H-PHE - 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 ○Other

Intake date [M|M] [D|D] [Y|Y]

Consent

Consent obtained ○Yes ○No ○IRB Exempt

Assent obtained ○Yes ○No

Type of assent ○Written ○Verbal

Permission to recontact ○Unknown ○Yes ○No

Protocol ID ○A

Demographics Information

Patient last name_____

Patient first name_____

Date of birth [M|M] [D|D] [Y|Y]

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 disorders

Specify amino acid disorder diagnosis for the patient □Benign Hyperphenylalaninemia (H-PHE)

Patient disorder identification method

□Unknown □Abnormal newborn screen □Abnormal labs

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☐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

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 ○No

Other research studies are clinical trials ○Unknown ○Yes ○No

Research study-other, specify_____

Clinicaltrials.gov identifier_____

Education

Maternal education

○Unknown

○8th grade/less

○9th-12th grade, no diploma

○High school graduate or GED completed

○Some college credit but no degree

○Associate degree (e.g., AA, AS)

○Bachelor's degree (e.g., BA, AB, BS)

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

○Doctorate (e.g., PhD, EdD) or Professional degree (e.g., MD, DDS, DVM, LLB, JD)

Paternal education

○Unknown

○8th grade/less

○9th-12th grade, no diploma

○High school graduate or GED completed

○Some college credit but no degree

○Associate degree (e.g., AA, AS)

○Bachelor's degree (e.g., BA, AB, BS)

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

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○Doctorate (e.g., PhD, EdD) or Professional degree (e.g., MD, DDS, DVM, LLB, JD)

Patient education

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

Special education services received prior to intake ○Unknown ○Yes ○No

Age patient qualified for special education services

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

Ancestral Origin, Race and Ethnicity

Ancestral Origin ☐Unknown ☐Africa ☐Asia ☐Europe ☐North America ☐South America ☐Oceania ☐Other

Ancestral Origin-Africa

☐Unknown ☐Egypt ☐Eritrea ☐Ethiopia ☐Liberia ☐Somalia ☐South Africa
☐Other

Ancestral Origin-Africa-Other, specify_____

Ancestral Origin-Asia

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

Ancestral Origin-Asia-Other, specify_____

Ancestral Origin-Europe

☐Unknown ☐Austria ☐Belgium ☐Bulgaria ☐Croatia
☐Czech Republic ☐Denmark ☐Finland ☐France ☐Germany
☐Greece ☐Hungary ☐Iceland ☐Ireland ☐Italy
☐Lithuania ☐Malta ☐Netherlands ☐Norway ☐Poland
☐Romania ☐Serbia ☐Slovakia ☐Slovenia ☐Spain

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☐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

Maternal age (in years) at patient's birth_____

Paternal age (in years) at patient's birth_____

Mother's marital status at patient's birth

○Unknown ○Married ○Widowed ○Divorced ○Separated

○Never married ○Living with partner

County mother resides in at patient's birth_____

State mother resides in at patient's birth

○Unknown	○Not Applicable	○AL	○AK	○AZ	○AR
○CA	○CO	○CT	○DE	○DC	○FL
○GA	○HI	○ID	○IL	○IN	○IA

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○KS	○KY	○LA	○ME	○MD	○MA
○MI	○MN	○MS	○MO	○MT	○NE
○NV	○NH	○NJ	○NM	○NY	○NC
○ND	○OH	○OK	○OR	○PA	○PR
○RI	○SC	○SD	○TN	○TX	○UT
○VT	○VA	○WA	○WV	○WI	○WY

Medical Coverage

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

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

Intake demographics comments

H-PHE - Intake Family History

Family History

Consanguinity ○Unknown ○Yes ○No

Patient 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 ○Half

Sibling 1: Half ○Maternal ○Paternal

Sibling 1: Gender ○Unknown ○Male ○Female

Sibling 1: Year of birth_____

Sibling 1: Affected with this condition ○Unknown ○Yes ○No

Sibling 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 screen

□Patient/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 ○Borderline

Sibling 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-mortem

Sibling 2: Sibling type ○Full ○Half

Sibling 2: Half ○Maternal ○Paternal

Sibling 2: Gender ○Unknown ○Male ○Female

Sibling 2: Year of birth_____

Sibling 2: Affected with this condition ○Unknown ○Yes ○No

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-mortem

Sibling 3: Sibling type ○Full ○Half

Sibling 3: Half ○Maternal ○Paternal

Sibling 3: Gender ○Unknown ○Male ○Female

Sibling 3: Year of birth_____

Sibling 3: Affected with this condition ○Unknown ○Yes ○No

Sibling 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

Sibling 3: Age of death (in years)_____

Sibling 3: Timing of diagnosis ○Unknown ○Pre-mortem ○Post-mortem

Sibling 4: Sibling type ○Full ○Half

Sibling 4: Half ○Maternal ○Paternal

Sibling 4: Gender ○Unknown ○Male ○Female

Sibling 4: Year of birth_____

Sibling 4: Affected with this condition ○Unknown ○Yes ○No

Sibling 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 ○Borderline

Sibling 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-mortem

Sibling 5: Sibling type ○Full ○Half

Sibling 5: Half ○Maternal ○Paternal

Sibling 5: Gender ○Unknown ○Male ○Female

Sibling 5: Year of birth_____

Sibling 5: Affected with this condition ○Unknown ○Yes ○No

Sibling 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

Sibling 5: Results of newborn screening for this condition

○Unavailable ○Presumptive positive ○Negative/Normal ○Borderline

Sibling 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-mortem

Sibling 6: Sibling type ○Full ○Half

Sibling 6: Half ○Maternal ○Paternal

Sibling 6: Gender ○Unknown ○Male ○Female

Sibling 6: Year of birth_____

Sibling 6: Affected with this condition ○Unknown ○Yes ○No

Sibling 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 ○Borderline

Sibling 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-mortem

Sibling 7: Sibling type ○Full ○Half

Sibling 7: Half ○Maternal ○Paternal

Sibling 7: Gender ○Unknown ○Male ○Female

Sibling 7: Year of birth_____

Sibling 7: Affected with this condition ○Unknown ○Yes ○No

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-mortem

Sibling 8: Sibling type ○Full ○Half

Sibling 8: Half ○Maternal ○Paternal

Sibling 8: Gender ○Unknown ○Male ○Female

Sibling 8: Year of birth_____

Sibling 8: Affected with this condition ○Unknown ○Yes ○No

Sibling 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

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 ○No

Biological 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 ○Borderline

Biological 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-mortem

Biological father: Affected with this condition ○Unknown ○Yes ○No

Biological 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 ○Borderline

Biological 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

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 ○No

Maternal 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-mortem

Maternal grandfather: Affected with this condition ○Unknown ○Yes ○No

Maternal 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-mortem

Paternal grandmother: Affected with this condition ○Unknown ○Yes ○No

Paternal grandmother: Enrolled in this study ○Unknown ○Yes ○No

Paternal grandmother: NBSTRN ID for this study_____

Paternal grandmother: Method of diagnosis

(Continued on next page.)

☐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_____

Relationship to affected family member 9_____

Relationship to affected family member 10_____

Comments

Intake family history comments

H-PHE - 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

Type of maternal treatment for affected fetus

Pregnancy complication/risk factor-other, specify_____

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

☐In vitro fertilization (IVF) ☐Preimplantation genetic diagnosis (PGD)
☐Intrauterine insemination (IUI) ☐Surrogate
☐Donor sperm ☐Donor egg
☐Donor embryo ☐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

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

(Continued on next page.)

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

Type of congenital anomalies-other, specify_____

Neonatal complications ○Unknown ○Yes ○No

Type of neonatal complications

☐Antibiotics ☐APGAR < 5
☐Apnea/Bradycardia spells ☐Hypoglycemia
☐Intubation/mechanical ventilation ☐Infection/sepsis
☐IV fluids ☐Jaundice
☐Premature (< 37 weeks gestation) ☐Respiratory distress
☐Transfused ☐Seizures
☐Other

Type of neonatal complications-other, specify_____

Type of neonatal nutrition

☐Unknown ☐TPN ☐Breast milk ☐Elemental formula
☐Human milk fortifier ☐Intralipid ☐Regular formula ☐Non-Lactose formula
☐Metabolic formula ☐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

Birth length_____

Birth length units ○cm ○in

Birth weight_____

Birth weight units ○lbs ○kg ○gm ○oz

Health History

Patient has had an outpatient speciality visit ○Unknown ○Yes ○No

Date of last outpatient specialty visit |M|M| |D|D| |Y|Y|

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 ○No

Most recent full scale IQ date |M|M| |D|D| |Y|Y|

Most recent full scale IQ test name_____

Most recent full scale IQ number_____

Days of age at initation of phenylalnine 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

(Continued on next page.)

☐Other

Provider of genentic counseling, other- specify_____

Comorbidities at time of intake

Prior testing

Echocardiogram prior to intake ○Unknown ○Yes ○No

Echocardiogram date |M|M| |D|D| |Y|Y|

Echocardiogram results ○Unknown ○Normal ○Abnormal

Echocardiogram comments

Electrocardiogram prior to intake ○Unknown ○Yes ○No

Electrocardiogram date |M|M| |D|D| |Y|Y|

Electrocardiogram results ○Unknown ○Normal ○Abnormal

Electrocardiogram comments

Electroencephalography prior to intake ○Unknown ○Yes ○No

Electroencephalography date |M|M| |D|D| |Y|Y|

Electroencephalography results ○Unknown ○Normal ○Abnormal

Electroencephalography comments

Neurological imaging prior to intake ○Unknown ○Yes ○No

Neurological imaging date |M|M| |D|D| |Y|Y|

Neurological imaging results ○Unknown ○Normal ○Abnormal

Neurological imaging comments

History of a seizure disorder ○Unknown ○Yes ○No

Patient has biospecimen stored ○Unknown ○Yes ○No

Type of patient biospecimen ☐Blood ☐Urine ☐Other tissue

Type of patient biospecimen-other tissue, specify_____

Specify biospecimen location_____

Age (in years) at time of first Dexa scan_____

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

Number of phenylalanine levels measured age 2 < 3 years_____

Average tyrosine level age 2 < 3 years_____

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

Number of tyrosine levels measured age 2 < 3 years_____

Average phenylalanine intake age 2 < 3 years_____

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

Average blood phenylalanine level age 3 < 4 years_____

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

Number of phenylalanine levels measured age 3 < 4 years_____

Average tyrosine level age 3 < 4 years_____

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

Number of tyrosine levels measured age 3 < 4 years_____

Average phenylalanine intake age 3 < 4 years_____

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

Average blood phenylalanine level age 4 < 5 years_____

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

Number of phenylalanine levels measured age 4 < 5 years_____

Average tyrosine level age 4 < 5 years_____

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

Number of tyrosine levels measured age 4 < 5 years_____

Average phenylalanine intake age 4 < 5 years_____

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

Average blood phenylalanine level age 5 < 6 years_____

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

Number of phenylalanine levels measured age 5 < 6 years_____

Average tyrosine level age 5 < 6 years_____

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

Number of tyrosine levels measured age 5 < 6 years_____

Average phenylalanine intake age 5 < 6 years_____

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

Average blood phenylalanine level age 6 < 7 years_____

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

Number of phenylalanine levels measured age 6 < 7 years_____

Average tyrosine level age 6 < 7 years_____

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

Number of tyrosine levels measured age 6 < 7 years_____

Average phenylalanine intake age 6 < 7 years_____

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

Average blood phenylalanine level age 7 < 8 years_____

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

Number of phenylalanine levels measured age 7 < 8 years_____

Average tyrosine level age 7 < 8 years_____

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

Number of tyrosine levels measured age 7 < 8 years_____

Average phenylalanine intake age 7 < 8 years_____

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

Average blood phenylalanine level age 8 < 9 years_____

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

Number of phenylalanine levels measured age 8 < 9 years_____

Average tyrosine level age 8 < 9 years_____

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

Number of tyrosine levels measured age 8 < 9 years_____

Average phenylalanine intake age 8 < 9 years_____

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

Average blood phenylalanine level age 9 < 10 years_____

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

Number of phenylalanine levels measured age 9 < 10 years_____

Average tyrosine level age 9 < 10 years_____

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

Number of tyrosine levels measured age 9 < 10 years_____

Average phenylalanine intake age 9 < 10 years_____

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

Average blood phenylalanine level age 10 < 12 years_____

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

Number of phenylalanine levels measured age 10 < 12 years_____

Average tyrosine level age 10 < 12 years_____

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

Number of tyrosine levels measured age 10 < 12 years_____

Average phenylalanine intake age 10 < 12 years_____

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

Average blood phenylalanine level age 12 < 14 years_____

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

Number of phenylalanine levels measured age 12 < 14 years_____

Average tyrosine level age 12 < 14 years_____

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

Number of tyrosine levels measured age 12 < 14 years_____

Average phenylalanine intake age 12 < 14 years_____

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

Average blood phenylalanine level age 14 < 16 years_____

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

Number of phenylalanine levels measured age 14 < 16 years_____

Average tyrosine level age 14 < 16 years_____

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

Number of tyrosine levels measured age 14 < 16 years_____

Average phenylalanine intake age 14 < 16 years_____

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

Average blood phenylalanine level age 16 < 18 years_____

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

Number of phenylalanine levels measured age 16 < 18 years_____

Average tyrosine level age 16 < 18 years_____

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

Number of tyrosine levels measured age 16 < 18 years_____

Average phenylalanine intake age 16 < 18 years_____

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

Average blood phenylalanine level age 18 < 20 years_____

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

Number of phenylalanine levels measured age 18 < 20 years_____

Average tyrosine level age 18 < 20 years_____

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

Number of tyrosine levels measured age 18 < 20 years_____

Average phenylalanine intake age 18 < 20 years_____

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

Average blood phenylalanine level age 20 < 25 years_____

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

Number of phenylalanine levels measured age 20 < 25 years_____

Average tyrosine level age 20 < 25 years_____

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

Number of tyrosine levels measured age 20 < 25 years_____

Average phenylalanine intake age 20 < 25 years_____

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

Average blood phenylalanine level age 25 < 30 years_____

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

Number of phenylalanine levels measured age 25 < 30 years_____

Average tyrosine level age 25 < 30 years_____

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

Number of tyrosine levels measured age 25 < 30 years_____

Average phenylalanine intake age 25 < 30 years_____

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

Average blood phenylalanine level age 30 < 35 years_____

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

Number of phenylalanine levels measured age 30 < 35 years_____

Average tyrosine level age 30 < 35 years_____

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

Number of tyrosine levels measured age 30 < 35 years_____

Average phenylalanine intake age 30 < 35 years_____

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

Average blood phenylalanine level age 35 < 40 years_____

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

Number of phenylalanine levels measured age 35 < 40 years_____

Average tyrosine level age 35 < 40 years_____

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

Number of tyrosine levels measured age 35 < 40 years_____

Average phenylalanine intake age 35 < 40 years_____

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

Average blood phenylalanine level age 40 < 50 years_____

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

Number of phenylalanine levels measured age 40 < 50 years_____

Average tyrosine level age 40 < 50 years_____

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

Number of tyrosine levels measured age 40 < 50 years_____

Average phenylalanine intake age 40 < 50 years_____

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

Average blood phenylalanine level age > 50 years_____

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

Number of phenylalanine levels measured age > 50 years_____

Average tyrosine level age > 50 years_____

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

Number of tyrosine levels measured age > 50 years_____

Average phenylalanine intake age > 50 years_____

Average phenylalanine intake units age > 50 years ○mg/day

Average blood phenylalanine level for current year of life through intake_____

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

○umol/dL ○mg/dL ○umol/L

Number of phenylalanine levels measured for current year of life through intake_____

Average tyrosine level for current year of life through intake_____

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

Number of tyrosine levels measured for current year of life through intake_____

Average phenylalanine intake for current year of life through intake_____

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

Emergency Management

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

Name of web-based emergency alert program_____

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

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

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

○Unknown ○Yes ○No

Comments

Intake past health history comments

H-PHE - Intake Newborn Screening

Newborn Screening

Newborn screening performed ○Unknown ○Yes ○No

Number of newborn screen results available ○Unknown ○0 ○1 ○2 ○3

Date first newborn screen collected |M|M| |D|D| |Y|Y|

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

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

Other result/report on second newborn screen units_____

Date third newborn screen collected M M D D Y Y

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 ○No

R Ear: Equipment Type ☐Unknown ☐DPOAE ☐TEOAE ☐ABR ☐AABR ☐ALGO ☐Other

Right ear: Screening test results ○Pass ○Refer ○Not Tested

L Ear: Equipment Type ☐Unknown ☐DPOAE ☐TEOAE ☐ABR ☐AABR ☐ALGO ☐Other

Left 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 M M D D Y Y

Right ear: Diagnosis: Hearing loss? ○Yes ○No

Right ear: Diagnosis: Degree of hearing loss

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

Right ear: Diagnosis: Type of hearing loss

☐Sensorineural ☐Conductive

☐Mixed ☐Auditory neuropathy/Auditory dys-synchrony

☐Other

Left ear: Diagnosis: Hearing loss? ○Yes ○No

Left ear: Diagnosis: Degree of hearing loss

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○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 |M|M| |D|D| |Y|Y|

H-PHE - Intake Initial Testing

Patient status at time of NBS reporting to specialty center

○Unknown ○Well ○Symptomatic ○Deceased

Patient symptoms at time of initial contact

☐Unknown ☐None

☐Alopecia ☐Apnea

☐Arrhythmia ☐Ataxia

☐Athetosis ☐Autistic-like features

☐Body odor ☐Brain abnormalities

☐Brain malformations ☐Candidiasis

☐Cardiomyopathy ☐Cataract(s)

☐Cerebral edema ☐Chorea

☐Cirrhosis ☐Clonus

☐Cognitive impairment ☐Coma

☐Confusion ☐Conjunctivitis

☐Contracture(s)-musculoskeletal ☐Corneal erosion

☐Dehydration ☐Dermatitis

☐Developmental delay(s) ☐Disorientation

☐Drooling/hypersalivation ☐Dysarthria

☐Dysmetria ☐Dysmorphism

☐Dysphagia ☐Dystonia

☐Eczema ☐Edema

☐Failure to thrive ☐Fatigue

☐Flapping tremor ☐Fluctuating level of alertness

☐Gait abnormality (other than ataxia) ☐Genital abnormalities

☐Headache ☐Hearing loss

☐Hepatic encephalopathy ☐Hepatomegaly

☐Hyperreflexia ☐Hypertension

☐Hypertonia ☐Hyporeflexia

☐Hypothermia ☐Hypotonia

☐Increased intracranial pressure ☐Infection/sepsis

☐Irritability ☐Jaundice

☐Keratosis ☐Learning disability

☐Lethargy ☐Liver failure-acute

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

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

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

Diagnostic Testing

Diagnostic labs performed

☐Unknown ☐None
☐Blood spot phenylalanine ☐Blood spot tyrosine
☐Plasma amino acid profile ☐Plasma phenylalanine
☐Plasma tyrosine ☐RBC dihydropteridine reductase (DHPR) activity
☐Urine organic acids ☐Urine pterins
☐Other

Blood spot phenylalanine ○Unknown ○Within normal limits ○Abnormal

Blood spot phenylalanine significance ○Diagnostic ○Non-diagnostic

Blood spot phenylalanine value_____

Blood spot phenylalanine units

○umol/L ○umol/dL ○umol/mL ○nmol/L ○nmol/dL ○nmol/mL ○mmol/L
○mmol/dL ○mmol/mL ○mg/dL ○g/dL ○ug/dL

Blood spot phenylalanine reference range_____

Blood spot tyrosine ○Unknown ○Within normal limits ○Abnormal

Blood spot tyrosine significance ○Diagnostic ○Non-diagnostic

Blood spot tyrosine value_____

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

Blood spot tyrosine reference range_____

Plasma amino acid profile ○Unknown ○Within normal limits ○Abnormal

Plasma amino acid profile significance ○Diagnostic ○Non-diagnostic

Plasma amino acid comments

Plasma phenylalanine ○Unknown ○Within normal limits ○Abnormal

Plasma phenylalanine significance ○Diagnostic ○Non-diagnostic

Plasma phenylalanine value_____

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

Plasma phenylalanine reference range_____

Plasma tyrosine ○Unknown ○Within normal limits ○Abnormal

Plasma tyrosine significance ○Diagnostic ○Non-diagnostic

Plasma tyrosine value_____

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

Plasma tyrosine reference range_____

RBC dihydropteridine reductase (DHPR) activity ○Unknown ○Within normal limits ○Abnormal

RBC 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 ○Abnormal

Urine organic acids significance ○Diagnostic ○Non-diagnostic

Urine organic acid comments

Urine pterins □Biopterin □Neopterin □Primapterin

Urine biopterin-number of samples ○1 ○2 ○3

Urine biopterin percentage_____

Urine biopterin value_____

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

Urine biopterin reference range_____

Urine biopterin percentage_____

Urine biopterin value_____

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

Urine biopterin reference range_____

Urine biopterin percentage_____

Urine biopterin value_____

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

Urine biopterin reference range_____

Urine neopterin-number of samples ○1 ○2 ○3

Urine neopterin percentage_____

Urine neopterin value_____

Urine neopterin units ○nmol/mol Cr ○nmol/L

Urine neopterin reference range_____

Urine neopterin percentage_____

Urine neopterin value_____

Urine neopterin units ○nmol/mol Cr ○nmol/L

Urine neopterin reference range_____

Urine neopterin percentage_____

Urine neopterin value_____

Urine neopterin units ○nmol/mol Cr ○nmol/L

Urine neopterin reference range_____

Urine primapterin-number of samples ○1 ○2 ○3

Urine primapterin percentage_____

Urine primapterin value_____

Urine primapterin units ○nmol/L

Urine primapterin reference range_____

Urine primapterin percentage_____

Urine primapterin value_____

Urine primapterin units ○nmol/L

Urine primapterin reference range_____

Urine primapterin percentage_____

Urine primapterin value_____

Urine primapterin units ○nmol/L

Urine primapterin reference range_____

Diagnostic lab-other, specify_____

Other diagnostic lab ○Unknown ○Within normal limits ○Abnormal

Other diagnostic lab significance ○Diagnostic ○Non-diagnostic

Other diagnostic lab comments

Other diagnostic lab value_____

Other diagnostic lab units_____

Other diagnostic lab reference range_____

Genetic Testing

Type of genetic/genomic testing

☐Unknown ☐Not done ☐Done, not available

☐Single gene ☐Mutation panel ☐Exome sequencing

☐Full genome sequencing ☐Copy number variant ☐Deletion/duplication testing

☐Other

Reason genotyping was done

☐Unknown ☐Diagnosis confirmation ☐Modifier genes ☐Pharmacogenetics

☐Other

Reason genotyping was done-other, specify_____

Reason genotyping was not done

☐Unknown ☐Clinician deemed unnecessary ☐Not covered by insurance

☐Parent refusal ☐Test not available ☐Other

Reason genotyping was not done-other, specify_____

Type of genetic/genomic testing-other, specify_____

Gene(s) associated with H-PHE ☐PAH ☐Other

Gene(s) associated with H-PHE-other, specify_____

PAH: Specify allele 1_____

PAH: Specify allele 2_____

Other: Specify allele 1_____

Other: Specify allele 2_____

Maternal genetic testing done ○Unknown ○Yes ○No ○Genotyping in progress

Mother: Allele 1_____

Mother: Allele 2_____

Paternal genetic testing done ○Unknown ○Yes ○No ○Genotyping in progress

Father: Allele 1_____

Father: Allele 2_____

Comments

Initial testing comments

H-PHE - Visit Demographics And History

Visit Date M M D D Y Y

Date of last outpatient speciality visit M M D D Y Y

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 ○No

Other 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

(Continued on next page.)

☐Medicare

☐Military

☐Newborn screening funds

☐Patient assistance program

☐State Children's Health Insurance Program (SCHIP)

☐State Children with Special Health Needs (CSHN) Program

☐Other

Medical coverage at visit-Patient assistance program, specify_____

Medical coverage at visit-Other, specify_____

Family History

Patient has new biological sibling since last visit ○Unknown ○Yes ○No

Complete sibling history questions on the Family History form.

Comments

Visit demographics and history comments

H-PHE - 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 |M|M| |D|D| |Y|Y|

Reason for sick visit 1 ○Unknown ○Condition related ○Condition unrelated

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

Reason for sick visit 2 ○Unknown ○Condition related ○Condition unrelated

Sick visit 2 was a condition exacerbation ○Unknown ○Yes ○No

Location for sick visit 2

○Unknown ○Emergency department ○Retail clinic

○Primary care ○Specialty center ○Urgent care

○Direct hospital admission ○Other

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

Name of hospital for sick visit 2_____

ICD-9 codes for sick visit 2 known ○Yes ○No

ICD-9 codes for sick visit 2_____

Diagnosis for sick visit 2_____

Number of inpatient days for sick visit 2_____

Number of ICU days for sick visit 2_____

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

Reason for sick visit 3 ○Unknown ○Condition related ○Condition unrelated

Sick visit 3 was a condition exacerbation ○Unknown ○Yes ○No

Location for sick visit 3

○Unknown ○Emergency department ○Retail clinic

○Primary care ○Specialty center ○Urgent care

○Direct hospital admission ○Other

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

Name of hospital for sick visit 3_____

ICD-9 codes for sick visit 3 known ○Yes ○No

ICD-9 codes for sick visit 3_____

Diagnosis for sick visit 3_____

Number of inpatient days for sick visit 3_____

Number of ICU days for sick visit 3_____

Date of sick visit 4 |M|M| |D|D| |Y|Y|

Reason for sick visit 4 ○Unknown ○Condition related ○Condition unrelated

Sick visit 4 was a condition exacerbation ○Unknown ○Yes ○No

Location for sick visit 4

○Unknown ○Emergency department ○Retail clinic

○Primary care ○Specialty center ○Urgent care

○Direct hospital admission ○Other

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

Name of hospital for sick visit 4_____

ICD-9 codes for sick visit 4 known ○Yes ○No

ICD-9 codes for sick visit 4_____

Diagnosis for sick visit 4_____

Number of inpatient days for sick visit 4_____

Number of ICU days for sick visit 4_____

Date of sick visit 5 |M|M| |D|D| |Y|Y|

Reason for sick visit 5 ○Unknown ○Condition related ○Condition unrelated

Sick visit 5 was a condition exacerbation ○Unknown ○Yes ○No

Location for sick visit 5

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

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

Name of hospital for sick visit 5_____

ICD-9 codes for sick visit 5 known ○Yes ○No

ICD-9 codes for sick visit 5_____

Diagnosis for sick visit 5_____

Number of inpatient days for sick visit 5_____

Number of ICU days for sick visit 5_____

Date of sick visit 6 |M|M| |D|D| |Y|Y|

Reason for sick visit 6 ○Unknown ○Condition related ○Condition unrelated

Sick visit 6 was a condition exacerbation ○Unknown ○Yes ○No

Location for sick visit 6

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

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

Name of hospital for sick visit 6_____

ICD-9 codes for sick visit 6 known ○Yes ○No

ICD-9 codes for sick visit 6_____

Diagnosis for sick visit 6_____

Number of inpatient days for sick visit 6_____

Number of ICU days for sick visit 6_____

Date of sick visit 7 |M|M| |D|D| |Y|Y|

Reason for sick visit 7 ○Unknown ○Condition related ○Condition unrelated

Sick visit 7 was a condition exacerbation ○Unknown ○Yes ○No

Location for sick visit 7

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

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

Name of hospital for sick visit 7_____

ICD-9 codes for sick visit 7 known ○Yes ○No

ICD-9 codes for sick visit 7_____

Diagnosis for sick visit 7_____

Number of inpatient days for sick visit 7_____

Number of ICU days for sick visit 7_____

Date of sick visit 8 |M|M| |D|D| |Y|Y|

Reason for sick visit 8 ○Unknown ○Condition related ○Condition unrelated

Sick visit 8 was a condition exacerbation ○Unknown ○Yes ○No

Location for sick visit 8

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

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

Name of hospital for sick visit 8_____

ICD-9 codes for sick visit 8 known ○Yes ○No

ICD-9 codes for sick visit 8_____

Diagnosis for sick visit 8_____

Number of inpatient days for sick visit 8_____

Number of ICU days for sick visit 8_____

Date of sick visit 9 |M|M| |D|D| |Y|Y|

Reason for sick visit 9 ○Unknown ○Condition related ○Condition unrelated

Sick visit 9 was a condition exacerbation ○Unknown ○Yes ○No

Location for sick visit 9

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

Patient was admitted to the hospital as a result of sick visit 9 ○Unknown ○Yes ○No

Name of hospital for sick visit 9_____

ICD-9 codes for sick visit 9 known ○Yes ○No

ICD-9 codes for sick visit 9_____

Diagnosis for sick visit 9_____

Number of inpatient days for sick visit 9_____

Number of ICU days for sick visit 9_____

Date of sick visit 10 |M|M| |D|D| |Y|Y|

Reason for sick visit 10 ○Unknown ○Condition related ○Condition unrelated

Sick visit 10 was a condition exacerbation ○Unknown ○Yes ○No

Location for sick visit 10

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

Patient was admitted to the hospital as a result of sick visit 10 ○Unknown ○Yes ○No

Name of hospital for sick visit 10_____

ICD-9 codes for sick visit 10 known ○Yes ○No

ICD-9 codes for sick visit 10_____

Diagnosis for sick visit 10_____

Number of inpatient days for sick visit 10_____

Number of ICU days for sick visit 10_____

Procedures

Anesthesia since last visit ○Unknown ○Yes ○No

Specific anesthesia precautions recommended due to metabolic condition ○Unknown ○Yes ○No

Surgical precautions taken

Anesthesia complications ○Unknown ○Yes ○No

Anesthesia complications-specify

Surgeries since last visit ○Unknown ○Yes ○No

Surgical procedure(s)_____

Surgery complications ○Unknown ○Yes ○No

Surgery complications-specify

Pregnancy

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

Please complete the pregnancy form.

Patient has become pregnant since the last visit ○Unknown ○Yes ○No

Please complete the pregnancy form.

Dialysis

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

Please complete the dialysis form.

Transplants

Transplant since last visit

○Unknown

○Yes-transplant received

○No-patient was evaluated for transplant but did not receive

○No

Please complete the transplant form.

Other Procedures

Major medical procedure since last visit ○Unknown ○Yes ○No

Description of major medical procedure_____

Comments

Visit health history comments

H-PHE - Visit Findings

Visit Measurements

Measurements taken at visit

☐Unknown ☐None ☐Blood pressure ☐Head circumference
☐Height/length ☐Weight

Blood pressure-systolic_____

Blood pressure-diastolic_____

Head circumference_____

Head circumference units ○cm ○in

Height/length_____

Height/length units ○cm ○in

How height/length measured ○Unknown ○Supine ○Standing

Weight_____

Weight units ○lbs ○kg ○gm ○oz

Body mass index_____

Visit findings ☐Unknown ☐None ☐Evidence of atopic dermatitis ☐Evidence of tremor ☐Other

Visit findings-other, specify_____

Comments

Visit findings comments

H-PHE - Visit Ancillary Care

Care Coordination

Missed subspecialty visits since last visit ○Unknown ○Yes ○No

Number of missed subspecialty visits_____

Other health services currently received ○Unknown ○Yes ○No

Specify other current health services

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

Specify other current health services-other, specify_____

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

Name of primary care provider_____

Preventive care status

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

Type of transplant service ☐Unknown ☐Evaluated for transplant ☐Received transplant

Transplant organ ☐Unknown ☐Heart ☐Kidney ☐Liver ☐Lung ☐Stem cell ☐Other

Transplant organ-other, specify_____

Community resources currently received ○Unknown ○Yes ○No

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

Developmental status ○Typical ○Atypical

Severity of atypical development ○Unknown ○Mild delay ○Moderate delay ○Severe delay

Developmental milestones that were achieved in a typical order and timeframe

☐Unknown ☐None ☐Cognitive ☐Fine motor ☐Gross motor

☐Social-emotional ☐Speech-language ☐Other

Developmental milestones that were achieved in typical order and timeframe-other, specify

_____

Referred for further developmental assessment

○Unknown ○Yes

○No ○Previously referred

○Family declined further assessments

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

☐Unknown ☐Developmental/behavioral pediatrician

☐Neuropsychologist ☐Psychiatric APRN/CNP/CNS

☐Psychiatrist ☐Psychologist

☐School psychologist ☐Other

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

_____

Neuropsychometric evaluation performed since last visit ○Unknown ○Yes ○No

Overall neuropyschometric impression ○Above average ○Average ○Below average ○In progress

Patient has mental health concerns ○Unknown ○Yes ○No

Referred for further mental health assessment

○Unknown ○Yes

○No ○Previously referred

○Family declined further assessments

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

☐Unknown ☐Developmental/behavioral pediatrician

☐Marriage and family therapist ☐Mental health counselor

☐Neuropsychologist ☐Pastoral counselor

☐Psychiatric APRN/CNP/CNS ☐Psychiatrist

☐Psychologist ☐School psychologist

☐Social worker ☐Other

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

_____

Behavioral concerns ○Unknown ○Yes ○No

Referred for further behavioral assessment ○Unknown ○Yes ○No

Referred for further behavioral assessment-Explain

○Previously referred ○Family declined further assessments

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

☐Unknown ☐Developmental/behavioral pediatrician

☐Marriage and family therapist ☐Mental health counselor

☐Neuropsychologist ☐Psychiatric APRN/CNP/CNS

☐Psychiatrist ☐Psychologist

☐School psychologist ☐Social worker

☐Other

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

_____

Education

Special education assessment recommended

○Unknown ○Yes

○No ○Special education services already received

Reason special education services received

☐Unknown ☐Cognitive disability ☐Developmental delay

☐Fine motor disability ☐Gross motor disability ☐Learning disability

☐Social-emotional disability ☐Speech/Language disability ☐Other health impairment (OHI)

☐Other

Reason special education services received-other, specify_____

Special education category

☐Unknown

☐Autism spectrum disorders

☐Blind-visually impaired

☐Deaf and hard of hearing

☐Deaf-Blind

☐Developmental cognitive disabilities: mild-moderate

☐Developmental cognitive disabilities: severe- profound

☐Developmental delay

☐Emotional/Behavioral disorders

☐Physically impaired

☐Severely multiply impaired

☐Specific learning disabilities

(Continued on next page.)

☐Speech or language impairments

☐Traumatic brain injury

☐Other health disabilities

Special education, other- specify_____

Comments

Visit ancillary care comments

H-PHE - Visit Lab Studies

Biochemical Labs

Biochemical labs associated with this visit

☐Unknown ☐None
☐Dihydropteridine reductase (DHPR) ☐Folic acid
☐Isoleucine ☐Leucine
☐Phenylalanine ☐Plasma amino acid profile
☐Plasma essential fatty acid profile ☐Plasma total homocysteine level
☐Selenium ☐Serum vitamin B12 level
☐Total 25-hydroxyvitamin D ☐Tyrosine
☐Urine methylmalonic acid (MMA) ☐Urine organic acids
☐Urine pterins ☐Valine
☐Other

Dihydropteridine reductase (DHPR)

○Within normal limits ○Abnormal ○In progress ○Results unavailable

Dihydropteridine reductase (DHPR) value_____

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

Dihydropteridine reductase (DHPR) reference range_____

Folic acid ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Folic acid value_____

Folic acid units ○nmol/L ○ng/mL

Folic acid reference range_____

Isoleucine ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Isoleucine value_____

Isoleucine units ○umol/dL ○umol/L ○mg/dL

Isoleucine reference range_____

Leucine ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Leucine value_____

Leucine units ○umol/dL ○umol/L ○mg/dL

Leucine reference range_____

Phenylalanine ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Phenylalanine value_____

Phenylalanine units ○umol/dL ○umol/L ○mg/dL

Phenylalanine reference range_____

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

Plasma amino acid comments

Plasma essential fatty acid profile

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

Alpha linolenic acid ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Alpha linolenic acid value_____

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

Alpha linoleic acid reference range_____

Docosahexaenoic acid (DHA) acid

○Within normal limits ○Abnormal ○In progress ○Results unavailable

Docosahexaenoic acid (DHA) acid value_____

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

Docosahexaenoic acid (DHA) acid reference range_____

EPA ○Within normal limits ○Abnormal ○In progress ○Results unavailable

EPA value_____

EPA units ○nmol/L ○ng/mL ○nmol/mL

EPA reference range_____

Linoleic acid ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Linoleic acid value_____

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

Linoleic acid reference range_____

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

Triene/tetraene ratio value_____

Triene/tetraene ratio reference range_____

Total fatty acids ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Total fatty acids comments_____

Other plasma essential fatty acid_____

Other plasma essential fatty acid result

○Within normal limits ○Abnormal ○In progress ○Results unavailable

Other plasma essential fatty acid value_____

Other plasma essential fatty acid units_____

Other plasma essential fatty acid reference range_____

Other plasma essential fatty acid comments

Plasma total homocysteine level

○Within normal limits ○Abnormal ○In progress ○Results unavailable

Plasma total homocysteine level value_____

Plasma total homocysteine level units ○umol/L ○mg/L

Plasma total homocysteine level reference range_____

Selenium ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Selenium value_____

Selenium units ○umol/L ○ug/L

Selenium reference range_____

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

Serum vitamin B12 level value_____

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

Serum vitamin B12 level reference range_____

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

Total 25-hydroxyvitamin D value_____

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

Total 25-hydroxyvitamin D reference range_____

Tyrosine ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Tyrosine value_____

Tyrosine units ○umol/dL ○umol/L ○mg/dL

Tyrosine reference range_____

Urine methylmalonic acid level

○Within normal limits ○Abnormal ○In progress ○Results unavailable

Urine methylmalonic acid level value_____

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

Urine methylmalonic acid level reference range_____

Urine organic acids ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Urine organic acid comments

Urine pterins ☐Biopterin ☐Neopterin ☐Primapterin

Urine biopterin-number of samples ○1 ○2 ○3

Urine biopterin percentage_____

Urine biopterin value_____

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

Urine biopterin percentage_____

Urine biopterin value_____

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

Urine biopterin percentage_____

Urine biopterin value_____

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

Urine neopterin-number of samples ○1 ○2 ○3

Urine neopterin percentage_____

Urine neopterin value_____

Urine neopterin units ○nmol/mol Cr ○nmol/L

Urine neopterin percentage_____

Urine neopterin value_____

Urine neopterin units ○nmol/mol Cr ○nmol/L

Urine neopterin percentage_____

Urine neopterin value_____

Urine neopterin units ○nmol/mol Cr ○nmol/L

Urine primapterin-number of samples ○1 ○2 ○3

Urine primapterin percentage_____

Urine primapterin value_____

Urine primapterin units ○nmol/L

Urine primapterin percentage_____

Urine primapterin value_____

Urine primapterin units ○nmol/L

Urine primapterin percentage_____

Urine primapterin value_____

Urine primapterin units ○nmol/L

Valine ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Valine value_____

Valine units ○umol/dL ○umol/L ○mg/dL

Valine reference range_____

Biochemical lab associated with this visit-other, specify_____

Other visit lab ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Other visit lab comments

Other visit lab value_____

Other visit lab units_____

Other visit lab reference range_____

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

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

Chemistry Labs

Chemistry labs associated with this visit

☐Unknown	☐None	☐Arterial blood gas
☐Anion gap	☐Ammonia	☐Calcium
☐Chloride	☐Carbon dioxide	☐Glucose
☐Potassium	☐Lactate dehydrogenase	☐Magnesium
☐Sodium	☐Phosphorous	☐Total cholesterol (fasting)
☐Total cholesterol (random)	☐Uric acid	☐Other

Arterial blood gas ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Arterial blood gas comments

Arterial blood gas value_____

Arterial blood gas units ○mmHg ○mEq/L

Arterial blood gas reference range_____

Anion gap ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Anion gap value_____

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

Anion gap reference range_____

Ammonia ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Ammonia value_____

Ammonia units ○umol/L ○ug/dL

Ammonia reference range_____

Calcium ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Calcium value_____

Calcium units ○mmol/L ○mg/dL

Calcium reference range_____

Chloride ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Chloride value_____

Chloride units ○mmol/L ○mg/L

Chloride reference range_____

Carbon dioxide ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Carbon dioxide value_____

Carbon dioxide units ○mEq/L ○mmol/L

Carbon dioxide reference range_____

Glucose ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Glucose value_____

Glucose units ○mmol/L ○mg/dL

Glucose reference range_____

Potassium ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Potassium value_____

Potassium units ○mEq/L ○mmol/L

Potassium reference range_____

Lactate dehydrogenase ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Lactate dehydrogenase value_____

Lactate dehydrogenase units ○U/L

Lactate dehydrogenase reference range_____

Magnesium ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Magnesium value_____

Magnesium units ○mmol/L ○mg/dL

Magnesium reference range_____

Sodium ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Sodium value_____

Sodium units ○mEq/L ○mmol/L

Sodium reference range_____

Phosphorous ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Phosphorous value_____

Phosphorous units ○mmol/L ○mg/dL

Phosphorous reference range_____

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

Total Cholesterol (fasting) value_____

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

Total Cholesterol (fasting) reference range_____

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

Total Cholesterol (random) value_____

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

Total Cholesterol (random) reference range_____

Uric acid ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Uric acid value_____

Uric acid units ○umol/L ○mg/dL

Uric acid reference range_____

Chemistry labs-other, specify_____

Other chemistry labs ○Within normal limits ○Abnormal ○In progress ○Results unavailable

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

□Unknown	□None	□Fibrinogen
□Hematocrit	□Hemoglobin	□International Normalized Ratio
□Partial Thromboplastin Time	□Prothrombin time test	□Peripheral blood smear
□Platelet count	□Red blood cell count	□White blood cell count
□Other		

Fibrinogen ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Fibrinogen value_____

Fibrinogen units ○umol/L ○mg/dL

Fibrinogen reference range_____

Hematocrit ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Hematocrit value_____

Hematocrit units ○% ○Proportion of total hemoglobin

Hematocrit reference range_____

Hemoglobin ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Hemoglobin value_____

Hemoglobin units ○g/dL ○g/L

Hemoglobin reference range_____

International Normalized Ratio (INR)

○Within normal limits ○Abnormal ○In progress ○Results unavailable

International Normalized Ratio (INR) value_____

International Normalized Ratio (INR) reference range_____

Partial Thromboplastin Time (PTT)

○Within normal limits ○Abnormal ○In progress ○Results unavailable

Partial Thromboplastin Time (PTT) value_____

Partial Thromboplastin Time (PTT) units ○seconds

Partial Thromboplastin Time (PTT) reference range_____

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

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

□Unknown	□None	□Albumin
□Alkaline phosphatase	□Alanine aminotransferase	□Aspartate aminotransferase
□Direct bilirubin	□Gamma-glutamyl transpeptide	□Globulin
□Prealbumin	□Total bilirubin	□Total protein
□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

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

☐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^2 ○mL/min/1.73m^3

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

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

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

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

Visit lab studies comments

H-PHE - Visit Studies Other

Home Monitoring

Home monitoring recommended ○Unknown ○Yes ○No

Home 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_____

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 ○Plasma

Frequency 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_____

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 ○Plasma

Frequency 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 dinitrophenylbydrazine home monitoring samples

○Unknown ○Precipitate present ○Precipitate absent

Ketones

Highest level of ketones ○Unknown ○None ○Trace ○Small ○Moderate ○Large

Frequency 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 ○Large

Frequency 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

Specify routine frequency_____

Number of other home monitoring samples reported since last outpatient metabolic visit_____

Laboratory Monitoring

Laboratory monitoring other than home monitoring done since last visit

☐Unknown ☐No ☐Phenylalanine ☐Tyrosine

Phenylalanine

Sample type of phenylalanine laboratory monitoring ○Unknown ○Filter paper blood ○Plasma

Frequency of phenylalanine laboratory monitoring

○Unknown ○Only when symptomatic ○Routinely

Specify routine frequency_____

Recommended therapeutic phenylalanine goal_____

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

Highest phenylalanine value on laboratory monitoring samples_____

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

Average phenylalanine on laboratory monitoring samples_____

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

Average percent above recommended phenylalanine therapeutic goal_____

Average percent below recommended phenylalanine therapeutic goal_____

Number of phenylalanine laboratory monitoring samples reported since last outpatient metabolic visit_____

Tyrosine

Sample type of tyrosine laboratory monitoring ○Unknown ○Filter paper blood ○Plasma

Frequency of tyrosine laboratory monitoring ○Unknown ○Only when symptomatic ○Routinely

Specify routine frequency_____

Recommended therapeutic tyrosine goal_____

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

Lowest tyrosine value on laboratory monitoring samples_____

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

Average tyrosine on laboratory monitoring samples_____

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

Average percent below recommended tyrosine therapeutic goal_____

Number of tyrosine laboratory monitoring samples reported since last outpatient metabolic visit_____

Other Studies

Physiological tests associated with this visit

☐Unknown ☐None ☐Electrocardiogram (ECG)

☐Electroencephalography (EEG) ☐Electromyography (EMG) ☐Holter monitoring

(Continued on next page.)

☐Cardiac stress test

Electrocardiogram

Electrocardiogram date M M D D Y Y

Electrocardiogram findings of note

Electroencephalography

Electroencephalography date M M D D Y Y

Electroencephalography findings of note

Electromyography

Electromyography date M M D D Y Y

Electromyography findings of note

Holter Monitoring

Holter monitoring date M M D D Y Y

Holter monitoring findings of note

Cardiac Stress Test

Cardiac stress test date M M D D Y Y

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
☐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

(Continued on next page.)

☐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 M M D D Y Y
Other imaging studies explanation

Dexa Scan

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

Number of Dexa scans performed ○1 ○2

First Dexa scan sites

☐Unknown ☐Heel ☐Hip ☐Pelvis
☐Spine ☐Wrist ☐Total body ☐Total body minus head
☐Other

First Dexa scan: Heel result ○Unknown ○Abnormal ○Normal
First Dexa scan: Heel Zscore_____
First Dexa scan: Hip result ○Unknown ○Abnormal ○Normal
First Dexa scan: Hip Zscore_____
First Dexa scan: Pelvis result ○Unknown ○Abnormal ○Normal

First Dexa scan: Pelvis Zscore_____

First Dexa scan: Spine result ○Unknown ○Abnormal ○Normal

First Dexa scan: Spine Zscore_____

First Dexa scan: Wrist result ○Unknown ○Abnormal ○Normal

First Dexa scan: Wrist Zscore_____

First Dexa scan: Total body result ○Unknown ○Abnormal ○Normal

First Dexa scan: Total body Zscore_____

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

First Dexa scan: Total body minus head Zscore_____

First Dexa scan site-other, specify_____

First Dexa scan: Other site result ○Unknown ○Abnormal ○Normal

First Dexa scan: Other site Zscore_____

Second Dexa scan sites

☐Unknown ☐Heel ☐Hip ☐Pelvis

☐Spine ☐Wrist ☐Total body ☐Total body minus head

☐Other

Second Dexa scan: Heel result ○Unknown ○Abnormal ○Normal

Second Dexa scan: Heel Zscore_____

Second Dexa scan: Hip result ○Unknown ○Abnormal ○Normal

Second Dexa scan: Hip Zscore_____

Second Dexa scan: Pelvis result ○Unknown ○Abnormal ○Normal

Second Dexa scan: Pelvis Zscore_____

Second Dexa scan: Spine result ○Unknown ○Abnormal ○Normal

Second Dexa scan: Spine Zscore_____

Second Dexa scan: Wrist result ○Unknown ○Abnormal ○Normal

Second Dexa scan: Wrist Zscore_____

Second Dexa scan: Total body result ○Unknown ○Abnormal ○Normal

Second Dexa scan: Total body Zscore_____

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

Second Dexa scan: Total body minus head Zscore_____

Second Dexa scan site-other, specify_____

Second Dexa scan: Other site result ○Unknown ○Abnormal ○Normal

Second Dexa scan: Other site Zscore_____

Comments

Visit studies-other comments

H-PHE - Visit Management And Treatment Pharmacotherapy

Pharmacotherapy

Medications

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

Other medications- specify_____

Homeopathic therapies ○Unknown ○Yes ○No

Specify homeopathic therapies

Disease Treatment

Treatment recommended/prescribed

☐Unknown ☐None
☐Large neutral amino acids (LNAAs) ☐Sapropterin dihydrochloride
☐Tyrosine ☐Other

Large Neutral Amino Acids

LNAA route recommended/prescribed ○Unknown ○Feeding tube ○IV ○Oral

LNAA dose recommended/prescribed_____

LNAA dose recommended/prescribed units ○g ○mg

LNAA frequency recommended/prescribed

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

(Continued on next page.)

○Other

LNAA frequency recommended/prescribed-other, specify_____

LNAA taken as recommended/prescribed ○Unknown ○Yes ○No

Actual LNAA dose reported_____

Actual LNAA dose reported units ○g ○mg

Actual LNAA frequency reported

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

○Other

Actual LNAA frequency reported-other, specify_____

Reason LNAA is not taken as recommended/prescribed_____

Method of payment for LNAA

☐Unknown

☐None

☐Commercial/private

☐Medicaid

☐Medicare

☐Military

☐Newborn screening funds

☐Patient assistance program

☐Self-pay

☐State Children's Health Insurance Program (SCHIP)

☐State Children with Special Health Needs (CSHN) Program

☐Other

Sapropterin dihydrochloride

Neuropsychological evaluation performed before prescribing sapropterin dihydrochloride

○Unknown ○Yes ○No

Neuropsychological evaluation performed while taking sapropterin dihydrochloride

○Unknown ○Yes ○No

Sapropterin dihydrochloride route recommended/prescribed

○Unknown ○Feeding tube ○Oral-dissolved ○Oral-tablet

Sapropterin dihydrochloride dose recommended/prescribed_____

Sapropterin dihydrochloride dose recommended/prescribed units ○g ○mg

Sapropterin 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 ○mg

Actual sapropterin dihydrochloride frequency reported ○Unknown ○Once/day ○Other

Actual sapropterin dihydrochloride frequency reported-other, specify_____

Reason sapropterin dihydrochloride is not taken as recommended/prescribed_____

Criterion used to determine sapropterin dihydrochloride responsiveness

☐Unknown

☐> 20% decrease in phenylalanine level post-medication

☐> 30% decrease in phenylalanine level post-medication

☐Improved academic performance

☐Improved attention

☐Improved behavior

☐Improved mood stability

☐Increased dietary phenylalanine tolerance

☐Other

Criterion used to determine sapropterin dihydrochloride responsiveness-Other, specify

_____

Method of payment for sapropterin dihydrochloride

☐Unknown

☐None

☐Commercial/private

☐Medicaid

☐Medicare

☐Military

☐Newborn screening funds

☐Patient assistance program

☐Self-pay

☐State Children's Health Insurance Program (SCHIP)

☐State Children with Special Health Needs (CSHN) Program

☐Other

Tyrosine

Tyrosine route recommended/prescribed ○Unknown ○Feeding tube ○Oral

Tyrosine dose recommended/prescribed_____

Tyrosine dose recommended/prescribed units ○g ○mg

Tyrosine frequency recommended/prescribed

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

Tyrosine frequency recommended/prescribed-other, specify_____

Tyrosine taken as recommended/prescribed ○Unknown ○Yes ○No

Actual tyrosine dose reported_____

Actual tyrosine dose reported units ○g ○mg

Actual tyrosine frequency reported

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

Actual tyrosine frequency reported-other, specify_____

Reason tyrosine is not taken as recommended/prescribed_____

Method of payment for tyrosine

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

Other Treatment

Oher treatment recommend/presribed_____

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 assistance program
- ☐Self-pay
- ☐State Children's Health Insurance Program (SCHIP)
- ☐State Children with Special Health Needs (CSHN) Program
- ☐Other

Comments

Medication and supplement comments

H-PHE - 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

☐Unknown	☐None	☐Baby formula (regular)
☐Baby formula (soy)	☐Elemental formula	☐Breast milk
☐Human milk fortifier	☐Almond milk	☐Rice milk
☐Skim milk	☐1% milk	☐2% milk
☐Soy milk	☐Special metabolic formula	☐Toddler formula (regular)
☐Toddler formula (soy)	☐Whole milk	☐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

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

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☐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_____

Phenylanine Restricted Diet

Phenylalanine restricted diet recommended/prescribed ○Unknown ○Yes ○No

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

_____

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

○mg

Average phenylalanine intake per day reported since last outpatient visit_____

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

Other

Average protein intake (grams per day) reported since last outpatient metabolic visit_____

Other amino acid supplements recommended/prescribed ○Unknown ○Yes ○No

Name(s) of other amino acid supplement(s) recommended/prescribed_____

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

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

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

_____

Amount of other amino acid supplement(s) recommended/prescribed_____

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

Other amino acid supplment(s) frequency recommended/prescribed_____

Comments

Nutrition comments

H-PHE - Study Status

First date of study status change |M|M| |D|D| |Y|Y|

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

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

Specify other non-IBEMC particpating 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 withdrawl, if known

Specify reason investigator initiated withdrawal, if known

Second date of study status change |M|M| |D|D| |Y|Y|

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

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

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○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 particpating 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 withdrawl, if known

Specify reason investigator initiated withdrawal, if known

Third date of study status change M M D D Y Y

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 M M D D Y Y

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

(Continued on next page.)

○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 particpating 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 withdrawl, if known

Specify reason investigator initiated withdrawal, if known

Fourth date of study status change |M|M| |D|D| |Y|Y|

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

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

Specify other non-IBEMC particpating center_____

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

(Continued on next page.)

☐Declined consent at age of majority or emancipation

☐Subject initiated withdrawal

☐Investigator initiated withdrawal

Specify reason subject initiated withdrawl, if known

Specify reason investigator initiated withdrawal, if known

Comments

Study status comments

H-PHE - 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

(Continued on next page.)

☐Hyperprolactinemia ☐Pelvic adhesions
☐Polycystic ovary syndrome (PCOS) ☐Premature ovarian insuffiency
☐Over-exercise ☐Ovulation disorder
☐Thyroid problems ☐Tobacco use
☐Uterine fibroid ☐Weight related
☐Other

Cause(s) of female infertility for first pregnancy-other, specify_____

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

_____

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

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

Type of assisted reproductive technologies used during first pregnancy

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

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

_____

Partner tested for patient's disorder during first pregnancy

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

Partner also affected by disorder during first pregnancy ○Unknown ○Yes ○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

(Continued on next page.)

○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

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)

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

- ☐Unknown
- ☐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
- ☐Ammonia - Abn high
- ☐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

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

- ☐Unknown
- ☐Hospitalizations for hyperemesis/IV fluids
- ☐Tube feedings
- ☐Home lab monitoring
- ☐ED visits for hyperemesis/IV fluids
- ☐TPN
- ☐Additional medications
- ☐Increased frequency of lab monitoring

(Continued on next page.)

☐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

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)

(Continued on next page.)

☐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

(Continued on next page.)

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

Describe abnormal newborn screen result for first newborn

Reason routine newborn screening was not done for first newborn

☐Unknown
☐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

_____

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

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

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

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

○Unknown ○Yes ○No

Abnormal newborn exam findings for first newborn

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

Abnormal newborn exam findings for first newborn-other, specify_____

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

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

First 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 insuffiency

☐Over-exercise ☐Ovulation disorder

☐Thyroid problems ☐Tobacco use

☐Uterine fibroid ☐Weight related

☐Other

Cause(s) of female infertility for second pregnancy-other, specify_____

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

_____

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

☐Unknown ☐None

☐Assisted reproductive technology ☐Fertility drugs

☐Surgery

Type of assisted reproductive technologies used during second pregnancy

☐Assisted hatching ☐ICSI ☐IVF

☐Surgical sperm aspiration ☐Other

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

_____

Partner tested for patient's disorder during second pregnancy

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

Partner also affected by disorder during second pregnancy ○Unknown ○Yes ○No

Preimplantation genetic diagnosis for disorder done for second pregnancy ○Unknown ○Yes ○No

Prenatal testing done for fetus for this disorder for second pregnancy ○Unknown ○Yes ○No

Type of prenatal testing performed for second pregnancy ☐Unknown ☐Biochemical ☐Molecular

Method of prenatal testing for second pregnancy

☐Unknown ☐Amniocentesis ☐Chorionic villus sampling

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

☐Unknown ☐None

☐Fetal echocardiogram ☐Increased frequency prenatal ultrasounds

☐Other

Additional prenatal testing performed on fetus as a result of parent's disorder for second pregnancy-other, specify_____

Second pregnancy terminated ○Unknown ○Yes ○No

Reason second pregnancy terminated

○Elective due to fetus affected with disorder

○Elective due to other fetal well-being unrelated to disorder

○Elective for other reason

○Elective due to maternal well-being

○Spontaneous

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

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for second pregnancy_____

Prenatal care received during second pregnancy ○Unknown ○Yes ○No

Weeks gestation prenatal care started during second pregnancy_____

Treatment prescribed for patient's disorder during second pregnancy

☐Unknown ☐Metabolic diet ☐Medications

☐Biochemical lab monitoring ☐Avoidance of fasting ☐Other

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

Patient in good metabolic condition prior to second pregnancy ○Unknown ○Yes ○No

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

Patient in good metabolic condition during second trimester of second pregnancy

○Unknown ○Pregnancy not sustained to second trimester

○Yes ○No

Patient in good metabolic condition during third trimester of second pregnancy

○Unknown ○Pregnancy not sustained to third trimester

○Yes ○No

Number of outpatient metabolic visits for patient during second pregnancy_____

Number of ED visits for management of disorder during second pregnancy

○Unknown ○0 ○1 ○2 ○3 ○4 ○5

○6 ○7 ○8 ○9 ○10 ○>10

Number of hospitalizations for management of disorder during second pregnancy

○Unknown ○0 ○1 ○2 ○3 ○4 ○5

○6 ○7 ○8 ○9 ○10 ○>10

Complications during second pregnancy

☐Unknown ☐Yes-related to disorder

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

Complications related to disorder during second pregnancy- specify

☐Unknown

☐Acute fatty liver of pregnancy (AFLP)

☐Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)

☐Intrauterine growth restriction (AUGR)

☐Mother affected with this condition

☐Other

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

(Continued on next page.)

☐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

(Continued on next page.)

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

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

Additional interventions required during second pregnancy due to this metabolic condition

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

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

_____

Total maternal weight gain (in kg) during second pregnancy_____

Additional interventions planned for labor/delivery related to patient's disorder for second pregnancy

☐Unknown
☐None

(Continued on next page.)

☐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

(Continued on next page.)

☐Other

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

Patient death during or shortly after second delivery ○Unknown ○Yes ○No

Disorder contributed to death of mother during second delivery ○Unknown ○Yes ○No

Weeks gestation at time of second delivery - round to nearest week_____

Method of second delivery

○Unknown ○Caesarean section (scheduled or non-urgent)

○Casesarean section (emergent) ○Vaginal

Maternal inpatient days post- second delivery_____

Live delivery of second newborn ○Unknown ○Yes ○No

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

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

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

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

☐Unknown ☐Blood/blood product transfusion

☐Glucose infusion ☐Medications

☐NICU/special care nursery monitoring ☐Resuscitation

☐TPN ☐Other

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

Birth measurements for second newborn ☐Unknown ☐Head circumference ☐Length ☐Weight

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

(Continued on next page.)

☐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
_____

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

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

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

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

○Unknown ○Yes ○No

Abnormal newborn exam findings for second newborn

☐Unknown ☐None ☐Congenital heart disease
☐Dysmorphism ☐Lethargy ☐Microcephaly
☐Other congenital anomalies ☐Poor feeding ☐Respiratory distress
☐Seizure(s) ☐Small for gestational age ☐Other
Abnormal newborn exam findings for second newborn-other, specify_____

Newborn death at or shortly after delivery for second newborn ○Unknown ○Yes ○No
Newborn's death related to maternal disorder for second newborn ○Unknown ○Yes ○No
Second newborn currently alive ○Unknown ○Yes ○No

Third Pregnancy

Patient's age at time of third pregnancy (in years)_____
Length of time patient tried to become pregnant before third pregnancy (in months)
○Unknown ○Unplanned ○1 ○2 ○3 ○4 ○5

(Continued on next page.)

○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 insuffiency

☐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

(Continued on next page.)

☐Surgical sperm aspiration ☐Other

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

_____

Partner tested for patient's disorder during third pregnancy

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

Partner also affected by disorder during third pregnancy ○Unknown ○Yes ○No

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

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

Type of prenatal testing performed for third pregnancy ☐Unknown ☐Biochemical ☐Molecular

Method of prenatal testing for third pregnancy

☐Unknown ☐Amniocentesis ☐Chorionic villus sampling

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

☐Unknown ☐None

☐Fetal echocardiogram ☐Increased frequency prenatal ultrasounds

☐Other

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

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

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

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

Patient in good metabolic condition during second trimester of third pregnancy

○Unknown ○Pregnancy not sustained to second trimester

○Yes ○No

Patient in good metabolic condition during third trimester of third pregnancy

○Unknown ○Pregnancy not sustained to third trimester

○Yes ○No

Number of outpatient metabolic visits for patient during third pregnancy_____

Number of ED visits for management of disorder during third pregnancy

○Unknown ○0 ○1 ○2 ○3 ○4 ○5

○6 ○7 ○8 ○9 ○10 ○>10

Number of hospitalizations for management of disorder during third pregnancy

○Unknown ○0 ○1 ○2 ○3 ○4 ○5

○6 ○7 ○8 ○9 ○10 ○>10

Complications during third pregnancy

☐Unknown ☐Yes-related to disorder

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

Complications related to disorder during third pregnancy- specify

☐Unknown

☐Acute fatty liver of pregnancy (AFLP)

☐Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)

☐Intrauterine growth restriction (AUGR)

☐Mother affected with this condition

☐Other

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

☐Unknown

☐Advanced maternal age (35+ years of age)

☐Ectopic pregnancy

☐Gestational diabetes

☐Group B strep

(Continued on next page.)

☐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

☐Unknown
☐Ammonia - WNL
☐Blood glucose - WNL
☐BNP - Abn high
☐CBC - Abn
☐C02 - Abn low
☐Ammonia - Abn high
☐Blood glucose - Abn low
☐Blood Glucose - Abn high
☐BNP - WNL
☐CBC - WNL
☐C02 - WNL

(Continued on next page.)

☐CK - Abn high
☐CK - WNL
☐Glucose Tolerance Test (oral) - Abn
☐Glucose Tolerance Test (oral) - WNL
☐INR - Abn
☐INR - WNL
☐Liver function tests - Abn high
☐Liver function tests - WNL
☐Plasma acylcarnitine profile - Abn
☐Plasma acylcarnitine profile - WNL
☐Plasma amino acids - Abn
☐Plasma amino acids - WNL
☐Plasma carnitine levels - Abn
☐Plasma carnitine levels - WNL
☐Prealbumin - Abn low
☐Prealbumin - WNL
☐Transferrin - Abn
☐Transferrin - WNL
☐Urine acylcarnitines - Abn
☐Urine acylcarnitines - WNL
☐Urine acylglycines - Abn
☐Urine acyglycines - WNL
☐Urine ketones - Abn high
☐Urine ketones - WNL
☐Urine organic acids - Abn
☐Urine organic acids - WNL
☐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

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

Additional interventions required during 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

☐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 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

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

Patient death during or shortly after third delivery ○Unknown ○Yes ○No

Disorder contributed to death of mother during third delivery ○Unknown ○Yes ○No

Weeks gestation at time of third delivery - round to nearest week_____

Method of third delivery

○Unknown ○Caesarean section (scheduled or non-urgent)

○Casesarean section (emergent) ○Vaginal

Maternal inpatient days post- third delivery_____

Live delivery of third newborn ○Unknown ○Yes ○No

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

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

Birth measurements for third newborn ☐Unknown ☐Head circumference ☐Length ☐Weight

Birth head circumference_____

Birth head circumference units ○cm ○in

Birth length_____

Birth length units ○cm ○in

Birth weight_____

Birth weight units ○lbs ○kg ○gm ○oz

APGAR score at 1 minute for third newborn

○Unknown ○0 ○1 ○2 ○3 ○4 ○5

○6 ○7 ○8 ○9 ○10

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

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 < 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

_____

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

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

Additional studies and results (examples: brain MRI, echocardiogram) done on 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

Cause(s) of male infertility for fourth pregnancy-other, specify_____

Cause(s) of female infertility for fourth pregnancy

☐Unknown ☐Age
☐Alcohol use ☐Cancer/cancer treatment related
☐Early menopause ☐Fallopian tube damage/blockage
☐Hyperprolactinemia ☐Pelvic adhesions
☐Polycystic ovary syndrome (PCOS) ☐Premature ovarian insuffiency
☐Over-exercise ☐Ovulation disorder
☐Thyroid problems ☐Tobacco use
☐Uterine fibroid ☐Weight related
☐Other

Cause(s) of female infertility for fourth pregnancy-other, specify_____

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

_____

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

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

Type of assisted reproductive technologies used during fourth pregnancy

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

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

_____

Partner tested for patient's disorder during fourth pregnancy

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

Partner also affected by disorder during fourth pregnancy ○Unknown ○Yes ○No

Preimplantation genetic diagnosis for disorder done for fourth pregnancy ○Unknown ○Yes ○No

Prenatal testing done for fetus for this disorder for fourth pregnancy ○Unknown ○Yes ○No

Type of prenatal testing performed for fourth pregnancy ☐Unknown ☐Biochemical ☐Molecular

Method of prenatal testing for fourth pregnancy

☐Unknown ☐Amniocentesis ☐Chorionic villus sampling

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

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

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

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○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

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

- ☐Unknown
- ☐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
- ☐Ammonia - Abn high
- ☐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 fourth pregnancy: describe test(s) and result(s)

Additional interventions required during fourth pregnancy due to this metabolic condition

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

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

_____

Total maternal weight gain (in kg) during fourth pregnancy_____

Additional interventions planned for labor/delivery related to patient's disorder for fourth pregnancy

- ☐Unknown
- ☐None
- ☐Additional maternal lab monitoring
- ☐Altered anesthesia plan
- ☐Change in delivery site
- ☐IV fluids
- ☐Letter to OB/MFM specialist
- ☐Planned C-section
- ☐Referral for high risk OB management
- ☐Other

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

Actual interventions for labor/delivery related to patient's disorder for fourth pregnancy

- ☐Unknown
- ☐No
- ☐Letter to OB/MFM specialist
- ☐IV fluids
- ☐Planned C-section
- ☐Additional maternal lab monitoring
- ☐Change in delivery site
- ☐Referral for high risk OB management
- ☐Altered anesthesia plan
- ☐Other

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

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

Abnormal patient lab results during fourth delivery

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

Abnormal patient lab results during fourth delivery-other, specify

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

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

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

Patient death during or shortly after fourth delivery ○Unknown ○Yes ○No

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

Weeks gestation at time of fourth delivery - round to nearest week_____

Method of fourth delivery

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

Maternal inpatient days post- fourth delivery_____

Live delivery of fourth newborn ○Unknown ○Yes ○No

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

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

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

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

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

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

Birth measurements for fourth newborn ☐Unknown ☐Head circumference ☐Length ☐Weight

Birth head circumference_____

Birth head circumference units ○cm ○in

Birth length_____

Birth length units ○cm ○in

Birth weight_____

Birth weight units ○lbs ○kg ○gm ○oz

APGAR score at 1 minute for fourth newborn

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

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

APGAR score at 5 minutes for fourth newborn

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

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

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

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

Routine newborn screening done for fourth newborn

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

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

_____

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

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

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

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

○Unknown ○Yes ○No

Abnormal newborn exam findings for fourth newborn

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

Abnormal newborn exam findings for fourth newborn-other, specify_____

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

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

Fourth newborn currently alive ○Unknown ○Yes ○No

Fifth Pregnancy

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

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

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

History of infertility for fifth pregnancy

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

Cause(s) of infertility for fifth pregnancy

☐Unknown ☐Male factor ☐Female factor
☐Combined male-female factor

Cause(s) of male infertility for fifth pregnancy

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

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☐Tobacco use ☐Other

Cause(s) of male infertility for fifth pregnancy-other, specify_____

Cause(s) of female infertility for fifth pregnancy

☐Unknown ☐Age
☐Alcohol use ☐Cancer/cancer treatment related
☐Early menopause ☐Fallopian tube damage/blockage
☐Hyperprolactinemia ☐Pelvic adhesions
☐Polycystic ovary syndrome (PCOS) ☐Premature ovarian insuffiency
☐Over-exercise ☐Ovulation disorder
☐Thyroid problems ☐Tobacco use
☐Uterine fibroid ☐Weight related
☐Other

Cause(s) of female infertility for fifth pregnancy-other, specify_____

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

_____

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

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

Type of assisted reproductive technologies used during fifth pregnancy

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

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

_____

Partner tested for patient's disorder during fifth pregnancy

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

Partner also affected by disorder during fifth pregnancy ○Unknown ○Yes ○No

Preimplantation genetic diagnosis for disorder done for fifth pregnancy ○Unknown ○Yes ○No

Prenatal testing done for fetus for this disorder for fifth pregnancy ○Unknown ○Yes ○No

Type of prenatal testing performed for fifth pregnancy ☐Unknown ☐Biochemical ☐Molecular

Method of prenatal testing for fifth pregnancy

☐Unknown ☐Amniocentesis ☐Chorionic villus sampling

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

☐Unknown ☐None
☐Fetal echocardiogram ☐Increased frequency prenatal ultrasounds

(Continued on next page.)

☐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

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

(Continued on next page.)

○6 ○7 ○8 ○9 ○10 ○>10

Duration of longest inpatient hospitalization (in days) for any reason during fifth pregnancy_____

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

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

Laboratory studies done on patient during fifth pregnancy

☐Unknown
☐Ammonia - 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

☐Ammonia - Abn high
☐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 fifth pregnancy: describe test(s) and result(s)

Additional interventions required during fifth pregnancy due to this metabolic condition

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

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

_____

Total maternal weight gain (in kg) during fifth pregnancy_____

Additional interventions planned for labor/delivery related to patient's disorder for fifth pregnancy

☐Unknown
☐None
☐Additional maternal lab monitoring
☐Altered anesthesia plan
☐Change in delivery site
☐IV fluids
☐Letter to OB/MFM specialist
☐Planned C-section
☐Referral for high risk OB management
☐Other

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

Actual interventions for labor/delivery related to patient's disorder for fifth pregnancy

☐Unknown
☐No
☐Letter to OB/MFM specialist
☐IV fluids
☐Planned C-section
☐Additional maternal lab monitoring
☐Change in delivery site
☐Referral for high risk OB management
☐Altered anesthesia plan
☐Other

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

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

Abnormal patient lab results during fifth delivery

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

Abnormal patient lab results during fifth delivery-other, specify

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

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

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

Patient death during or shortly after fifth delivery ○Unknown ○Yes ○No

Disorder contributed to death of mother during fifth delivery ○Unknown ○Yes ○No

Weeks gestation at time of fifth delivery - round to nearest week_____

Method of fifth delivery

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

Maternal inpatient days post- fifth delivery_____

Live delivery of fifth newborn ○Unknown ○Yes ○No

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

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

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

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

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

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

Birth measurements for fifth newborn ☐Unknown ☐Head circumference ☐Length ☐Weight

Birth head circumference_____

Birth head circumference units ○cm ○in

Birth length_____

Birth length units ○cm ○in

Birth weight_____

Birth weight units ○lbs ○kg ○gm ○oz

APGAR score at 1 minute for fifth newborn

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

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

APGAR score at 5 minutes for fifth newborn

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

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

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

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

Routine newborn screening done for fifth newborn

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

Describe abnormal newborn screen result for fifth newborn

Reason routine newborn screening was not done for fifth newborn

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

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

_____

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

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

Additional studies and results (examples: brain MRI, echocardiogram) done on 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

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 insuffiency
☐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

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

(Continued on next page.)

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

Complications during sixth pregnancy

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

Complications related to disorder during sixth pregnancy- specify

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

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

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

Complications during sixth pregnancy-other, specify_____

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

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

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

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

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

- ☐Unknown
- ☐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
- ☐Ammonia - Abn high
- ☐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 sixth pregnancy: describe test(s) and result(s)

Additional interventions required during sixth pregnancy due to this metabolic condition

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

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

_____

Total maternal weight gain (in kg) during sixth pregnancy_____

Additional interventions planned for labor/delivery related to patient's disorder for sixth pregnancy

☐Unknown
☐None
☐Additional maternal lab monitoring
☐Altered anesthesia plan
☐Change in delivery site
☐IV fluids
☐Letter to OB/MFM specialist
☐Planned C-section
☐Referral for high risk OB management
☐Other

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

Actual interventions for labor/delivery related to patient's disorder for sixth pregnancy

☐Unknown
☐No
☐Letter to OB/MFM specialist
☐IV fluids
☐Planned C-section
☐Additional maternal lab monitoring
☐Change in delivery site
☐Referral for high risk OB management
☐Altered anesthesia plan
☐Other

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

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

Abnormal patient lab results during sixth delivery

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

Abnormal patient lab results during sixth delivery-other, specify

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

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

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

Patient death during or shortly after sixth delivery ○Unknown ○Yes ○No

Disorder contributed to death of mother during sixth delivery ○Unknown ○Yes ○No

Weeks gestation at time of sixth delivery - round to nearest week_____

Method of sixth delivery

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

Maternal inpatient days post- sixth delivery_____

Live delivery of sixth newborn ○Unknown ○Yes ○No

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

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

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

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

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

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

Birth measurements for sixth newborn ☐Unknown ☐Head circumference ☐Length ☐Weight

Birth head circumference_____

Birth head circumference units ○cm ○in

Birth length_____

Birth length units ○cm ○in

Birth weight_____

Birth weight units ○lbs ○kg ○gm ○oz

APGAR score at 1 minute for sixth newborn

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

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

APGAR score at 5 minutes for sixth newborn

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

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

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

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

Routine newborn screening done for sixth newborn

○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

_____

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

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

Additional studies and results (examples: brain MRI, echocardiogram) done on 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

Cause(s) of male infertility for seventh pregnancy-other, specify_____

Cause(s) of female infertility for seventh pregnancy

☐Unknown ☐Age
☐Alcohol use ☐Cancer/cancer treatment related
☐Early menopause ☐Fallopian tube damage/blockage
☐Hyperprolactinemia ☐Pelvic adhesions
☐Polycystic ovary syndrome (PCOS) ☐Premature ovarian insuffiency
☐Over-exercise ☐Ovulation disorder
☐Thyroid problems ☐Tobacco use
☐Uterine fibroid ☐Weight related
☐Other

Cause(s) of female infertility for seventh pregnancy-other, specify_____

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

_____

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

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

Type of assisted reproductive technologies used during seventh pregnancy

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

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

_____

Partner tested for patient's disorder during seventh pregnancy

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

Partner also affected by disorder during seventh pregnancy ○Unknown ○Yes ○No

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

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

Type of prenatal testing performed for seventh pregnancy ☐Unknown ☐Biochemical ☐Molecular

Method of prenatal testing for seventh pregnancy

☐Unknown ☐Amniocentesis ☐Chorionic villus sampling

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

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

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

(Continued on next page.)

○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

Duration of longest inpatient hospitalization (in days) for any reason during seventh pregnancy_____

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

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

Laboratory studies done on patient during seventh pregnancy

- ☐Unknown
- ☐Ammonia - 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
- ☐Ammonia - Abn high
- ☐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 seventh pregnancy: describe test(s) and result(s)

Additional interventions required during seventh pregnancy due to this metabolic condition

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

Additional interventions required during 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
☐None
☐Additional maternal lab monitoring
☐Altered anesthesia plan
☐Change in delivery site
☐IV fluids
☐Letter to OB/MFM specialist
☐Planned C-section
☐Referral for high risk OB management
☐Other

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

Actual interventions for labor/delivery related to patient's disorder for seventh pregnancy

☐Unknown
☐No
☐Letter to OB/MFM specialist
☐IV fluids
☐Planned C-section
☐Additional maternal lab monitoring
☐Change in delivery site
☐Referral for high risk OB management
☐Altered anesthesia plan
☐Other

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

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)

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

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

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

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

Routine newborn screening done for seventh newborn

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

Describe abnormal newborn screen result for seventh newborn

Reason routine newborn screening was not done for seventh newborn

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

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

_____

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

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

Additional studies and results (examples: brain MRI, echocardiogram) done on 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

(Continued on next page.)

☐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 insuffiency
☐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

(Continued on next page.)

☐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

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

(Continued on next page.)

○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

☐Unknown
☐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

☐Ammonia - Abn high
☐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 eighth pregnancy: describe test(s) and result(s)

Additional interventions required during eighth pregnancy due to this metabolic condition

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

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

_____

Total maternal weight gain (in kg) during eighth pregnancy_____

Actual interventions for labor/delivery related to patient's disorder for eighth pregnancy

- ☐Unknown
- ☐No
- ☐Letter to OB/MFM specialist
- ☐IV fluids
- ☐Planned C-section
- ☐Additional maternal lab monitoring
- ☐Change in delivery site
- ☐Referral for high risk OB management
- ☐Altered anesthesia plan
- ☐Other

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

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

Abnormal patient lab results during eighth delivery

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

Abnormal patient lab results during eighth delivery-other, specify

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

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

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

Patient death during or shortly after eighth delivery ○Unknown ○Yes ○No

Disorder contributed to death of mother during eighth delivery ○Unknown ○Yes ○No

Weeks gestation at time of eighth delivery - round to nearest week_____

Method of eighth delivery

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

Maternal inpatient days post- eighth delivery_____

Live delivery of eighth newborn ○Unknown ○Yes ○No

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

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

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

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

(Continued on next page.)

☐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

(Continued on next page.)

○Unknown ○Yes - normal for all screened disorders

○Yes -abnormal ○Yes - results pending

○No

Describe abnormal newborn screen result for eighth newborn

Reason routine newborn screening was not done for eighth newborn

☐Unknown

☐Died prior to collection of NBS

☐Refused

☐Transferred to another facility prior to NBS collection

☐Transfused prior to collection of NBS

☐Other

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

_____

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

☐Unknown ☐Yes - biochemical normal

☐Yes - bichemical abnormal ☐Yes - molecular normal

☐Yes - molecular abnormal affected ☐Yes - molecular abnormal unaffected carrier

☐No

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

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

○Unknown ○Yes ○No

Abnormal newborn exam findings for eighth newborn

☐Unknown ☐None ☐Congenital heart disease

☐Dysmorphism ☐Lethargy ☐Microcephaly

☐Other congenital anomalies ☐Poor feeding ☐Respiratory distress

☐Seizure(s) ☐Small for gestational age ☐Other

Abnormal newborn exam findings for eighth newborn-other, specify_____

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

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

Eighth newborn currently alive ○Unknown ○Yes ○No

Current Pregnancy

Patient is pregnant ○Unknown ○Yes ○No

Patient's age at time of current pregnancy (in years)_____

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

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

History of infertility for current pregnancy

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

Cause(s) of infertility for current pregnancy

□Unknown □Male factor □Female factor
□Combined male-female factor

Cause(s) of male infertility for current pregnancy

□Unknown □Abnormal sperm production or function
□Age □Cancer/cancer treatment related
□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 insuffiency
□Over-exercise □Ovulation disorder
□Thyroid problems □Tobacco use
□Uterine fibroid □Weight related
□Other

Cause(s) of female infertility for current pregnancy-other, specify_____

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

_____

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

☐Unknown ☐None

☐Assisted reproductive technology ☐Fertility drugs

☐Surgery

Type of assisted reproductive technologies used during current pregnancy

☐Assisted hatching ☐ICSI ☐IVF

☐Surgical sperm aspiration ☐Other

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

_____

Partner tested for patient's disorder during current pregnancy

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

Partner also affected by disorder during current pregnancy ○Unknown ○Yes ○No

Preimplantation genetic diagnosis for disorder done for current pregnancy ○Unknown ○Yes ○No

Prenatal testing done for fetus for this disorder for current pregnancy ○Unknown ○Yes ○No

Type of prenatal testing performed for current pregnancy ☐Unknown ☐Biochemical ☐Molecular

Method of prenatal testing for current pregnancy

☐Unknown ☐Amniocentesis ☐Chorionic villus sampling

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

☐Unknown ☐None

☐Fetal echocardiogram ☐Increased frequency prenatal ultrasounds

☐Other

Additional prenatal testing performed on fetus as a result of parent's disorder for current pregnancy-other, specify_____

Current pregnancy terminated ○Unknown ○Yes ○No

Reason current pregnancy terminated

○Elective due to fetus affected with disorder

○Elective due to other fetal well-being unrelated to disorder

○Elective for other reason

○Elective due to maternal well-being

○Spontaneous

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

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for current pregnancy_____

Prenatal care received during current pregnancy ○Unknown ○Yes ○No

Weeks gestation prenatal care started during current pregnancy_____

Treatment prescribed for patient's disorder during current pregnancy

☐Unknown ☐Metabolic diet ☐Medications

☐Biochemical lab monitoring ☐Avoidance of fasting ☐Other

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

Patient in good metabolic condition prior to current pregnancy ○Unknown ○Yes ○No

Patient in good metabolic condition during first trimester of current pregnancy

○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)

(Continued on next page.)

☐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)

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

- ☐Unknown
- ☐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
- ☐Ammonia - Abn high
- ☐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

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

- ☐Unknown
- ☐Hospitalizations for hyperemesis/IV fluids
- ☐Tube feedings
- ☐Home lab monitoring
- ☐ED visits for hyperemesis/IV fluids
- ☐TPN
- ☐Additional medications
- ☐Increased frequency of lab monitoring

(Continued on next page.)

☐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

Pregnancy comments

H-PHE - 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
☐Periotoneal 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 |M|M| |D|D| |Y|Y|

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_____

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

☐Periotoneal 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 |M|M| |D|D| |Y|Y|

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

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

□Periotoneal 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 M M D D Y Y

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

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

□Periotoneal dialysis

Reason for fourth episode of dialysis

□Unknown □Hyperammonemia □MSUD □Organ failure □Sepsis

□Other

Reason for fourth episode of dialysis-other, specify

Start date of fourth episode of dialysis treatment |M|M| |D|D| |Y|Y|

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

☐Periotoneal dialysis

Reason for fifth episode of dialysis

(Continued on next page.)

☐Unknown ☐Hyperammonemia ☐MSUD ☐Organ failure ☐Sepsis
☐Other

Reason for fifth episode of dialysis-other, specify

Start date of fifth episode of dialysis treatment M M D D Y Y

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

Sixth Dialysis Treatment

Type(s) of dialysis received during sixth episode

☐Unknown ☐APD/CCPD ☐ECMO ☐CAPD

☐CVVH ☐CVVD ☐CVVHDF ☐Hemodialysis

☐Periotoneal dialysis

Reason for sixth episode of dialysis

☐Unknown ☐Hyperammonemia ☐MSUD ☐Organ failure ☐Sepsis

☐Other

Reason for sixth episode of dialysi-other, specify

Start date of sixth episode dialysis treatment |M|M| |D|D| |Y|Y|

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

Peak value of other metabolite during sixth episode of dialysis_____

Peak value of other metabolite during sixth episode of dialysis units_____

Reason sixth episode of dialysis treatment was stopped

○Unknown ○Acute episode resolved ○Treatment withdrawn ○Death

○Other

Reason sixth episode of dialysis treatment was stopped-other, specify_____

Seventh Dialysis Treatment

Type(s) of dialysis received during seventh episode

☐Unknown ☐APD/CCPD ☐ECMO ☐CAPD

☐CVVH ☐CVVD ☐CVVHDF ☐Hemodialysis

☐Periotoneal 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 |M|M| |D|D| |Y|Y|

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

(Continued on next page.)

○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

□Periotoneal 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 |M|M| |D|D| |Y|Y|

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

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

□Periotoneal 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 |M|M| |D|D| |Y|Y|

Duration (in days) of ninth episode of dialysis treatment_____

Metabolite(s) of concern during ninth episode of dialysis

□Unknown □None □Ammonia □Blood urea nitrogen

(Continued on next page.)

☐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
☐Periotoneal dialysis

Reason for tenth episode of dialysis

☐Unknown ☐Hyperammonemia ☐MSUD ☐Organ failure ☐Sepsis
☐Other

Reason for tenth episode of dialysis-other, specify

Start date of tenth episode of dialysis treatment |M|M| |D|D| |Y|Y|

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_____

If >10 dialysis treatments, for each episode: enter start date, duration (days), and reason for stopping dialysis

Comments

Dialysis comments

H-PHE - 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

☐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

(Continued on next page.)

☐Reappearance of liver disease ☐Rejection

☐Transplant related renal impairment ☐Other

Known complications post first transplant, other- specify_____

Second Transplant

Second organ received by transplant

☐Unknown ☐Bone marrow ☐Heart ☐Kidney ☐Liver ☐Lung

☐Pancreas ☐Stem Cell ☐Other

Second organ received by transplant, other- specify_____

Age (in days) at second transplant_____

Reason for second transplant

☐Unknown ☐Treatment of disorder ☐Renal failure ☐Liver failure

☐Heart failure ☐Other

Reason for second transplant, other- specify_____

Patient followed by Metabolism on an outpatient basis post second transplant ○Unknown ○Yes ○No

Number of outpatient metabolic visits in the last year post transplant

○1 ○2 ○3 ○4 ○5 ○6 ○7 ○8 ○9 ○10 ○>10

Metabolic labs monitored post second transplant ○Unknown ○Yes ○No

Metabolic lab monitoring post second transplant: Note date(s), test(s), and normal or abnormal result(s)

Complications during the second transplant procedure ○Unknown ○Yes ○No

Complications during the second transplant procedure- specify

☐Clotting ☐Death ☐Major bleeding ☐Other

Complications during the second transplant procedure, other- specify_____

Complications post second transplant ○Unknown ○Yes ○No

Complications post second transplant- specify

☐Unknown ☐Cancer

☐Cataract ☐Clotting

☐Death ☐Delayed graft function

☐Diabetes mellitus ☐High cholesterol

☐Hypertension ☐Infection

☐Major bleeding ☐Osteoporosis

☐Reappearance of heart disease ☐Reappearance of kidney disease

☐Reappearance of liver disease ☐Rejection

(Continued on next page.)

☐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

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_____

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_____

Provide details for any other transplants not listed above

Comments

Transplant comments