Results unavailable

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

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

Partial Thromboplastin Time (PTT)

 Within normal limits  Abnormal  In progress  Results unavailable

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

Partial Thromboplastin Time (PTT) units  seconds

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

Prothrombin time test  Within normal limits  Abnormal  In progress  Results unavailable

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

Prothrombin time test units  seconds

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

Peripheral blood smear  Within normal limits  Abnormal  In progress  Results unavailable

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

Peripheral blood smear units  platelet/RBC/WBC count

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

Platelet count  Within normal limits  Abnormal  In progress  Results unavailable

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

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

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

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

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

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

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

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

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

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

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

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

Other hematology labs  Within normal limits  Abnormal  In progress  Results unavailable

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

Date      

Other hematology lab comments

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

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

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

## Liver Labs

---

Liver labs associated with this visit

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

Albumin     Within normal limits     Abnormal     In progress     Results unavailable

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

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

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

Alkaline phosphatase     Within normal limits     Abnormal     In progress     Results unavailable

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

Alkaline phosphatase units     U/L     ukat/L

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

Alanine aminotransferase     Within normal limits     Abnormal     In progress     Results unavailable

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

Alanine aminotransferase units     U/L     ukat/L

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

Aspartate aminotransferase     Within normal limits     Abnormal     In progress     Results unavailable

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

Aspartate aminotransferase units     U/L     ukat/L

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

Direct bilirubin     Within normal limits     Abnormal     In progress     Results unavailable

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

Direct bilirubin units

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

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

Date |||||

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

Gamma-glutamyl transpeptidase

 Within normal limits  Abnormal  In progress  Results unavailable

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

Gamma-glutamyl transpeptidase units  IU/L  U/L

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

Globulin  Within normal limits  Abnormal  In progress  Results unavailable

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

Globulin units  g/dL  g/L

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

Prealbumin  Within normal limits  Abnormal  In progress  Results unavailable

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

Prealbumin units  mg/dL  mg/L

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

Total bilirubin  Within normal limits  Abnormal  In progress  Results unavailable

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

Total bilirubin units  umol/L  mg/dL

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

Total protein  Within normal limits  Abnormal  In progress  Results unavailable

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

Total protein units  g/dL  g/L

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

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

Other liver labs  Within normal limits  Abnormal  In progress  Results unavailable

Other liver lab comments

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

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

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

### Renal Labs

---

Renal labs associated with this visit

- Unknown  None  
 Blood urea nitrogen  Serum creatinine

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"/>
--

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 \_\_\_\_\_

### 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                          |

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

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

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

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

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

Creatine phosphokinase  Within normal limits  Abnormal  In progress  Results unavailable

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

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

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

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

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

C-reactive protein units  nmol/L  mg/dL

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

Erythrocyte sedimentation rate  Within normal limits  Abnormal  In progress  Results unavailable

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

Erythrocyte sedimentation rate units  mm/h

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

Ferritin  Within normal limits  Abnormal  In progress  Results unavailable

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

Ferritin units  pmol/L  ng/mL

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

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

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

Thyroid-stimulating hormone units  mIU/L

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

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

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

Thyroxine (free) units  pmol/L  ng/mL

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

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

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

Thyroxine (total) units  pmol/L  ng/mL

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

Transferrin  Within normal limits  Abnormal  In progress  Results unavailable

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

Transferrin units  mg/dL  g/L



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

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      ***BIOPT-REG - Visit Studies Other*****Home Monitoring**Home monitoring recommended  Unknown  Yes  NoHome monitoring done since the last outpatient visit  Unknown  Yes  No

Type of home monitoring

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

**Glucose**Frequency of glucose home monitoring  Unknown  Only when symptomatic  Routinely

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

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

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

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

 mmol/L  mg/dL**Branched Chain Amino Acids**

Type of branched chain amino acid home monitoring

- Unknown  Alloisoleucine  Isoleucine  Leucine  Valine

Sample type of branched chain amino acid home monitoring

- Unknown  Filter paper blood  Plasma

Frequency of branched chain amino acid home monitoring

- Unknown  Only when symptomatic  Routinely

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

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

**Alloisoleucine**

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

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

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

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

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

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

Date      

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

**Isoleucine**

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

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

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

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

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

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

**Leucine**

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

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

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

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

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

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

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

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

**Valine**

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Date      

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

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

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

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

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

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

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

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

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

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

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

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

**Dinitrophenylhydrazine**

Frequency of dinitrophenylhydrazine home monitoring

 Unknown  Only when symptomatic  Routinely

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

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

Result of dinitrophenylhydrazine home monitoring samples

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

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

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

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

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

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

**Other**

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

Frequency of other home monitoring  Unknown  Only when symptomatic  Routinely

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

Date

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

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

**Other Studies**

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Physiological tests associated with this visit

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

**Electrocardiogram**

Electrocardiogram date

Electrocardiogram findings of note

**Electroencephalography**

Electroencephalography date

Electroencephalography findings of note

**Electromyography**

Electromyography date

Electromyography findings of note

**Holter Monitoring**

Holter monitoring date

Holter monitoring findings of note

**Cardiac Stress Test**

Cardiac stress test date

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

Date

Cardiac stress test findings of note

**Imaging Studies**

Imaging studies associated with this visit

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

Abdominal imaging

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

Abdominal findings of note

Cardiac Imaging

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

Cardiac findings of note

Musculoskeletal imaging

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

Musculoskeletal findings of note

Neurological imaging

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

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

Positron emission tomography (PET) scan Abn

Evidence of abnormal myelination on CNS imaging     Unknown     Yes     No

Neurological findings of note

Renal/Pelvic/Genital imaging

Unknown

Genitogram WNL

Genitogram Abn

Nuclear medicine dimercapto succinic acid (DMSA) renal scan WNL

Nuclear medicine dimercapto succinic acid (DMSA) renal scan Abn

Pelvic ultrasound WNL

Pelvic ultrasound Abn

Renal ultrasound WNL

Renal ultrasound Abn

Testicular ultrasound WNL

Testicular ultrasound Abn

VCUG WNL

VCUG Abn

Other

Renal/pelvic/genital findings of note

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

Other imaging studies date

Other imaging studies explanation

**Comments**

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

Visit studies-other comments



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

Date      

## ***BIOPT-REG - 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"/> 5-hydroxy tryptophan (5-HTP) |
| <input type="checkbox"/> Folinic acid                | <input type="checkbox"/> L-deprenyl | <input type="checkbox"/> L-dopa                       |
| <input type="checkbox"/> Sapropterin dihydrochloride | <input type="checkbox"/> Other      |   |

#### **5-hydroxy tryptophan (5-HTP)**

5-hydroxy tryptophan (5-HTP) route recommended/prescribed    Unknown    Feeding tube    Oral

5-hydroxy tryptophan (5-HTP) dose recommended/prescribed \_\_\_\_\_

5-hydroxy tryptophan (5-HTP) dose recommended/prescribed units    mg

5-hydroxy tryptophan (5-HTP) frequency recommended/prescribed

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

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

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

5-hydroxy tryptophan (5-HTP) frequency recommended/prescribed-other, specify \_\_\_\_\_

5-hydroxy tryptophan (5-HTP) taken as recommended/prescribed     Unknown     Yes     No

Actual 5-hydroxy tryptophan (5-HTP) dose reported \_\_\_\_\_

Actual 5-hydroxy tryptophan (5-HTP) dose reported units     mg

Actual 5-hydroxy tryptophan (5-HTP) frequency reported

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

Actual 5-hydroxy tryptophan (5-HTP) frequency reported-other, specify \_\_\_\_\_

Reason 5-hydroxy tryptophan (5-HTP) is not taken as recommended/prescribed \_\_\_\_\_

Method of payment for 5-hydroxy tryptophan (5-HTP)

- 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

### **Folinic acid**

Folinic acid route recommended/prescribed     Unknown     Feeding tube     Oral

Folinic acid dose recommended/prescribed \_\_\_\_\_

Folinic acid dose recommended/prescribed units     mg     ug

Folinic acid frequency recommended/prescribed

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

Folinic acid frequency recommended/prescribed-other, specify \_\_\_\_\_

Folinic acid taken as recommended/prescribed     Unknown     Yes     No

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"/>

Actual folinic acid dose reported \_\_\_\_\_

Actual folinic acid dose reported units  mg  ug

Actual folinic acid frequency reported

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

Actual folinic acid frequency reported-other, specify \_\_\_\_\_

Reason folinic acid is not taken as recommended/prescribed \_\_\_\_\_

Method of payment for folinic acid

- 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

**L-deprenyl**

L-deprenyl formulation recommended/prescribed

- Unknown
- 10% carbidopa
- 25% carbidopa
- Slow release

L-deprenyl route recommended/prescribed  Unknown  Feeding tube  Oral

L-deprenyl dose recommended/prescribed \_\_\_\_\_

L-deprenyl dose recommended/prescribed units  mg

L-deprenyl frequency recommended/prescribed  Unknown  Once/day  Twice/day  Other

L-deprenyl frequency recommended/prescribed-other, specify \_\_\_\_\_

L-deprenyl taken as recommended/prescribed  Unknown  Yes  No

Actual L-deprenyl dose reported \_\_\_\_\_

Actual L-deprenyl dose reported units  mg

Actual L-deprenyl frequency reported  Unknown  Once/day  Twice/day  Other

Actual L-deprenyl frequency reported-other, specify \_\_\_\_\_

Reason L-deprenyl is not taken as recommended/prescribed \_\_\_\_\_

Method of payment for L-deprenyl

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

- 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

**L-dopa**

L-dopa formulation recommended/prescribed

- Unknown
- 10% carbidopa
- 25% carbidopa
- Slow release

L-dopa route recommended/prescribed  Unknown  Feeding tube  Oral

L-dopa dose recommended/prescribed \_\_\_\_\_

L-dopa dose recommended/prescribed units  mg

L-dopa frequency recommended/prescribed

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

L-dopa frequency recommended/prescribed-other, specify \_\_\_\_\_

L-dopa taken as recommended/prescribed  Unknown  Yes  No

Actual L-dopa dose reported \_\_\_\_\_

Actual L-dopa dose reported units  mg

Actual L-dopa frequency reported

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

Actual L-dopa frequency reported-other, specify \_\_\_\_\_

Reason L-dopa is not taken as recommended/prescribed \_\_\_\_\_

Method of payment for L-dopa

- Unknown
- None
- Commercial/private
- Medicaid

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

Date      

- 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

\_\_\_\_\_

Sapropterin dihydrochloride taken as recommended/prescribed     Unknown     Yes     No

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

Actual sapropterin dihydrochloride dose reported units     g     mgActual 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

Patient Name _____ Date  M M   D D   Y Y
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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

**Other Treatment**

Other treatment recommend/prescribed \_\_\_\_\_

Other treatment route recommended/prescribed \_\_\_\_\_

Other treatment dose recommended/prescribed \_\_\_\_\_

Other treatment dose recommended/prescribed units \_\_\_\_\_

Other treatment frequency recommended/prescribed \_\_\_\_\_

Other treatment taken as recommended/prescribed     Unknown     Yes     No

Actual other treatment dose reported \_\_\_\_\_

Actual other treatment dose reported units \_\_\_\_\_

Actual other treatment frequency reported \_\_\_\_\_

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

Method of payment for other treatment

- Unknown
- None
- Commercial/private
- Medicaid
- Medicare
- Military
- Newborn screening funds

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

Date

- 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      ***BIOPT-REG - 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" 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 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"/>
--

- 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 \_\_\_\_\_

**Modified Low Protein Foods**

Modified low protein foods recommended/prescribed     Unknown     Yes     No

Amount of protein grams recommended/prescribed from food per day (not including metabolic formula) for protein restricted diet \_\_\_\_\_

Method of payment for modified low protein foods

- 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/primary caregiver reports adherence to modified low protein foods     Unknown     Yes     No

Actual frequency of adherence to modified low protein foods

- Unknown     0 days/week     1 day/week     2 days/week     3 days/week     4 days/week
- 5 days/week     6 days/week

Reason for poor adherence to modified low protein foods \_\_\_\_\_

**Phenylalanine Restricted Diet**

Phenylalanine restricted diet recommended/prescribed     Unknown     Yes     No

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

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

Date

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**

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        ***BIOPT-REG - Study Status***First date of study status change      Condition follow-up status    Active    Inactive

Reason for inactive status

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

Date of death      

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

Moved-specify

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

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

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

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

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

Specify reason subject initiated withdrawal, if known

Specify reason investigator initiated withdrawal, if known

Second date of study status change

Condition follow-up status     Active     Inactive

Reason for inactive status

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

Date of death

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

Moved-specify

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

Specify other IBEMC participating center

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

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

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

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

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

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

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

Specify reason subject initiated withdrawal, if known

Specify reason investigator initiated withdrawal, if known

Third date of study status change

Condition follow-up status     Active     Inactive

Reason for inactive status

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

Date of death

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

Moved-specify

- Moved to another IBEMC participating center
- Moved to another non-IBEMC participating center
- Moved-condition follow-up status unknown
- Specify other IBEMC participating center
  - IL-Ann & Robert H. Lurie Children's Hospital of Chicago
  - IL-University of Illinois
  - IN- Riley Hospital for Children Indiana University Health
  - KY-University of Louisville
  - MI-University of Michigan
  - MI-Wayne State University Children's Hospital of Michigan
  - MN-University of Minnesota
  - MO-University of Missouri
  - NE-University of Nebraska
  - NJ-Hackensack University
  - NY-University of Rochester
  - NY-Women's & Children's Hospital of Buffalo
  - OH-Cincinnati Children's Hospital
  - OH-Nationwide Children's Hospital

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

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

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

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

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

Specify reason subject initiated withdrawal, if known

Specify reason investigator initiated withdrawal, if known

Fourth date of study status change   |   |

Condition follow-up status    Active    Inactive

Reason for inactive status

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

Date of death   |   |

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



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

Moved-specify

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

Specify other IBEMC participating center

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

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

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

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

Date

Declined consent at age of majority or emancipation

Subject initiated withdrawal

Investigator initiated withdrawal

Specify reason subject initiated withdrawal, if known

Specify reason investigator initiated withdrawal, if known

### Comments

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Study status comments

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

Date      **BIOPT-REG - 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 |||||

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

Abnormal patient lab results during first delivery-other, specify

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

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

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

Patient death during or shortly after first delivery     Unknown     Yes     No

Disorder contributed to death of mother during first delivery     Unknown     Yes     No

Weeks gestation at time of first delivery - round to nearest week\_\_\_\_\_

Method of first delivery

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

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

Live delivery of first newborn     Unknown     Yes     No

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

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

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

Additional interventions for first newborn during or shortly after delivery (other than labs)

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

Date

- Unknown
- Blood/blood product transfusion
- Glucose infusion
- Medications
- NICU/special care nursery monitoring
- Resuscitation
- TPN
- Other

Additional interventions for first newborn during or shortly after delivery-other, specify

Birth measurements for first newborn  Unknown  Head circumference  Length  Weight

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

Birth head circumference units  cm  in

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

Birth length units  cm  in

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

Birth weight units  lbs  kg  gm  oz

APGAR score at 1 minute for first newborn

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

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

APGAR score at 5 minutes for first newborn

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

Health concern(s) with APGAR score at five minutes < 8 for first newborn

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

- Unknown  <24 hours
- 24-28 hours  3-5 days
- 6-14 days  >14 days
- N/A - baby was not born in a hospital  baby is still in the hospital

Routine newborn screening done for first newborn

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

- 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

- Ammonia - WNL
  - Blood glucose - WNL
  - BNP - Abn high
  - CBC - Abn
  - C02 - Abn low
  - CK - Abn high
  - Glucose Tolerance Test (oral) - Abn
  - INR - Abn
  - Liver function tests - Abn high
  - Plasma acylcarnitine profile - Abn
  - Plasma amino acids - Abn
  - Plasma carnitine levels - Abn
  - Prealbumin - Abn low
  - Transferrin - Abn
  - Urine acylcarnitines - Abn
  - Urine acylglycines - Abn
  - Urine ketones - Abn high
  - Urine organic acids - Abn
  - Other
- Blood glucose - Abn low
  - Blood Glucose - Abn high
  - BNP - WNL
  - CBC - WNL
  - C02 - WNL
  - CK - WNL
  - Glucose Tolerance Test (oral) - WNL
  - INR - WNL
  - Liver function tests - WNL
  - Plasma acylcarnitine profile - WNL
  - Plasma amino acids - WNL
  - Plasma carnitine levels - WNL
  - Prealbumin - WNL
  - Transferrin - WNL
  - Urine acylcarnitines - WNL
  - Urine acyglycines - WNL
  - Urine ketones - WNL
  - Urine organic acids - WNL

Other laboratory studies done on patient during second pregnancy: describe test(s) and result(s)

Additional interventions required during second pregnancy due to this metabolic condition

- Unknown
- 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 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

- Unknown
- 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 |M|M| |D|D| |Y|Y|

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  NoNewborn's death related to maternal disorder for second newborn  Unknown  Yes  NoSecond newborn currently alive  Unknown  Yes  No

### Third Pregnancy

---

Patient's age at time of third pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before third pregnancy (in months)

- Unknown  Unplanned  1  2  3  4  5

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

- 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"/> CO2 - Abn low       | <input type="checkbox"/> CO2 - WNL                |



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

Date      

- |  |  |
|--|--|
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

Other laboratory studies done on patient during third pregnancy: describe test(s) and result(s)

Additional interventions required during third pregnancy due to this metabolic condition

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                                    | <input type="checkbox"/> ED visits for hyperemesis/IV fluids   |
| <input type="checkbox"/> Hospitalizations for hyperemesis/IV fluids | <input type="checkbox"/> TPN                                   |
| <input type="checkbox"/> Tube feedings                              | <input type="checkbox"/> Additional medications                |
| <input type="checkbox"/> Home lab monitoring                        | <input type="checkbox"/> Increased frequency of lab monitoring |
| <input type="checkbox"/> More intensive fetal monitoring            | <input type="checkbox"/> Bedrest                               |
| <input type="checkbox"/> Other                                      |  |

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

\_\_\_\_\_

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

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 |||||

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

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

Complications during fourth pregnancy

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

Complications related to disorder during fourth pregnancy- specify

- Unknown
- Acute fatty liver of pregnancy (AFLP)
- Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)
- Intrauterine growth restriction (AUGR)
- Mother affected with this condition
- Other

Complications not known to be related to disorder during fourth pregnancy-specify

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

Complications during fourth pregnancy-other, specify\_\_\_\_\_

Number of ED visits for complications during fourth pregnancy, unrelated to management of disorder

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

Number of hospitalizations for complications during fourth pregnancy, unrelated to management of disorder

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



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

Date    |    |   

Duration of longest inpatient hospitalization (in days) for any reason during fourth pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during fourth pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during fourth pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during fourth pregnancy

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> Ammonia - Abn high                  |
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> CO <sub>2</sub> - Abn low           | <input type="checkbox"/> CO <sub>2</sub> - WNL               |
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

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

Date |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 <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"/>

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

Abnormal patient lab results during fourth 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 fourth delivery-other, specify

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

Patient death during or shortly after fourth delivery     Unknown     Yes     No

Disorder contributed to death of mother during fourth delivery     Unknown     Yes     No

Weeks gestation at time of fourth delivery - round to nearest week\_\_\_\_\_

Method of fourth delivery

- |   |   |
|---|---|
| <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- fourth delivery\_\_\_\_\_

Live delivery of fourth newborn     Unknown     Yes     No

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

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

Date

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

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

Additional interventions for fourth newborn during or shortly after delivery (other than labs)

- Unknown  Blood/blood product transfusion
- Glucose infusion  Medications
- NICU/special care nursery monitoring  Resuscitation
- TPN  Other

Additional interventions for fourth newborn during or shortly after delivery-other, specify

Birth measurements for fourth newborn  Unknown  Head circumference  Length  Weight

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

Birth head circumference units  cm  in

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

Birth length units  cm  in

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

Birth weight units  lbs  kg  gm  oz

APGAR score at 1 minute for fourth newborn

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

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

APGAR score at 5 minutes for fourth newborn

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

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

Date      

Health concern(s) with APGAR score at five minutes &lt; 8 for fourth newborn

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

- Unknown  <24 hours  
 24-28 hours  3-5 days  
 6-14 days  >14 days  
 N/A - baby was not born in a hospital  baby is still in the hospital

Routine newborn screening done for fourth newborn

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

Describe abnormal newborn screen result for fourth newborn

Reason routine newborn screening was not done for fourth newborn

- Unknown  
 Died prior to collection of NBS  
 Refused  
 Transferred to another facility prior to NBS collection  
 Transfused prior to collection of NBS  
 Other

Reason routine newborn screening was not done for fourth newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the fourth baby after birth to rule out the mother's disorder

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

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

Date      

Additional studies and results (examples: brain MRI, echocardiogram) done on fourth newborn to assess for effects of maternal disorder

Fourth newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

Unknown  Yes  No

Abnormal newborn exam findings for fourth newborn

- |   |  |   |
|---|--|---|
| <input type="checkbox"/> Unknown                    | <input type="checkbox"/> None                      | <input type="checkbox"/> Congenital heart disease |
| <input type="checkbox"/> Dysmorphism                | <input type="checkbox"/> Lethargy                  | <input type="checkbox"/> Microcephaly             |
| <input type="checkbox"/> Other congenital anomalies | <input type="checkbox"/> Poor feeding              | <input type="checkbox"/> Respiratory distress     |
| <input type="checkbox"/> Seizure(s)                 | <input type="checkbox"/> Small for gestational age | <input type="checkbox"/> Other                    |

Abnormal newborn exam findings for fourth newborn-other, specify \_\_\_\_\_

Newborn death at or shortly after delivery for fourth newborn  Unknown  Yes  No

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

Fourth newborn currently alive  Unknown  Yes  No

## **Fifth Pregnancy**

---

Patient's age at time of fifth pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before fifth pregnancy (in months)

- Unknown  Unplanned  1  2  3  4  5  
 6  7  8  9  10  11  12  
 13-24  25-36  >36

History of infertility for fifth pregnancy

- Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for fifth pregnancy

- Unknown  Male factor  Female factor  
 Combined male-female factor

Cause(s) of male infertility for fifth pregnancy

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

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

Date      

- 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  NoPreimplantation genetic diagnosis for disorder done for fifth pregnancy  Unknown  Yes  NoPrenatal testing done for fetus for this disorder for fifth pregnancy  Unknown  Yes  NoType of prenatal testing performed for fifth pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for fifth pregnancy

- Unknown  Amniocentesis  Chorionic villus sampling

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

- Unknown  None  
 Fetal echocardiogram  Increased frequency prenatal ultrasounds

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

Other laboratory studies done on patient during fifth pregnancy: describe test(s) and result(s)

[Empty text box for laboratory studies]

Additional interventions required during fifth 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 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
- 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 fifth pregnancy-other, specify

[Empty text box for additional interventions]

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

[Empty text box for actual interventions]

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

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

[Empty text box for lab tests and results]

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

[Empty text box for health concerns]

Additional interventions for fifth newborn during or shortly after delivery (other than labs)

- Unknown
- Blood/blood product transfusion
- Glucose infusion
- Medications
- NICU/special care nursery monitoring
- Resuscitation
- TPN
- Other

Additional interventions for fifth newborn during or shortly after delivery-other, specify

[Empty text box for additional interventions]

Birth measurements for fifth newborn  Unknown  Head circumference  Length  Weight

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

Birth head circumference units  cm  in

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

Birth length units  cm  in

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

Birth weight units  lbs  kg  gm  oz

APGAR score at 1 minute for fifth newborn

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

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

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

APGAR score at 5 minutes for fifth newborn

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

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

Date      

Health concern(s) with APGAR score at five minutes &lt; 8 for fifth newborn

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

- Unknown  <24 hours  
 24-28 hours  3-5 days  
 6-14 days  >14 days  
 N/A - baby was not born in a hospital  baby is still in the hospital

Routine newborn screening done for fifth newborn

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

Describe abnormal newborn screen result for fifth newborn

Reason routine newborn screening was not done for fifth newborn

- Unknown  
 Died prior to collection of NBS  
 Refused  
 Transferred to another facility prior to NBS collection  
 Transfused prior to collection of NBS  
 Other

Reason routine newborn screening was not done for fifth newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the fifth baby after birth to rule out the mother's disorder

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

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

Date      

Additional studies and results (examples: brain MRI, echocardiogram) done on fifth newborn to assess for effects of maternal disorder

Fifth newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

Unknown  Yes  No

Abnormal newborn exam findings for fifth newborn

Unknown  None  Congenital heart disease  
 Dysmorphism  Lethargy  Microcephaly  
 Other congenital anomalies  Poor feeding  Respiratory distress  
 Seizure(s)  Small for gestational age  Other

Abnormal newborn exam findings for fifth newborn-other, specify \_\_\_\_\_

Newborn death at or shortly after delivery for fifth newborn  Unknown  Yes  No

Newborn's death related to maternal disorder for fifth newborn  Unknown  Yes  No

Fifth newborn currently alive  Unknown  Yes  No

## Sixth Pregnancy

Patient's age at time of sixth pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before sixth pregnancy (in months)

Unknown  Unplanned  1  2  3  4  5  
 6  7  8  9  10  11  12  
 13-24  25-36  >36

History of infertility for sixth pregnancy

Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for sixth pregnancy

Unknown  Male factor  Female factor  
 Combined male-female factor

Cause(s) of male infertility for sixth pregnancy

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

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

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

Cause(s) of female infertility for sixth 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 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

- Unknown
- None
- Assisted reproductive technology
- Fertility drugs
- Surgery

Type of assisted reproductive technologies used during sixth pregnancy

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

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

\_\_\_\_\_

Partner tested for patient's disorder during sixth pregnancy

- Unknown
- Yes-biochemical
- Yes-molecular
- No

Partner also affected by disorder during sixth pregnancy  Unknown  Yes  No

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

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

Type of prenatal testing performed for sixth pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for sixth pregnancy

- Unknown
- Amniocentesis
- Chorionic villus sampling

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

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



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 |||||

Duration of longest inpatient hospitalization (in days) for any reason during sixth pregnancy \_\_\_\_\_

Highest value of primary metabolite of concern during sixth pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during sixth pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during sixth pregnancy

- |  |  |
|--|--|
| <input type="checkbox"/> Unknown                             | <input type="checkbox"/> Ammonia - Abn high                  |
| <input type="checkbox"/> Ammonia - WNL                       | <input type="checkbox"/> Blood glucose - Abn low             |
| <input type="checkbox"/> Blood glucose - WNL                 | <input type="checkbox"/> Blood Glucose - Abn high            |
| <input type="checkbox"/> BNP - Abn high                      | <input type="checkbox"/> BNP - WNL                           |
| <input type="checkbox"/> CBC - Abn                           | <input type="checkbox"/> CBC - WNL                           |
| <input type="checkbox"/> CO <sub>2</sub> - Abn low           | <input type="checkbox"/> CO <sub>2</sub> - WNL               |
| <input type="checkbox"/> CK - Abn high                       | <input type="checkbox"/> CK - WNL                            |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn                           | <input type="checkbox"/> INR - WNL                           |
| <input type="checkbox"/> Liver function tests - Abn high     | <input type="checkbox"/> Liver function tests - WNL          |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn  | <input type="checkbox"/> Plasma acylcarnitine profile - WNL  |
| <input type="checkbox"/> Plasma amino acids - Abn            | <input type="checkbox"/> Plasma amino acids - WNL            |
| <input type="checkbox"/> Plasma carnitine levels - Abn       | <input type="checkbox"/> Plasma carnitine levels - WNL       |
| <input type="checkbox"/> Prealbumin - Abn low                | <input type="checkbox"/> Prealbumin - WNL                    |
| <input type="checkbox"/> Transferrin - Abn                   | <input type="checkbox"/> Transferrin - WNL                   |
| <input type="checkbox"/> Urine acylcarnitines - Abn          | <input type="checkbox"/> Urine acylcarnitines - WNL          |
| <input type="checkbox"/> Urine acylglycines - Abn            | <input type="checkbox"/> Urine acylglycines - WNL            |
| <input type="checkbox"/> Urine ketones - Abn high            | <input type="checkbox"/> Urine ketones - WNL                 |
| <input type="checkbox"/> Urine organic acids - Abn           | <input type="checkbox"/> Urine organic acids - WNL           |
| <input type="checkbox"/> Other                               |  |

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

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

Other laboratory studies done on patient during sixth pregnancy: describe test(s) and result(s)

[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      

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

Cause(s) of female infertility for seventh pregnancy

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                          | <input type="checkbox"/> Age                             |
| <input type="checkbox"/> Alcohol use                      | <input type="checkbox"/> Cancer/cancer treatment related |
| <input type="checkbox"/> Early menopause                  | <input type="checkbox"/> Fallopian tube damage/blockage  |
| <input type="checkbox"/> Hyperprolactinemia               | <input type="checkbox"/> Pelvic adhesions                |
| <input type="checkbox"/> Polycystic ovary syndrome (PCOS) | <input type="checkbox"/> Premature ovarian insufficiency |
| <input type="checkbox"/> Over-exercise                    | <input type="checkbox"/> Ovulation disorder              |
| <input type="checkbox"/> Thyroid problems                 | <input type="checkbox"/> Tobacco use                     |
| <input type="checkbox"/> Uterine fibroid                  | <input type="checkbox"/> Weight related                  |
| <input type="checkbox"/> Other                            |  |

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

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

\_\_\_\_\_

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

- |   |  |
|---|--|
| <input type="checkbox"/> Unknown                          | <input type="checkbox"/> None            |
| <input type="checkbox"/> Assisted reproductive technology | <input type="checkbox"/> Fertility drugs |
| <input type="checkbox"/> Surgery                          |  |

Type of assisted reproductive technologies used during seventh pregnancy

- |  |                                |                              |
|--|--------------------------------|------------------------------|
| <input type="checkbox"/> Assisted hatching         | <input type="checkbox"/> ICSI  | <input type="checkbox"/> IVF |
| <input type="checkbox"/> Surgical sperm aspiration | <input type="checkbox"/> Other |                              |

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

\_\_\_\_\_

Partner tested for patient's disorder during seventh pregnancy

- |                                  |  |  |                             |
|----------------------------------|--|--|-----------------------------|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Yes-biochemical | <input type="checkbox"/> Yes-molecular | <input type="checkbox"/> No |
|----------------------------------|--|--|-----------------------------|

Partner also affected by disorder during seventh pregnancy  Unknown  Yes  NoPreimplantation genetic diagnosis for disorder done for seventh pregnancy  Unknown  Yes  NoPrenatal testing done for fetus for this disorder for seventh pregnancy  Unknown  Yes  NoType of prenatal testing performed for seventh pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for seventh pregnancy

- |                                  |  |  |
|----------------------------------|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Amniocentesis | <input type="checkbox"/> Chorionic villus sampling |
|----------------------------------|--|--|

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

- |   |   |
|---|---|
| <input type="checkbox"/> Unknown              | <input type="checkbox"/> None                                     |
| <input type="checkbox"/> Fetal echocardiogram | <input type="checkbox"/> Increased frequency prenatal ultrasounds |
| <input type="checkbox"/> Other                |   |

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

Date      

Additional prenatal testing performed on fetus as a result of parent's disorder for seventh pregnancy-other, specify \_\_\_\_\_

Seventh pregnancy terminated  Unknown  Yes  No

Reason seventh pregnancy terminated

- Elective due to fetus affected with disorder  
 Elective due to other fetal well-being unrelated to disorder  
 Elective for other reason  
 Elective due to maternal well-being  
 Spontaneous

Gestational age (in weeks) at time of seventh pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for seventh pregnancy \_\_\_\_\_

Prenatal care received during seventh pregnancy  Unknown  Yes  No

Weeks gestation prenatal care started during seventh pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during seventh pregnancy

- Unknown  Metabolic diet  Medications  
 Biochemical lab monitoring  Avoidance of fasting  Other

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

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

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

Patient in good metabolic condition during second trimester of seventh pregnancy

- Unknown  Pregnancy not sustained to second trimester  
 Yes  No

Patient in good metabolic condition during third trimester of seventh pregnancy

- Unknown  Pregnancy not sustained to third trimester  
 Yes  No

Number of outpatient metabolic visits for patient during seventh pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during seventh pregnancy

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

Number of hospitalizations for management of disorder during seventh pregnancy

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

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

Complications during 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      

Health concern(s) with APGAR score at five minutes &lt; 8 for seventh newborn

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

- Unknown  <24 hours  
 24-28 hours  3-5 days  
 6-14 days  >14 days  
 N/A - baby was not born in a hospital  baby is still in the hospital

Routine newborn screening done for seventh newborn

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

Describe abnormal newborn screen result for seventh newborn

Reason routine newborn screening was not done for seventh newborn

- Unknown  
 Died prior to collection of NBS  
 Refused  
 Transferred to another facility prior to NBS collection  
 Transfused prior to collection of NBS  
 Other

Reason routine newborn screening was not done for seventh newborn-other, specify

\_\_\_\_\_

Additional testing (beyond newborn screening) done for the seventh baby after birth to rule out the mother's disorder

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



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

Date      

Additional studies and results (examples: brain MRI, echocardiogram) done on seventh newborn to assess for effects of maternal disorder

Seventh newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

Unknown  Yes  No

Abnormal newborn exam findings for seventh newborn

Unknown  None  Congenital heart disease  
 Dysmorphism  Lethargy  Microcephaly  
 Other congenital anomalies  Poor feeding  Respiratory distress  
 Seizure(s)  Small for gestational age  Other

Abnormal newborn exam findings for seventh newborn-other, specify \_\_\_\_\_

Newborn death at or shortly after delivery for seventh newborn  Unknown  Yes  No

Newborn's death related to maternal disorder for seventh newborn  Unknown  Yes  No

Seventh newborn currently alive  Unknown  Yes  No

## **Eighth Pregnancy**

---

Patient's age at time of eighth pregnancy (in years) \_\_\_\_\_

Length of time patient tried to become pregnant before eighth pregnancy (in months)

Unknown  Unplanned  1  2  3  4  5  
 6  7  8  9  10  11  12  
 13-24  25-36  >36

History of infertility for eighth pregnancy

Unknown  
 Yes- not known to be related to inborn error of metabolism  
 Yes-related to inborn error of metabolism  
 No

Cause(s) of infertility for eighth pregnancy

Unknown  Male factor  Female factor  
 Combined male-female factor

Cause(s) of male infertility for eighth pregnancy

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

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

- Tobacco use
- Other

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

Cause(s) of female infertility for eighth pregnancy

- Unknown
- Age
- Alcohol use
- Cancer/cancer treatment related
- Early menopause
- Fallopian tube damage/blockage
- Hyperprolactinemia
- Pelvic adhesions
- Polycystic ovary syndrome (PCOS)
- Premature ovarian insufficiency
- Over-exercise
- Ovulation disorder
- Thyroid problems
- Tobacco use
- Uterine fibroid
- Weight related
- Other

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

Cause(s) of infertility for eighth pregnancy, combined male-female factor- specify \_\_\_\_\_

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

- Unknown
- None
- Assisted reproductive technology
- Fertility drugs
- Surgery

Type of assisted reproductive technologies used during eighth pregnancy

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

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

Partner tested for patient's disorder during eighth pregnancy

- Unknown
- Yes-biochemical
- Yes-molecular
- No

Partner also affected by disorder during eighth pregnancy  Unknown  Yes  No

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

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

Type of prenatal testing performed for eighth pregnancy  Unknown  Biochemical  Molecular

Method of prenatal testing for eighth pregnancy

- Unknown
- Amniocentesis
- Chorionic villus sampling

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

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

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

Date

Other

Additional prenatal testing performed on fetus as a result of parent's disorder for eighth pregnancy-other, specify \_\_\_\_\_

Eighth pregnancy terminated  Unknown  Yes  No

Reason eighth pregnancy terminated

- Elective due to fetus affected with disorder
- Elective due to other fetal well-being unrelated to disorder
- Elective for other reason
- Elective due to maternal well-being
- Spontaneous

Gestational age (in weeks) at time of eighth pregnancy termination - elective or spontaneous (round to the nearest week) \_\_\_\_\_

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for eighth pregnancy \_\_\_\_\_

Prenatal care received during eighth pregnancy  Unknown  Yes  No

Weeks gestation prenatal care started during eighth pregnancy \_\_\_\_\_

Treatment prescribed for patient's disorder during eighth pregnancy

- Unknown  Metabolic diet  Medications
- Biochemical lab monitoring  Avoidance of fasting  Other

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

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

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

Patient in good metabolic condition during second trimester of eighth pregnancy

- Unknown  Pregnancy not sustained to second trimester
- Yes  No

Patient in good metabolic condition during third trimester of eighth pregnancy

- Unknown  Pregnancy not sustained to third trimester
- Yes  No

Number of outpatient metabolic visits for patient during eighth pregnancy \_\_\_\_\_

Number of ED visits for management of disorder during eighth pregnancy

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

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

Date      

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"/> 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 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 <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 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 <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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- 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 |M|M| |D|D| |Y|Y|

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"/> <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 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      ***BIOPT-REG - Dialysis***

Number of different episodes during which dialysis (any type) was used

 1     2     3     4     5     6     7     8     9     10     >10
**First Dialysis Treatment**

Type(s) of dialysis received during first episode

- Unknown                       APD/CCPD                       ECMO                       CAPD  
 CVVH                       CVVD                       CVVHDF                       Hemodialysis  
 Peritoneal dialysis

Reason for first episode of dialysis

- Unknown                       Hyperammonemia     MSUD                       Organ failure                       Sepsis  
 Other

Reason for first episode of dialysis-other, specify

Start date of first episode of dialysis treatment      

Duration (in days) of first episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during first episode of dialysis

- Unknown                       None                       Ammonia                       Blood urea nitrogen  
 Plasma alloisoleucine     Plasma isoleucine                       Plasma leucine                       Plasma valine  
 Serum creatinine                       Other

Metabolite(s) of concern during first episode of dialysis-other,specify \_\_\_\_\_

Peak value of ammonia during first episode of dialysis \_\_\_\_\_

Peak value of ammonia during first episode of dialysis units

 umol/L                       ug/dL                       ug/L                       ug/mL                       g/dL

Peak value of blood urea nitrogen during first episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during first episode of dialysis units     mg/dL     mmol/L

Peak value of plasma alloisoleucine during first episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during first episode of dialysis units

 umol/dL                       umol/L                       mg/dL

Peak value of plasma isoleucine during first episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during first episode of dialysis units     umol/dL     umol/L     mg/dL

Peak value of plasma leucine during first episode of dialysis \_\_\_\_\_



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

Date      Peak value of plasma leucine during first episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of plasma valine during first episode of dialysis \_\_\_\_\_

Peak value of plasma valine during first episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of serum creatinine during first episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during first episode of dialysis units  umol/L  mg/dL

Peak value of other metabolite during first episode of dialysis \_\_\_\_\_

Peak value of other metabolite during first episode of dialysis units \_\_\_\_\_

Reason first episode of dialysis treatment was stopped

 Unknown  Acute episode resolved  Treatment withdrawn  Death Other

Reason first episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

**Second Dialysis Treatment**

Type(s) of dialysis received during second episode

 Unknown APD/CCPD ECMO CAPD CVVH CVVD CVVHDF Hemodialysis Peritoneal dialysis

Reason for second episode of dialysis

 Unknown Hyperammonemia MSUD Organ failure Sepsis Other

Reason for second episode of dialysis-other, specify

Start date of second episode of dialysis treatment      

Duration (in days) of second episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during second episode of dialysis

 Unknown None Ammonia Blood urea nitrogen Plasma alloisoleucine 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 <input type="text" value="MM"/> <input type="text" value="DD"/> <input type="text" value="YY"/>

Start date of fourth episode of dialysis treatment

Duration (in days) of fourth episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during fourth episode of dialysis

- Unknown                       None                       Ammonia                       Blood urea nitrogen
- Plasma alloisoleucine    Plasma isoleucine         Plasma leucine               Plasma valine
- Serum creatinine         Other

Metabolite(s) of concern during fourth episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during fourth episode of dialysis \_\_\_\_\_

Peak value of ammonia during fourth episode of dialysis units

- umol/L                       ug/dL                       ug/L                       ug/mL                       g/dL

Peak value of blood urea nitrogen during fourth episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during fourth episode of dialysis units    mg/dL    mmol/L

Peak value of plasma alloisoleucine during fourth episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during fourth episode of dialysis units

- umol/dL                       umol/L                       mg/dL

Peak value of plasma isoleucine during fourth episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during fourth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of plasma leucine during fourth episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during fourth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of plasma valine during fourth episode of dialysis \_\_\_\_\_

Peak value of plasma valine during fourth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of serum creatinine during fourth episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during fourth episode of dialysis units    umol/L    mg/dL

Peak value of other metabolite during fourth episode of dialysis \_\_\_\_\_

Peak value of other metabolite during fourth episode of dialysis units \_\_\_\_\_

Reason fourth episode of dialysis treatment was stopped

- Unknown                       Acute episode resolved    Treatment withdrawn     Death
- Other

Reason fourth episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

**Fifth Dialysis Treatment**

---

Type(s) of dialysis received during fifth episode

- Unknown                       APD/CCPD                       ECMO                       CAPD
- CVVH                       CVVD                       CVVHDF                       Hemodialysis
- Peritoneal dialysis

Reason for fifth episode of dialysis

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

Date

- Unknown       Hyperammonemia     MSUD                       Organ failure             Sepsis
- Other

Reason for fifth episode of dialysis-other, specify

Start date of fifth episode of dialysis treatment

Duration (in days) of fifth episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during fifth episode of dialysis

- Unknown                       None                       Ammonia                       Blood urea nitrogen
- Plasma alloisoleucine     Plasma isoleucine         Plasma leucine               Plasma valine
- Serum creatinine         Other

Metabolite(s) of concern during fifth episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during fifth episode of dialysis \_\_\_\_\_

Peak value of ammonia during fifth episode of dialysis units

- umol/L                       ug/dL                       ug/L                       ug/mL                       g/dL

Peak value of blood urea nitrogen during fifth episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during fifth episode of dialysis units     mg/dL     mmol/L

Peak value of plasma alloisoleucine during fifth episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during fifth episode of dialysis units

- umol/dL                       umol/L                       mg/dL

Peak value of plasma isoleucine during fifth episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during fifth episode of dialysis units     umol/dL     umol/L     mg/dL

Peak value of plasma leucine during fifth episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during fifth episode of dialysis units     umol/dL     umol/L     mg/dL

Peak value of plasma valine during fifth episode of dialysis \_\_\_\_\_

Peak value of plasma valine during fifth episode of dialysis units     umol/dL     umol/L     mg/dL

Peak value of serum creatinine during fifth episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during fifth episode of dialysis units     umol/L     mg/dL

Peak value of other metabolite during fifth episode of dialysis \_\_\_\_\_

Peak value of other metabolite during fifth episode of dialysis units \_\_\_\_\_

Reason fifth episode of dialysis treatment was stopped

- Unknown                       Acute episode resolved     Treatment withdrawn     Death
- Other

Reason fifth episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

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

Date      **Sixth Dialysis Treatment**

Type(s) of dialysis received during sixth episode

- Unknown       APD/CCPD       ECMO       CAPD  
 CVVH       CVVD       CVVHDF       Hemodialysis  
 Peritoneal dialysis

Reason for sixth episode of dialysis

- Unknown       Hyperammonemia       MSUD       Organ failure       Sepsis  
 Other

Reason for sixth episode of dialysis-other, specify

Start date of sixth episode dialysis treatment      

Duration (in days) of sixth episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during sixth episode of dialysis

- Unknown       None       Ammonia       Blood urea nitrogen  
 Plasma alloisoleucine       Plasma isoleucine       Plasma leucine       Plasma valine  
 Serum creatinine       Other

Metabolite(s) of concern during sixth episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during sixth episode of dialysis \_\_\_\_\_

Peak value of ammonia during sixth episode of dialysis units

- umol/L       ug/dL       ug/L       ug/mL       g/dL

Peak value of blood urea nitrogen during sixth episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during sixth episode of dialysis units  mg/dL  mmol/L

Peak value of plasma alloisoleucine during sixth episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during sixth episode of dialysis units

- umol/dL       umol/L       mg/dL

Peak value of plasma isoleucine during sixth episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during sixth episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of plasma leucine during sixth episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during sixth episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of plasma valine during sixth episode of dialysis \_\_\_\_\_

Peak value of plasma valine during sixth episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of serum creatinine during sixth episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during sixth episode of dialysis units  umol/L  mg/dL

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

Date    |    |

Peak value of other metabolite during sixth episode of dialysis \_\_\_\_\_

Peak value of other metabolite during sixth episode of dialysis units \_\_\_\_\_

Reason sixth episode of dialysis treatment was stopped

- Unknown                       Acute episode resolved  Treatment withdrawn     Death
- Other

Reason sixth episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

### Seventh Dialysis Treatment

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Type(s) of dialysis received during seventh episode

- Unknown                       APD/CCPD                       ECMO                               CAPD
- CVVH                               CVVD                               CVVHDF                           Hemodialysis
- Peritoneal dialysis

Reason for seventh episode of dialysis

- Unknown                       Hyperammonemia     MSUD                               Organ failure                       Sepsis
- Other

Reason for seventh episode of dialysis -other, specify

Start date of seventh episode of dialysis treatment    |    |

Duration (in days) of seventh episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during seventh episode of dialysis

- Unknown                       None                               Ammonia                           Blood urea nitrogen
- Plasma alloisoleucine     Plasma isoleucine               Plasma leucine                   Plasma valine
- Serum creatinine               Other

Metabolite(s) of concern during seventh episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during seventh episode of dialysis \_\_\_\_\_

Peak value of ammonia during seventh episode of dialysis units

- umol/L                       ug/dL                               ug/L                               ug/mL                               g/dL

Peak value of blood urea nitrogen during seventh episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during seventh episode of dialysis units     mg/dL     mmol/L

Peak value of plasma alloisoleucine during seventh episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during seventh episode of dialysis units

- umol/dL                               umol/L                               mg/dL

Peak value of plasma isoleucine during seventh episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during seventh episode of dialysis units

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

Date

umol/dL                       umol/L                       mg/dL

Peak value of plasma leucine during seventh episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during seventh episode of dialysis units     umol/dL     umol/L     mg/dL

Peak value of plasma valine during seventh episode of dialysis \_\_\_\_\_

Peak value of plasma valine during seventh episode of dialysis units     umol/dL     umol/L     mg/dL

Peak value of serum creatinine during seventh episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during seventh episode of dialysis units     umol/L     mg/dL

Peak value of other metabolite during seventh episode of dialysis \_\_\_\_\_

Peak value of other metabolite during seventh episode of dialysis units \_\_\_\_\_

Reason seventh episode of dialysis treatment was stopped

- Unknown                       Acute episode resolved     Treatment withdrawn     Death
- Other

Reason seventh episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

### **Eighth Dialysis Treatment**

Type(s) of dialysis received during eighth episode

- Unknown                       APD/CCPD                       ECMO                       CAPD
- CVVH                       CVVD                       CVVHDF                       Hemodialysis
- Peritoneal dialysis

Reason for eighth episode of dialysis

- Unknown                       Hyperammonemia     MSUD                       Organ failure                       Sepsis
- Other

Reason for eighth episode of dialysis-other, specify

Start date of eighth episode of dialysis treatment

Duration (in days) of eighth episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during eighth episode of dialysis

- Unknown                       None                       Ammonia                       Blood urea nitrogen
- Plasma alloisoleucine     Plasma isoleucine                       Plasma leucine                       Plasma valine
- Serum creatinine                       Other

Metabolite(s) of concern during eighth episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during eighth episode of dialysis \_\_\_\_\_

Peak value of ammonia during eighth episode of dialysis units

umol/L                       ug/dL                       ug/L                       ug/mL                       g/dL



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

Date

Peak value of blood urea nitrogen during eighth episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during eighth episode of dialysis units  mg/dL  mmol/L

Peak value of plasma alloisoleucine during eighth episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during eighth episode of dialysis units

umol/dL

umol/L

mg/dL

Peak value of plasma isoleucine during eighth episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during eighth episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of plasma leucine during eighth episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during eighth episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of plasma valine during eighth episode of dialysis \_\_\_\_\_

Peak value of plasma valine during eighth episode of dialysis units  umol/dL  umol/L  mg/dL

Peak value of serum creatinine during eighth episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during eighth episode of dialysis units  umol/L  mg/dL

Peak value of other metabolite during eighth episode of dialysis \_\_\_\_\_

Peak value of other metabolite during eighth episode of dialysis units \_\_\_\_\_

Reason eighth episode of dialysis treatment was stopped

Unknown

Acute episode resolved  Treatment withdrawn  Death

Other

Reason eighth episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

### Ninth Dialysis Treatment

Type(s) of dialysis received during ninth episode

Unknown

APD/CCPD

ECMO

CAPD

CVVH

CVVD

CVVHDF

Hemodialysis

Peritoneal dialysis

Reason for ninth episode of dialysis

Unknown

Hyperammonemia  MSUD

Organ failure

Sepsis

Other

Reason for ninth episode of dialysis-other, specify

Start date of ninth episode of dialysis treatment

Duration (in days) of ninth episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during ninth episode of dialysis

Unknown

None

Ammonia

Blood urea nitrogen

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

Date       Plasma alloisoleucine    Plasma isoleucine    Plasma leucine    Plasma valine Serum creatinine    Other

Metabolite(s) of concern during ninth episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during ninth episode of dialysis \_\_\_\_\_

Peak value of ammonia during ninth episode of dialysis units

 umol/L    ug/dL    ug/L    ug/mL    g/dL

Peak value of blood urea nitrogen during ninth episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during ninth episode of dialysis units    mg/dL    mmol/L

Peak value of plasma alloisoleucine during ninth episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during ninth episode of dialysis units

 umol/dL    umol/L    mg/dL

Peak value of plasma isoleucine during ninth episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during ninth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of plasma leucine during ninth episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during ninth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of plasma valine during ninth episode of dialysis \_\_\_\_\_

Peak value of plasma valine during ninth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of serum creatinine during ninth episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during ninth episode of dialysis units    umol/L    mg/dL

Peak value of other metabolite during ninth episode of dialysis \_\_\_\_\_

Peak value of other metabolite during ninth episode of dialysis units \_\_\_\_\_

Reason ninth episode of dialysis treatment was stopped

 Unknown    Acute episode resolved    Treatment withdrawn    Death Other

Reason ninth episode of dialysis treatment was stopped-other, specify \_\_\_\_\_

**Tenth Dialysis Treatment**

Type(s) of dialysis received during tenth episode

 Unknown    APD/CCPD    ECMO    CAPD CVVH    CVVD    CVVHDF    Hemodialysis Peritoneal dialysis

Reason for tenth episode of dialysis

 Unknown    Hyperammonemia    MSUD    Organ failure    Sepsis Other

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

Date

Reason for tenth episode of dialysis-other, specify

Start date of tenth episode of dialysis treatment

Duration (in days) of tenth episode of dialysis treatment \_\_\_\_\_

Metabolite(s) of concern during tenth episode of dialysis

- Unknown                       None                       Ammonia                       Blood urea nitrogen
- Plasma alloisoleucine    Plasma isoleucine         Plasma leucine               Plasma valine
- Serum creatinine         Other

Metabolite(s) of concern during tenth episode of dialysis-other, specify \_\_\_\_\_

Peak value of ammonia during tenth episode of dialysis \_\_\_\_\_

Peak value of ammonia during tenth episode of dialysis units

- umol/L                       ug/dL                       ug/L                       ug/mL                       g/dL

Peak value of blood urea nitrogen during tenth episode of dialysis \_\_\_\_\_

Peak value of blood urea nitrogen during tenth episode of dialysis units    mg/dL    mmol/L

Peak value of plasma alloisoleucine during tenth episode of dialysis \_\_\_\_\_

Peak value of plasma alloisoleucine during tenth episode of dialysis units

- umol/dL                       umol/L                       mg/dL

Peak value of plasma isoleucine during tenth episode of dialysis \_\_\_\_\_

Peak value of plasma isoleucine during tenth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of plasma leucine during tenth episode of dialysis \_\_\_\_\_

Peak value of plasma leucine during tenth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of plasma valine during tenth episode of dialysis \_\_\_\_\_

Peak value of plasma valine during tenth episode of dialysis units    umol/dL    umol/L    mg/dL

Peak value of serum creatinine during tenth episode of dialysis \_\_\_\_\_

Peak value of serum creatinine during tenth episode of dialysis units    umol/L    mg/dL

Peak value of other metabolite during tenth episode of dialysis \_\_\_\_\_

Peak value of other metabolite during tenth episode of dialysis units \_\_\_\_\_

Reason tenth episode of dialysis treatment was stopped

- Unknown                       Acute episode resolved    Treatment withdrawn     Death
- Other

Reason tenth episode of dialysis treatment was stopped-other, specify \_\_\_\_\_



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

Date      ***BIOPT-REG - 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       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