

Patient Name _____

Date ***bKT - Intake Demographics***

NBSTRN ID _____

Version 2.1.2

Center name

 A B C D E F G H I J K L M N O P Q R S T U V OtherIntake date **Consent**Consent obtained Yes No IRB ExemptAssent obtained Yes NoType of assent Written VerbalPermission to recontact Unknown Yes NoProtocol ID A**Demographics Information**

Patient last name _____

Patient first name _____

Date of birth

Age _____

Gestational age in weeks _____

Societal sex Unknown Male Female

Biological sex

Not tested Unknown XX genotype/Female

XY genotype/Male XXX Triple X syndrome XXY Klinefelter's syndrome

XO Turner's syndrome XXXY syndrome XXYY syndrome

Mosaic including XXXXY Penta X syndrome Other

Biological sex-other, specify _____

Biological mother's maiden name _____

Zip code _____

ConditionPatient condition category Organic acid disordersSpecify organic acid disorder diagnosis for the patient Beta-Ketothiolase deficiency (bKT)

Patient disorder identification method

 Unknown Abnormal newborn screen Abnormal labs

Patient Name _____

Date | |

Clinical presentation Family member with this condition

Family member with this condition

Biological mother Biological father Full sibling Half sibling Other

Family member with this condition _____

Care and Other Studies

Miles from home to primary care _____

Miles from home to specialty care _____

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

Name of primary care provider _____

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

Specify medical home-other, specify _____

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

Research study-other, specify _____

Clinicaltrials.gov identifier _____

Education

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)

Patient Name _____

Date

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

Patient education

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

Special education services received prior to intake Unknown Yes No

Age patient qualified for special education services

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

Ancestral Origin, Race and Ethnicity

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

Ancestral Origin-Africa

- Unknown Egypt Eritrea Ethiopia Liberia Somalia South Africa
- Other

Ancestral Origin-Africa-Other, specify _____

Ancestral Origin-Asia

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

Ancestral Origin-Asia-Other, specify _____

Ancestral Origin-Europe

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

Patient Name _____

Date Sweden Switzerland Ukraine United Kingdom OtherAncestral Origin-Europe-Italy Unknown SicilyAncestral Origin-Europe-Romania Unknown TransylvaniaAncestral Origin-Europe-United Kingdom Unknown England Northern Ireland Scotland Wales

Ancestral Origin-Europe-Other, specify _____

Ancestral Origin-North America

 Unknown Aleutian Islands Canada Dominican Republic Honduras Mexico Puerto Rico United States OtherAncestral Origin-North America-Canada Unknown French Canadian

Ancestral Origin-North America-Other, specify _____

Ancestral Origin-South America Unknown Colombia Venezuela Other

Ancestral Origin-South America-Other, specify _____

Ancestral Origin-Oceania Unknown Australia Other

Ancestral Origin-Oceania-Other, specify _____

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

Ancestral Origin-Other, specify _____

Race

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

Race-American Indian/Alaskan Native-Other, specify _____

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

Maternal age (in years) at patient's birth _____

Paternal age (in years) at patient's birth _____

Mother's marital status at patient's birth

 Unknown Married Widowed Divorced Separated Never married Living with partner

County mother resides in at patient's birth _____

State mother resides in at patient's birth

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

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

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

Medical Coverage

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

Patient Name _____

Date ***bKT - Intake Family History*****Family History**Consanguinity Unknown Yes NoPatient was adopted Unknown Yes No

Number of pregnancies for patient's mother _____

Number of live births for patient's mother _____

Siblings

Number of biological siblings in the patient's family _____

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

Sibling 1: Year of birth _____

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

Sibling 1: NBSTRN ID for this study _____

Sibling 1: Method of diagnosis

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

Sibling 1: Method of diagnosis-other, specify _____

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

Sibling 1: Results of newborn screening for this condition

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

Sibling 1: Specify the type of diagnostic tests performed

Unknown Biochemical Molecular Imaging Other

Sibling 1: Diagnostic tests performed-other, specify _____

Sibling 1: Deceased Unknown Yes No

Sibling 1: Age of death (in years) _____

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

Sibling 2: Year of birth _____

Sibling 2: Affected with this condition Unknown Yes No

Patient Name _____

Date Sibling 2: Enrolled in this study Unknown Yes No

Sibling 2: NBSTRN ID for this study _____

Sibling 2: Method of diagnosis

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

Sibling 2: Method of diagnosis-other, specify _____

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

Sibling 2: Results of newborn screening for this condition

 Unavailable Presumptive positive Negative/Normal BorderlineSibling 2: Diagnostic tests performed for this condition Unknown Yes No

Sibling 2: Specify the type of diagnostic tests performed

 Unknown Biochemical Molecular Imaging Other

Sibling 2: Diagnostic tests performed-other, specify _____

Sibling 2: Deceased Unknown Yes No

Sibling 2: Age of death (in years) _____

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

Sibling 3: Year of birth _____

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

Sibling 3: NBSTRN ID for this study _____

Sibling 3: Method of diagnosis

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

Sibling 3: Method of diagnosis-other, specify _____

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

Sibling 3: Results of newborn screening for this condition

 Unavailable Presumptive positive Negative/Normal BorderlineSibling 3: Diagnostic tests performed for this condition Unknown Yes No

Sibling 3: Specify the type of diagnostic tests performed

 Unknown Biochemical Molecular Imaging Other

Sibling 3: Diagnostic tests performed-other, specify _____

Sibling 3: Deceased Unknown Yes No

Patient Name _____

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

Sibling 3: Age of death (in years)_____

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

Sibling 4: Year of birth_____

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

Sibling 4: NBSTRN ID for this study_____

Sibling 4: Method of diagnosis

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

Sibling 4: Method of diagnosis-other, specify_____

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

Sibling 4: Results of newborn screening for this condition

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

Sibling 4: Specify the type of diagnostic tests performed

Unknown Biochemical Molecular Imaging Other

Sibling 4: Diagnostic tests performed-other, specify_____

Sibling 4: Deceased Unknown Yes No

Sibling 4: Age of death (in years)_____

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

Sibling 5: Year of birth_____

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

Sibling 5: NBSTRN ID for this study_____

Sibling 5: Method of diagnosis

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

Sibling 5: Method of diagnosis-other, specify_____

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

Patient Name _____

Date

Sibling 5: Results of newborn screening for this condition

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

Sibling 5: Specify the type of diagnostic tests performed

 Unknown Biochemical Molecular Imaging Other

Sibling 5: Diagnostic tests performed-other, specify _____

Sibling 5: Deceased Unknown Yes No

Sibling 5: Age of death (in years) _____

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

Sibling 6: Year of birth _____

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

Sibling 6: NBSTRN ID for this study _____

Sibling 6: Method of diagnosis

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

Sibling 6: Method of diagnosis-other, specify _____

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

Sibling 6: Results of newborn screening for this condition

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

Sibling 6: Specify the type of diagnostic tests performed

 Unknown Biochemical Molecular Imaging Other

Sibling 6: Diagnostic tests performed-other, specify _____

Sibling 6: Deceased Unknown Yes No

Sibling 6: Age of death (in years) _____

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

Sibling 7: Year of birth _____

Sibling 7: Affected with this condition Unknown Yes No

Patient Name _____

Date Sibling 7: Enrolled in this study Unknown Yes No

Sibling 7: NBSTRN ID for this study _____

Sibling 7: Method of diagnosis

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

Sibling 7: Method of diagnosis-other, specify _____

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

Sibling 7: Results of newborn screening for this condition

Unavailable Presumptive positive Negative/Normal Borderline

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

Sibling 7: Specify the type of diagnostic tests performed

Unknown Biochemical Molecular Imaging Other

Sibling 7: Diagnostic tests performed-other, specify _____

Sibling 7: Deceased Unknown Yes No

Sibling 7: Age of death (in years) _____

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

Sibling 8: Year of birth _____

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

Sibling 8: NBSTRN ID for this study _____

Sibling 8: Method of diagnosis

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

Sibling 8: Method of diagnosis-other, specify _____

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

Sibling 8: Results of newborn screening for this condition

Unavailable Presumptive positive Negative/Normal Borderline

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

Sibling 8: Specify the type of diagnostic tests performed

Unknown Biochemical Molecular Imaging Other

Sibling 8: Diagnostic tests performed-other, specify _____

Sibling 8: Deceased Unknown Yes No

Patient Name _____

Date

Sibling 8: Age of death (in years) _____

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

Biological mother: NBSTRN ID for this study _____

Biological mother: Method of diagnosis

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

Biological mother: Method of diagnosis-other, specify _____

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

Biological mother: Results of newborn screening for this condition

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

Biological mother: Specify the type of diagnostic tests performed

 Unknown Biochemical Molecular Imaging Other

Biological mother: Diagnostic tests performed-other, specify _____

Biological mother: Deceased Unknown Yes No

Biological mother: Age of death (in years) _____

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

Biological father: NBSTRN ID for this study _____

Biological father: Method of diagnosis

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

Biological father: Method of diagnosis-other, specify _____

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

Biological father: Results of newborn screening for this condition

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

Biological father: Specify the type of diagnostic tests performed

 Unknown Biochemical Molecular Imaging Other

Biological father: Diagnostic tests performed-other, specify _____

Biological father: Deceased Unknown Yes No

Patient Name _____

Date

Biological father: Age of death (in years) _____

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

Maternal grandmother: NBSTRN ID for this study _____

Maternal grandmother: Method of diagnosis

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

Maternal grandmother: Method of diagnosis-other, specify _____

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

Maternal grandmother: Specify the type of diagnostic tests performed

 Unknown Biochemical Molecular Imaging Other

Maternal grandmother: Diagnostic tests performed-other, specify _____

Maternal grandmother: Deceased Unknown Yes No

Maternal grandmother: Age of death (in years) _____

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

Maternal grandfather: NBSTRN ID for this study _____

Maternal grandfather: Method of diagnosis

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

Maternal grandfather: Method of diagnosis-other, specify _____

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

Maternal grandfather: Specify the type of diagnostic tests performed

 Unknown Biochemical Molecular Imaging Other

Maternal grandfather: Diagnostic tests performed-other, specify _____

Maternal grandfather: Deceased Unknown Yes No

Maternal grandfather: Age of death (in years) _____

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

Paternal grandmother: NBSTRN ID for this study _____

Paternal grandmother: Method of diagnosis

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

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

Paternal grandmother: Method of diagnosis-other, specify_____

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

Paternal grandmother: Specify the type of diagnostic tests performed

- Unknown Biochemical Molecular Imaging Other

Paternal grandmother: Diagnostic tests performed-other, specify_____

Paternal grandmother: Deceased Unknown Yes No

Paternal grandmother: Age of death (in years)_____

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

Paternal grandfather: Affected with this condition Unknown Yes No

Paternal grandfather: Enrolled in this study Unknown Yes No

Paternal grandfather: NBSTRN ID for this study_____

Paternal grandfather: Method of diagnosis

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

Paternal grandfather: Method of diagnosis-other, specify_____

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

Paternal grandfather: Specify the type of diagnostic tests performed

- Unknown Biochemical Molecular Imaging Other

Paternal grandfather: Diagnostic tests performed-other, specify_____

Paternal grandfather: Deceased Unknown Yes No

Paternal grandfather: Age of death (in years)_____

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

Other Family

Other affected family members NOT listed above Unknown Yes No

Number of other affected family members that are NOT listed above_____

Relationship to affected family member 1_____

Relationship to affected family member 2_____

Relationship to affected family member 3_____

Relationship to affected family member 4_____

Relationship to affected family member 5_____

Relationship to affected family member 6_____

Relationship to affected family member 7_____

Relationship to affected family member 8_____

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

Relationship to affected family member 9 _____

Relationship to affected family member 10 _____

Comments

Intake family history comments

Patient Name _____

Date

bKT - 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 DNAChorionic villus sampling (CVS) diagnosis Biochemical/enzyme DNA

Issues concerning mother's pregnancy with this patient

- Unknown None Pregnancy complications
 Assisted reproduction

Pregnancy complications/risk factors

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

Type of prenatal substance exposure

- Unknown Alcohol Tobacco Illicit drugs
 Harmful chemicals Known teratogens

Maternal treatment for affected fetus Unknown None Yes No

Patient Name _____

Date

Type of maternal treatment for affected fetus

--

Pregnancy complication/risk factor-other, specify _____

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

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

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

Pregnancy

Patient has biological children Unknown Yes No

Please complete the pregnancy form.

Patient is pregnant Unknown Yes No

Please complete the pregnancy form.

Neonatal History

Patient's birth was a result of multiple gestation pregnancy

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

Specify other number of multiples _____

Congenital anomalies Unknown Yes No

Type of congenital anomalies

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

Patient Name _____

Date

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

Type of congenital anomalies-other, specify _____

Neonatal complications Unknown Yes No

Type of neonatal complications

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

Type of neonatal complications-other, specify _____

Type of neonatal nutrition

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

Type of neonatal nutrition-other, specify _____

Birth MeasurementsBirth measurements Unknown Head circumference Length Weight

Birth head circumference _____

Birth head circumference units cm in

Patient Name _____

Date

Birth length _____

Birth length units cm in

Birth weight _____

Birth weight units lbs kg gm oz

Health History

Patient has had an outpatient speciality visit Unknown Yes No

Date of last outpatient specialty visit

Days of age from birth until intervention for this condition _____

Days of age from birth until first seen by subspecialist _____

Dialysis

Dialysis (any type) prior to intake Unknown Yes No

Please complete the dialysis form.

Transplants

Transplant prior to intake

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

Please complete the transplant form.

Hospitalizations prior to intake Unknown Yes No

Number of hospitalizations prior to intake related to this condition _____

Number of hospitalizations prior to intake not related to this condition _____

Genetic counseling provided Unknown Yes No

Provider of genetic counseling

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

Provider of genetic counseling, other- specify _____

Comorbidities at time of intake

Patient Name _____

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

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

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 NoPatient has biospecimen stored Unknown Yes NoType of patient biospecimen Blood Urine Other tissue

Type of patient biospecimen-other tissue, specify _____

Specify biospecimen location _____

Age (in years) at time of first Dexa scan _____

Emergency Management

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

Name of web-based emergency alert program _____

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

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

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

Unknown

Yes

No

Comments

Intake past health history comments

Patient Name _____

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

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

Reason for first newborn screen Routine Research

Carnitine free (C0) on first newborn screen _____

Carnitine free (C0) on first newborn screen units umol/L

Tiglylcarnitine (C5:1) on first newborn screen _____

Tiglylcarnitine (C5:1) on first newborn screen units umol/L

3-Hydroxyisovalerylcarnitine (C5-OH) on first newborn screen _____

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

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

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

Octanoylcarnitine (C8) on first newborn screen _____

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

Other result/report on first newborn screen

Other result/report on first newborn screen units _____

Date second newborn screen collected

Reason for second newborn screen

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

Reason for second newborn screen-other, specify _____

Carnitine free (C0) on second newborn screen _____

Carnitine free (C0) on second newborn screen units umol/L

Tiglylcarnitine (C5:1) on second newborn screen _____

Tiglylcarnitine (C5:1) on second newborn screen units umol/L

Patient Name _____

Date

3-Hydroxyisovalerylcarnitine (C5-OH) on second newborn screen _____

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

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

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

Octanoylcarnitine (C8) on second newborn screen _____

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

Other result/report on second newborn screen

Other result/report on second newborn screen units _____

Date third newborn screen collected

Reason for third newborn screen

- Unsatisfactory Borderline Result NICU Protocol TPN Transfused
- State Mandate Other

Reason for third newborn screen-other, specify _____

Carnitine free (C0) on third newborn screen _____

Carnitine free (C0) on third newborn screen units umol/L

Tiglylcarnitine (C5:1) on third newborn screen _____

Tiglylcarnitine (C5:1) on third newborn screen units umol/L

3-Hydroxyisovalerylcarnitine (C5-OH) on third newborn screen _____

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

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

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

Octanoylcarnitine (C8) on third newborn screen _____

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

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

Patient Name _____

Date

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

Right ear: Diagnosis: Hearing loss? Yes No

Right ear: Diagnosis: Degree of hearing loss

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

Right ear: Diagnosis: Type of hearing loss

Sensorineural

Conductive

Mixed

Auditory neuropathy/Auditory dys-synchrony

Other

Left ear: Diagnosis: Hearing loss? Yes No

Left ear: Diagnosis: Degree of hearing loss

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

Left ear: Diagnosis: Type of hearing loss

Sensorineural

Conductive

Mixed

Auditory neuropathy/Auditory dys-synchrony

Other

Comments

Intake newborn screening comments

Patient Name _____

Date ***bKT - Intake Initial Testing***

Patient status at time of NBS reporting to specialty center

 Unknown Well Symptomatic Deceased

Patient symptoms at time of initial contact

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

Patient Name _____

Date

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

Patient symptoms at time of initial contact-other, specify _____

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

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

Patient Name _____

Date

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

Diagnostic Testing

Diagnostic labs performed

- Unknown
- None
- Mitochondrial acetoacetyl-CoA thiolase enzyme assay from fibroblasts
- Plasma acylcarnitine profile
- Plasma amino acid profile
- Plasma carnitine
- Urine acylcarnitine
- Urine organic acids
- Other

Number of times mitochondrial acetoacetyl-CoA enzyme assay from fibroblasts done 1 2

Mitochondrial acetoacetyl-CoA enzyme assay from fibroblasts

- Unknown Within normal limits Abnormal

Mitochondrial acetoacetyl-CoA enzyme assay from fibroblasts significance

- Diagnostic Non-diagnostic

Mitochondrial acetoacetyl-CoA enzyme assay from fibroblasts value _____

Mitochondrial acetoacetyl-CoA enzyme assay from fibroblasts units nmol/min nmol/h

Mitochondrial acetoacetyl-CoA enzyme assay from fibroblasts reference range

Second mitochondrial acetoacetyl-CoA enzyme assay from fibroblasts

- Unknown Within normal limits Abnormal

Second mitochondrial acetoacetyl-CoA enzyme assay from fibroblasts significance

- Diagnostic Non-diagnostic

Second mitochondrial acetoacetyl-CoA enzyme assay from fibroblasts value _____

Second mitochondrial acetoacetyl-CoA enzyme assay from fibroblasts units nmol/min nmol/h

Second mitochondrial acetoacetyl-CoA enzyme assay from fibroblasts reference range

Plasma acylcarnitine profile Unknown Within normal limits Abnormal

Plasma acylcarnitine profile significance Diagnostic Non-diagnostic

Patient Name _____

Date |||||

Plasma acylcarnitine profile comments

Plasma amino acid profile Unknown Within normal limits Abnormal

Plasma amino acid profile significance Diagnostic Non-diagnostic

Plasma amino acid comments

Plasma carnitine levels

Unknown Free carnitine Total carnitine

Esterified carnitine Esterified:Free carnitine ratio

Free plasma carnitine Unknown Within normal limits Abnormal

Free plasma carnitine significance Diagnostic Non-diagnostic

Free plasma carnitine value_____

Free plasma carnitine units umol/L mg/dL

Free plasma carnitine reference range_____

Total plasma carnitine Unknown Within normal limits Abnormal

Total plasma carnitine significance Diagnostic Non-diagnostic

Total plasma carnitine value_____

Total plasma carnitine units umol/L mg/dL

Total plasma carnitine reference range_____

Esterified plasma carnitine Unknown Within normal limits Abnormal

Esterified plasma carnitine significance Diagnostic Non-diagnostic

Esterified plasma carnitine value_____

Esterified plasma carnitine units umol/L mg/dL

Esterified plasma carnitine reference range_____

Esterified:Free plasma carnitine ratio Unknown Within normal limits Abnormal

Esterified:Free plasma carnitine ratio significance Diagnostic Non-diagnostic

Esterified:Free plasma carnitine ratio value_____

Esterified:Free plasma carnitine ratio reference range_____

Urine acylcarnitine

Unknown Urine C4 acylcarnitine Urine C5-OH acylcarnitine

Urine C5-DC acylcarnitine

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

Urine C4 acylcarnitine Unknown Within normal limits Abnormal

Urine C4 acylcarnitine significance Diagnostic Non-diagnostic

Urine C4 acylcarnitine value _____

Urine C4 acylcarnitine units mmol/mol Cr

Urine C4 acylcarnitine reference range _____

Urine C5-OH acylcarnitine Unknown Within normal limits Abnormal

Urine C5-OH acylcarnitine significance Diagnostic Non-diagnostic

Urine C5-OH acylcarnitine value _____

Urine C5-OH acylcarnitine units mmol/mol Cr

Urine C5-OH acylcarnitine reference range _____

Urine C5-DC acylcarnitine Unknown Within normal limits Abnormal

Urine C5-DC acylcarnitine significance Diagnostic Non-diagnostic

Urine C5-DC acylcarnitine value _____

Urine C5-DC acylcarnitine units mmol/mol Cr

Urine C5-DC acylcarnitine reference range _____

Urine organic acids Unknown Within normal limits Abnormal

Urine organic acids significance Diagnostic Non-diagnostic

Urine organic acid comments

Diagnostic lab-other, specify _____

Other diagnostic lab Unknown Within normal limits Abnormal

Other diagnostic lab significance Diagnostic Non-diagnostic

Other diagnostic lab comments

Other diagnostic lab value _____

Other diagnostic lab units _____

Other diagnostic lab reference range _____

Genetic Testing

Type of genetic/genomic testing

- | | | |
|--------------------------------------|---|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Not done | <input type="checkbox"/> Done, not available |
| <input type="checkbox"/> Single gene | <input type="checkbox"/> Mutation panel | <input type="checkbox"/> Exome sequencing |

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

- 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 BKT ACAT1 Other

Gene(s) associated with BKT-other, specify _____

ACAT1: Specify allele 1 _____

ACAT1: Specify allele 2 _____

Other: Specify allele 1 _____

Other: Specify allele 2 _____

Maternal genetic testing done Unknown Yes No Genotyping in progress

Mother: Allele 1 _____

Mother: Allele 2 _____

Paternal genetic testing done Unknown Yes No Genotyping in progress

Father: Allele 1 _____

Father: Allele 2 _____

Comments

Initial testing comments

Patient Name _____

Date ***bKT - Visit Demographics And History***Visit Date Date of last outpatient speciality visit **Consent**Patient consent valid Unknown Yes No

Obtain new consent prior to completing data entry.

Care and Other Studies

Providers seen at this visit

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

Providers seen at this visit, other- specify _____

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

Location of visit-other, specify _____

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

Miles from home to primary care _____

Miles from home to specialty care _____

Name of primary care center _____

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

Identify the research study _____

Clinicaltrials.gov identifier _____

EducationEducation status has changed since the last visit Unknown Yes No

Complete education questions on the Demographics form.

Medical Coverage

Medical coverage at visit

- Unknown
 None
 Commercial/private
 Medicaid

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

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

Medical coverage at visit-Patient assistance program, specify _____

Medical coverage at visit-Other, specify _____

Family History

Patient has new biological sibling since last visit Unknown Yes No

Complete sibling history questions on the Family History form.

Comments

Visit demographics and history comments

Patient Name _____

Date |||||

bKT - 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

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

 Unknown Yes No

Lowest blood glucose value reported since last outpatient metabolic visit _____

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

Lowest blood glucose range reported since last outpatient metabolic visit _____

Sick Visits

Sick visits since last outpatient visit Unknown Yes No

Number of sick visits _____

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

Location for sick visit 1

- Unknown Emergency department Retail clinic
 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

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

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 | |

Reason for sick visit 2 Unknown Condition related Condition unrelated

Sick visit 2 was a condition exacerbation Unknown Yes No

Location for sick visit 2

Unknown Emergency department Retail clinic

Primary care Specialty center Urgent care

Direct hospital admission Other

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

Name of hospital for sick visit 2 _____

ICD-9 codes for sick visit 2 known Yes No

ICD-9 codes for sick visit 2 _____

Diagnosis for sick visit 2 _____

Number of inpatient days for sick visit 2 _____

Number of ICU days for sick visit 2 _____

Date of sick visit 3 | |

Reason for sick visit 3 Unknown Condition related Condition unrelated

Sick visit 3 was a condition exacerbation Unknown Yes No

Location for sick visit 3

Unknown Emergency department Retail clinic

Primary care Specialty center Urgent care

Direct hospital admission Other

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

Name of hospital for sick visit 3 _____

ICD-9 codes for sick visit 3 known Yes No

ICD-9 codes for sick visit 3 _____

Diagnosis for sick visit 3 _____

Number of inpatient days for sick visit 3 _____

Number of ICU days for sick visit 3 _____

Date of sick visit 4 | |

Reason for sick visit 4 Unknown Condition related Condition unrelated

Sick visit 4 was a condition exacerbation Unknown Yes No

Patient Name _____

Date

Location for sick visit 4

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

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

Name of hospital for sick visit 4 _____

ICD-9 codes for sick visit 4 known Yes No

ICD-9 codes for sick visit 4 _____

Diagnosis for sick visit 4 _____

Number of inpatient days for sick visit 4 _____

Number of ICU days for sick visit 4 _____

Date of sick visit 5 Reason for sick visit 5 Unknown Condition related Condition unrelatedSick visit 5 was a condition exacerbation Unknown Yes No

Location for sick visit 5

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

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

Name of hospital for sick visit 5 _____

ICD-9 codes for sick visit 5 known Yes No

ICD-9 codes for sick visit 5 _____

Diagnosis for sick visit 5 _____

Number of inpatient days for sick visit 5 _____

Number of ICU days for sick visit 5 _____

Date of sick visit 6 Reason for sick visit 6 Unknown Condition related Condition unrelatedSick visit 6 was a condition exacerbation Unknown Yes No

Location for sick visit 6

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

Patient Name _____

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

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 unrelatedSick visit 7 was a condition exacerbation Unknown Yes No

Location for sick visit 7

Unknown Emergency department Retail clinicPrimary care Specialty center Urgent careDirect hospital admission OtherPatient was admitted to the hospital as a result of sick visit 7 Unknown Yes No

Name of hospital for sick visit 7 _____

ICD-9 codes for sick visit 7 known Yes No

ICD-9 codes for sick visit 7 _____

Diagnosis for sick visit 7 _____

Number of inpatient days for sick visit 7 _____

Number of ICU days for sick visit 7 _____

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

Reason for sick visit 8 Unknown Condition related Condition unrelatedSick visit 8 was a condition exacerbation Unknown Yes No

Location for sick visit 8

Unknown Emergency department Primary careSpecialty center Urgent care Direct hospital admissionOtherPatient 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 unrelatedSick visit 9 was a condition exacerbation Unknown Yes No

Patient Name _____

Date

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

Location for sick visit 10

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

ProceduresAnesthesia since last visit Unknown Yes NoSpecific anesthesia precautions recommended due to metabolic condition Unknown Yes No

Surgical precautions taken

Anesthesia complications Unknown Yes No

Patient Name _____

Date

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

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

Visit health history comments

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

bKT - 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 _____

Comments

Visit findings comments

Patient Name _____

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

Number of missed subspecialty visits _____

Other health services currently received Unknown Yes No

Specify other current health services

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

Specify other current health services-other, specify _____

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

Name of primary care provider _____

Preventive care status

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

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

Transplant organ-other, specify _____

Community resources currently received Unknown Yes No

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

Specify current community resources

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

Specify current community resources-other, specify _____

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

Specify current family support-other, specify _____

Specify medical home Unknown Primary care center Speciality care center Other

Specify medical home-other, specify _____

Specify current social services

- Unknown County Developmental disability
- Medical Mental health Other

Specify current social services-other, specify _____

Emergency Management

Patient currently has emergency specialty contact information

Unknown Yes No

Not needed for this condition

Type of emergency contact information

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

Type of emergency contact information-other, specify _____

Developmental Assessment

Developmental assessment done at this visit Unknown Yes No

Standardized developmental screening tool(s) used Unknown Yes No

Patient Name _____

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

Referred for further behavioral assessment Unknown Yes No

Referred for further behavioral assessment-Explain

 Previously referred Family declined further assessments

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

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

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

Education

Special education assessment recommended

 Unknown Yes No Special education services already received

Reason special education services received

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

Reason special education services received-other, specify _____

Special education category

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

Patient Name _____

Date

Speech or language impairments

Traumatic brain injury

Other health disabilities

Special education, other- specify _____

Comments

Visit ancillary care comments

Patient Name _____

Date ***bKT - Visit Lab Studies*****Biochemical Labs**

Biochemical labs associated with this visit

- Unknown
 None
 Blood spot acylcarnitine profile
 Mitochondrial acetoacetyl-CoA thiolase enzyme assay from fibroblasts
 Plasma acylcarnitine profile
 Plasma amino acid profile
 Plasma carnitine
 Urine acylcarnitine
 Urine ketones
 Urine organic acids
 Other

Blood spot acylcarnitine profile

- Within normal limits Abnormal In progress Results unavailable

Blood spot acylcarnitine profile comments

Please complete information about the mitochondrial acetoacetyl-CoA thiolase enzyme assay from fibroblasts on the Intake Initial Testing form.

- Plasma acylcarnitine profile Within normal limits Abnormal In progress Results unavailable

Plasma acylcarnitine profile comments

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

Plasma amino acid comments

Plasma carnitine levels

Patient Name _____

Date

- Unknown Free carnitine Total carnitine
 Esterified carnitine Esterified:Free carnitine ratio
 Free plasma carnitine Within normal limits Abnormal In progress Results unavailable

Free plasma carnitine value_____

Free plasma carnitine units umol/L mg/dL

Free plasma carnitine reference range_____

- Total plasma carnitine Within normal limits Abnormal In progress Results unavailable

Total plasma carnitine value_____

Total plasma carnitine units umol/L mg/dL

Total plasma carnitine reference range_____

- Esterified plasma carnitine Within normal limits Abnormal In progress Results unavailable

Esterified plasma carnitine value_____

Esterified plasma carnitine units umol/L mg/dL

Esterified plasma carnitine reference range_____

- Esterified:Free plasma carnitine ratio

Within normal limits Abnormal In progress Results unavailable

Esterified:Free plasma carnitine ratio value_____

Esterified:Free plasma carnitine ratio reference range_____

Urine acylcarnitine

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

- Urine C4 acylcarnitine Within normal limits Abnormal In progress Results unavailable

Urine C4 acylcarnitine value_____

Urine C4 acylcarnitine units mmol/mol Cr

Urine C4 acylcarnitine reference range_____

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

Urine C5-OH acylcarnitine value_____

Urine C5-OH acylcarnitine units mmol/mol Cr

Urine C5-OH acylcarnitine reference range_____

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

Urine C5-DC acylcarnitine value_____

Urine C5-DC acylcarnitine units mmol/mol Cr

Urine C5-DC acylcarnitine reference range_____

- Urine ketones Within normal limits Abnormal In progress Results unavailable

Patient Name _____

Date

Urine ketones comments

Urine organic acids Within normal limits Abnormal In progress Results unavailable

Urine organic acid comments

Biochemical lab associated with this visit-other, specify _____

Other visit lab Within normal limits Abnormal In progress Results unavailable

Other visit lab comments

Other visit lab value _____

Other visit lab units _____

Other visit lab reference range _____

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

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

Chemistry Labs

Chemistry labs associated with this visit

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

Arterial blood gas Within normal limits Abnormal In progress Results unavailable

Arterial blood gas comments

Arterial blood gas value _____

Patient Name _____

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

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

Patient Name _____

Date

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

Patient Name _____

Date

Platelet count units THOU/uL 10⁹/L 10³/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 10⁶/uL 10¹²/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 10³/uL 10⁹/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

Patient Name _____

Date

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 _____

Patient Name _____

Date

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/1.73m² mL/min/1.73m³

Nuclear medicine glomerular filtration rate reference range _____

Renal labs-other, specify _____

Other renal labs Within normal limits Abnormal In progress Results unavailable

Patient Name _____

Date

Other renal lab comments

Other renal lab value _____

Other renal lab units _____

Other renal lab reference range _____

Miscellaneous Labs

Miscellaneous labs associated with this visit

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

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

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

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 _____

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

Other miscellaneous lab units _____

Other miscellaneous lab reference range _____

Comments

Visit lab studies comments

Patient Name _____

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

Type of home monitoring

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

GlucoseFrequency of glucose home monitoring Unknown Only when symptomatic Routinely

Specify routine frequency _____

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

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

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

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

Type of branched chain amino acid home monitoring

- Unknown Alloisoleucine Isoleucine Leucine Valine

Sample type of branched chain amino acid home monitoring

- Unknown Filter paper blood Plasma

Frequency of branched chain amino acid home monitoring

- Unknown Only when symptomatic Routinely

Specify routine frequency _____

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

Alloisoleucine

Recommended therapeutic alloisoleucine goal _____

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

Average alloisoleucine on home monitoring samples _____

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

Average percent above recommended alloisoleucine therapeutic goal _____

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

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 _____

Patient Name _____

Date

Average percent below recommended phenylalanine therapeutic goal _____

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

TyrosineSample type of tyrosine home monitoring Unknown Filter paper blood PlasmaFrequency of tyrosine home monitoring Unknown Only when symptomatic Routinely

Specify routine frequency _____

Recommended therapeutic tyrosine goal _____

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

Lowest tyrosine value on home monitoring samples _____

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

Average tyrosine on home monitoring samples _____

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

Average percent below recommended tyrosine therapeutic goal _____

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

Dinitrophenylhydrazine

Frequency of dinitrophenylhydrazine home monitoring

 Unknown Only when symptomatic Routinely

Specify routine frequency _____

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

Result of dinitrophenylhydrazine home monitoring samples

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

Specify routine frequency _____

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

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

Specify routine frequency _____

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

Other

Other type of home monitoring, specify _____

Frequency of other home monitoring Unknown Only when symptomatic Routinely

Patient Name _____

Date

Specify routine frequency _____

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

Other Studies

Physiological tests associated with this visit

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

Electrocardiogram

Electrocardiogram date

Electrocardiogram findings of note

Electroencephalography

Electroencephalography date

Electroencephalography findings of note

Electromyography

Electromyography date

Electromyography findings of note

Holter Monitoring

Holter monitoring date

Holter monitoring findings of note

Cardiac Stress Test

Cardiac stress test date

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

Cardiac stress test findings of note

Imaging Studies

Imaging studies associated with this visit

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

Abdominal imaging

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

Abdominal findings of note

Cardiac Imaging

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

Cardiac findings of note

Musculoskeletal imaging

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

Musculoskeletal findings of note

Neurological imaging

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

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

Positron emission tomography (PET) scan Abn

Evidence of abnormal myelination on CNS imaging Unknown Yes No

Neurological findings of note

Renal/Pelvic/Genital imaging

Unknown

Genitogram WNL

Genitogram Abn

Nuclear medicine dimercapto succinic acid (DMSA) renal scan WNL

Nuclear medicine dimercapto succinic acid (DMSA) renal scan Abn

Pelvic ultrasound WNL

Pelvic ultrasound Abn

Renal ultrasound WNL

Renal ultrasound Abn

Testicular ultrasound WNL

Testicular ultrasound Abn

VCUG WNL

VCUG Abn

Other

Renal/pelvic/genital findings of note

Imaging studies-other, specify _____

Other imaging studies date

Other imaging studies explanation

Dexa Scan

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

Number of Dexa scans performed 1 2

First Dexa scan sites

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

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

First Dexa scan: Heel result Unknown Abnormal Normal

First Dexa scan: Heel Zscore _____

First Dexa scan: Hip result Unknown Abnormal Normal

First Dexa scan: Hip Zscore _____

First Dexa scan: Pelvis result Unknown Abnormal Normal

First Dexa scan: Pelvis Zscore _____

First Dexa scan: Spine result Unknown Abnormal Normal

First Dexa scan: Spine Zscore _____

First Dexa scan: Wrist result Unknown Abnormal Normal

First Dexa scan: Wrist Zscore _____

First Dexa scan: Total body result Unknown Abnormal Normal

First Dexa scan: Total body Zscore _____

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

First Dexa scan: Total body minus head Zscore _____

First Dexa scan site-other, specify _____

First Dexa scan: Other site result Unknown Abnormal Normal

First Dexa scan: Other site Zscore _____

Second Dexa scan sites

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

Second Dexa scan: Heel result Unknown Abnormal Normal

Second Dexa scan: Heel Zscore _____

Second Dexa scan: Hip result Unknown Abnormal Normal

Second Dexa scan: Hip Zscore _____

Second Dexa scan: Pelvis result Unknown Abnormal Normal

Second Dexa scan: Pelvis Zscore _____

Second Dexa scan: Spine result Unknown Abnormal Normal

Second Dexa scan: Spine Zscore _____

Second Dexa scan: Wrist result Unknown Abnormal Normal

Second Dexa scan: Wrist Zscore _____

Second Dexa scan: Total body result Unknown Abnormal Normal

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

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

Patient Name _____

Date ***bKT - Visit Management And Treatment Pharmacotherapy*****Pharmacotherapy**

Medications

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

Other medications- specify _____

Homeopathic therapies Unknown Yes No

Specify homeopathic therapies

Disease Treatment

Treatment recommended/prescribed

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

Glucose paste/gel/tablets

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

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

L-carnitineL-carnitine route recommended/prescribed Unknown Feeding tube IV Oral

L-carnitine dose recommended/prescribed _____

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

L-carnitine dose recommended/prescribed units g mg

L-carnitine frequency recommended/prescribed

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

L-carnitine frequency recommended/prescribed-other, specify _____

L-carnitine taken as recommended/prescribed Unknown Yes No

Actual L-carnitine dose reported _____

Actual L-carnitine dose reported units g mg

Actual L-carnitine frequency reported

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

Actual L-carnitine frequency reported-other, specify _____

Reason L-carnitine is not taken as recommended/prescribed _____

Method of payment for L-carnitine

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

Other Treatment

Other treatment recommend/prescribed _____

Other treatment route recommended/prescribed _____

Other treatment dose recommended/prescribed _____

Other treatment dose recommended/prescribed units _____

Other treatment frequency recommended/prescribed _____

Other treatment taken as recommended/prescribed Unknown Yes No

Actual other treatment dose reported _____

Actual other treatment dose reported units _____

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

Actual other treatment frequency reported _____

Reason other treatment is not taken as recommended/prescribed _____

Method of payment for other treatment

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

Comments

Medication and supplement comments

--

Patient Name _____

Date ***bKT - Visit Management And Treatment Nutrition*****Nutrition**Mode of nutrition delivery Unknown Oral NG tube NJ tube G-tube GJ tube TPN

Types of milk/formula taken

- | | | |
|--|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> None | <input type="checkbox"/> Baby formula (regular) |
| <input type="checkbox"/> Baby formula (soy) | <input type="checkbox"/> Elemental formula | <input type="checkbox"/> Breast milk |
| <input type="checkbox"/> Human milk fortifier | <input type="checkbox"/> Almond milk | <input type="checkbox"/> Rice milk |
| <input type="checkbox"/> Skim milk | <input type="checkbox"/> 1% milk | <input type="checkbox"/> 2% milk |
| <input type="checkbox"/> Soy milk | <input type="checkbox"/> Special metabolic formula | <input type="checkbox"/> Toddler formula (regular) |
| <input type="checkbox"/> Toddler formula (soy) | <input type="checkbox"/> Whole milk | <input type="checkbox"/> Other |

Number of special metabolic formulas recommended/prescribed Unknown 1 2 3

Name of special metabolic formula 1 _____

Amount of special metabolic formula 1 (grams) recommended/prescribed per 24 hours _____

Special metabolic formula 1 taken as recommended/prescribed Unknown Yes No

Actual frequency of use of special metabolic formula 1

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

Reason special metabolic formula 1 is not taken as recommended/prescribed _____

Fat grams from metabolic formula 1 recommended/prescribed per 24 hours _____

Protein grams from metabolic formula 1 recommended/prescribed per 24 hours _____

Method of payment for special metabolic formula 1

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

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

Name of special metabolic formula 2 _____

Amount of special metabolic formula 2 (grams) recommended/prescribed per 24 hours _____

Special metabolic formula 2 taken as recommended/prescribed Unknown Yes No

Actual frequency of use of special metabolic formula 2

Unknown 0 days/week 1 day/week 2 days/week 3 days/week 4 days/week

5 days/week 6 days/week

Reason special metabolic formula 2 is not taken as recommended/prescribed _____

Fat grams from metabolic formula 2 recommended/prescribed per 24 hours _____

Protein grams from metabolic formula 2 recommended/prescribed per 24 hours _____

Method of payment for special metabolic formula 2

Unknown

None

Commercial/private

Medicaid

Medicare

Military

Newborn screening funds

Patient assistance program

Self-pay

State Children's Health Insurance Program (SCHIP)

State Children with Special Health Needs (CSHN) Program

Other

Name of special metabolic formula 3 _____

Amount of special metabolic formula 3 (grams) recommended/prescribed per 24 hours _____

Special metabolic formula 3 taken as recommended/prescribed Unknown Yes No

Actual frequency of use of special metabolic formula 3

Unknown 0 days/week 1 day/week 2 days/week 3 days/week 4 days/week

5 days/week 6 days/week

Reason special metabolic formula 3 is not taken as recommended/prescribed _____

Fat grams from metabolic formula 3 recommended/prescribed per 24 hours _____

Protein grams from metabolic formula 3 recommended/prescribed per 24 hours _____

Method of payment for special metabolic formula 3

Unknown

None

Commercial/private

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

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

Type milk/formula, other- specify _____

Modified Low Protein Foods

Modified low protein foods recommended/prescribed Unknown Yes No

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

Method of payment for modified low protein foods

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

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

Actual frequency of adherence to modified low protein foods

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

Reason for poor adherence to modified low protein foods _____

Feeding Intervals

Specific information given to avoid fasting Unknown Yes No

Longest interval recommended between feedings in well state during DAY (in hours)

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

- Unknown 1 2 3 4 5 6
 7 8 9 10 11 12

Longest reported interval between feedings in well state during DAY (in hours)

- Unknown 1 2 3 4 5 6
 7 8 9 10 11 12

Longest interval recommended between feedings in well state during NIGHT (in hours)

- Unknown 1 2 3 4 5 6
 7 8 9 10 11 12

Longest reported interval between feedings in well state during NIGHT (in hours)

- Unknown 1 2 3 4 5 6
 7 8 9 10 11 12

Comments

Nutrition comments

Patient Name _____

Date ***bKT - Study Status***First date of study status change Condition follow-up status Active Inactive

Reason for inactive status

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

Date of death

Age of death (in years) _____

Moved-specify

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

Patient Name _____

Date

Specify other non-IBEMC participating center _____

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

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

Specify reason subject initiated withdrawal, if known

Specify reason investigator initiated withdrawal, if known

Second date of study status change

Condition follow-up status Active Inactive

Reason for inactive status

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

Date of death

Age of death (in years) _____

Moved-specify

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

Specify other IBEMC participating center

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

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

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

Specify other non-IBEMC participating center _____

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

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

Patient Name _____

Date

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

Specify other non-IBEMC participating center _____

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

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

Specify reason subject initiated withdrawal, if known

Specify reason investigator initiated withdrawal, if known

Fourth date of study status change

Condition follow-up status Active Inactive

Reason for inactive status

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

Date of death

Age of death (in years) _____

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

Moved-specify

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

Specify other IBEMC participating center

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

Specify other non-IBEMC participating center _____

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

Patient Name _____

Date

Declined consent at age of majority or emancipation

Subject initiated withdrawal

Investigator initiated withdrawal

Specify reason subject initiated withdrawal, if known

Specify reason investigator initiated withdrawal, if known

Comments

Study status comments

Patient Name _____

Date ***bKT - Pregnancy***Patient has had one or more pregnancies Unknown Yes No

Number of pregnancies _____

Number of term pregnancies _____

Number of preterm pregnancies _____

Number of pregnancies ending in abortion/miscarriage _____

Number of pregnancies resulting in live birth _____

Patient has biological children Unknown Yes No

Number of biological children _____

First Pregnancy

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

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

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

History of infertility for first pregnancy

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

Cause(s) of infertility for first pregnancy

- Unknown Male factor Female factor
 Combined male-female factor

Cause(s) of male infertility for first pregnancy

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

Cause(s) of male infertility for first pregnancy-other, specify _____

Cause(s) of female infertility for first pregnancy

- Unknown Age
 Alcohol use Cancer/cancer treatment related
 Early menopause Fallopian tube damage/blockage

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

- | | |
|---|--|
| <input type="checkbox"/> Hyperprolactinemia | <input type="checkbox"/> Pelvic adhesions |
| <input type="checkbox"/> Polycystic ovary syndrome (PCOS) | <input type="checkbox"/> Premature ovarian insufficiency |
| <input type="checkbox"/> Over-exercise | <input type="checkbox"/> Ovulation disorder |
| <input type="checkbox"/> Thyroid problems | <input type="checkbox"/> Tobacco use |
| <input type="checkbox"/> Uterine fibroid | <input type="checkbox"/> Weight related |
| <input type="checkbox"/> Other | |

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

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

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

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

Type of assisted reproductive technologies used during first pregnancy

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

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

Partner tested for patient's disorder during first pregnancy

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

Partner also affected by disorder during first pregnancy Unknown Yes No

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

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

Type of prenatal testing performed for first pregnancy Unknown Biochemical Molecular

Method of prenatal testing for first pregnancy

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

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

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

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

First pregnancy terminated Unknown Yes No

Reason first pregnancy terminated

- Elective due to fetus affected with disorder

Patient Name _____

Date

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

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

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for first pregnancy _____

Prenatal care received during first pregnancy Unknown Yes No

Weeks gestation prenatal care started during first pregnancy _____

Treatment prescribed for patient's disorder during first pregnancy

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

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

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

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

Patient in good metabolic condition during second trimester of first pregnancy

- Unknown Pregnancy not sustained to second trimester
- Yes No

Patient in good metabolic condition during third trimester of first pregnancy

- Unknown Pregnancy not sustained to third trimester
- Yes No

Number of outpatient metabolic visits for patient during first pregnancy _____

Number of ED visits for management of disorder during first pregnancy

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

Number of hospitalizations for management of disorder during first pregnancy

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

Complications during first pregnancy

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

Patient Name _____

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

Complications related to disorder during first pregnancy- specify

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

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

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

Complications during first pregnancy-other, specify_____

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

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

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

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

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

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

Patient Name _____

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

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

Laboratory studies done on patient during first pregnancy

- | | |
|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Ammonia - Abn high |
| <input type="checkbox"/> Ammonia - WNL | <input type="checkbox"/> Blood glucose - Abn low |
| <input type="checkbox"/> Blood glucose - WNL | <input type="checkbox"/> Blood Glucose - Abn high |
| <input type="checkbox"/> BNP - Abn high | <input type="checkbox"/> BNP - WNL |
| <input type="checkbox"/> CBC - Abn | <input type="checkbox"/> CBC - WNL |
| <input type="checkbox"/> C02 - Abn low | <input type="checkbox"/> C02 - WNL |
| <input type="checkbox"/> CK - Abn high | <input type="checkbox"/> CK - WNL |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn | <input type="checkbox"/> INR - WNL |
| <input type="checkbox"/> Liver function tests - Abn high | <input type="checkbox"/> Liver function tests - WNL |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn | <input type="checkbox"/> Plasma acylcarnitine profile - WNL |
| <input type="checkbox"/> Plasma amino acids - Abn | <input type="checkbox"/> Plasma amino acids - WNL |
| <input type="checkbox"/> Plasma carnitine levels - Abn | <input type="checkbox"/> Plasma carnitine levels - WNL |
| <input type="checkbox"/> Prealbumin - Abn low | <input type="checkbox"/> Prealbumin - WNL |
| <input type="checkbox"/> Transferrin - Abn | <input type="checkbox"/> Transferrin - WNL |
| <input type="checkbox"/> Urine acylcarnitines - Abn | <input type="checkbox"/> Urine acylcarnitines - WNL |
| <input type="checkbox"/> Urine acylglycines - Abn | <input type="checkbox"/> Urine acylglycines - WNL |
| <input type="checkbox"/> Urine ketones - Abn high | <input type="checkbox"/> Urine ketones - WNL |
| <input type="checkbox"/> Urine organic acids - Abn | <input type="checkbox"/> Urine organic acids - WNL |
| <input type="checkbox"/> Other | |

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

Additional interventions required during first pregnancy due to this metabolic condition

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

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

- More intensive fetal monitoring
- Bedrest
- Other

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

Total maternal weight gain (in kg) during first pregnancy _____

Number of documented episodes of hyperammonemia during first pregnancy

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

Number of documented episodes of ketonuria during first pregnancy

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

Number of documented episodes of metabolic acidosis during first pregnancy

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

Treatment(s) for hyperammonemic episodes during first pregnancy

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

Maternal protein intake restricted during first pregnancy Unknown Yes No

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

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

Ammonia scavenger drugs used during first pregnancy

- Unknown No scavengers used Buphenyl Sodium benzoate

Other medications/supplements used during first pregnancy

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

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

Patient Name _____

Date

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

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

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

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

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

Abnormal patient lab results during first delivery

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

Abnormal patient lab results during first delivery-other, specify

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

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

Patient Name _____

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

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

[Empty text box for maternal interventions]

Patient death during or shortly after first delivery Unknown Yes No

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

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

Method of first delivery

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

Maternal inpatient days post- first delivery_____

Live delivery of first newborn Unknown Yes No

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

[Empty text box for acute health concerns]

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

[Empty text box for lab tests and results]

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

[Empty text box for health concerns]

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

- 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

[Empty text box for additional interventions]

Birth measurements for first newborn Unknown Head circumference Length Weight

Patient Name _____

Date

Birth head circumference _____

Birth head circumference units cm in

Birth length _____

Birth length units cm in

Birth weight _____

Birth weight units lbs kg gm oz

APGAR score at 1 minute for first newborn

Unknown 0 1 2 3 4 5

6 7 8 9 10

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

APGAR score at 5 minutes for first newborn

Unknown 0 1 2 3 4 5

6 7 8 9 10

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

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

Unknown <24 hours

24-28 hours 3-5 days

6-14 days >14 days

N/A - baby was not born in a hospital baby is still in the hospital

Routine newborn screening done for first newborn

Unknown Yes - normal for all screened disorders

Yes -abnormal Yes - results pending

No

Describe abnormal newborn screen result for first newborn

Reason routine newborn screening was not done for first newborn

Unknown

Patient Name _____

Date

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

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

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

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

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

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

- Unknown Yes No

Abnormal newborn exam findings for first newborn

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

Abnormal newborn exam findings for first newborn-other, specify _____

Newborn death at or shortly after delivery for first newborn Unknown Yes NoNewborn's death related to maternal disorder for first newborn Unknown Yes NoFirst newborn currently alive Unknown Yes No

Second Pregnancy

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

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

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

Patient Name _____

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

History of infertility for second pregnancy

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

Cause(s) of infertility for second pregnancy

- Unknown Male factor Female factor
 Combined male-female factor

Cause(s) of male infertility for second pregnancy

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

Cause(s) of male infertility for second pregnancy-other, specify _____

Cause(s) of female infertility for second pregnancy

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

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

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

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

- Unknown None
 Assisted reproductive technology Fertility drugs
 Surgery

Type of assisted reproductive technologies used during second pregnancy

- Assisted hatching ICSI IVF
 Surgical sperm aspiration Other

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

Patient Name _____

Date

Partner tested for patient's disorder during second pregnancy

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

Partner also affected by disorder during second pregnancy Unknown Yes No

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

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

Type of prenatal testing performed for second pregnancy Unknown Biochemical Molecular

Method of prenatal testing for second pregnancy

- Unknown
- Amniocentesis
- Chorionic villus sampling

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

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

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

Second pregnancy terminated Unknown Yes No

Reason second pregnancy terminated

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

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

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

Prenatal care received during second pregnancy Unknown Yes No

Weeks gestation prenatal care started during second pregnancy _____

Treatment prescribed for patient's disorder during second pregnancy

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

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

Patient Name _____

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

Patient in good metabolic condition prior to second pregnancy Unknown Yes NoPatient in good metabolic condition during first trimester of second pregnancy Unknown Yes No

Patient in good metabolic condition during second trimester of second pregnancy

UnknownPregnancy not sustained to second trimesterYesNo

Patient in good metabolic condition during third trimester of second pregnancy

UnknownPregnancy not sustained to third trimesterYesNo

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 56 7 8 9 10 >10

Number of hospitalizations for management of disorder during second pregnancy

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

Complications during second pregnancy

UnknownYes-related to disorderYes- not known to be related to disorder No

Complications related to disorder during second pregnancy- specify

UnknownAcute fatty liver of pregnancy (AFLP)Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)Intrauterine growth restriction (AUGR)Mother affected with this conditionOther

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

UnknownAdvanced maternal age (35+ years of age)Ectopic pregnancyGestational diabetesGroup B strepHemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)Intrauterine growth restriction (AUGR)Inadequate prenatal careMaternal prenatal substance exposure

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

- Preeclampsia
- Rh isoimmunization
- Toxemia
- Young maternal age (15 years of age + under)
- Preterm labor
- Other

Complications during second pregnancy-other, specify _____

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

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

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

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

Duration of longest inpatient hospitalization (in days) for any reason during second pregnancy _____

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

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

Laboratory studies done on patient during second pregnancy

- | | |
|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Ammonia - Abn high |
| <input type="checkbox"/> Ammonia - WNL | <input type="checkbox"/> Blood glucose - Abn low |
| <input type="checkbox"/> Blood glucose - WNL | <input type="checkbox"/> Blood Glucose - Abn high |
| <input type="checkbox"/> BNP - Abn high | <input type="checkbox"/> BNP - WNL |
| <input type="checkbox"/> CBC - Abn | <input type="checkbox"/> CBC - WNL |
| <input type="checkbox"/> CO2 - Abn low | <input type="checkbox"/> CO2 - WNL |
| <input type="checkbox"/> CK - Abn high | <input type="checkbox"/> CK - WNL |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn | <input type="checkbox"/> INR - WNL |
| <input type="checkbox"/> Liver function tests - Abn high | <input type="checkbox"/> Liver function tests - WNL |

Patient Name _____

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

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

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

Additional interventions required during second pregnancy due to this metabolic condition

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

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

Total maternal weight gain (in kg) during second pregnancy _____

Number of documented episodes of hyperammonemia during second pregnancy

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

Number of documented episodes of ketonuria during second pregnancy

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

Number of documented episodes of metabolic acidosis during second pregnancy

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

Treatment(s) for hyperammonemic episodes during second pregnancy

- Unknown
- No hyperammonemic episodes
- Hospital admission

Patient Name _____

Date

- Protein restriction
- Ammonul
- Arginine

Maternal protein intake restricted during second pregnancy Unknown Yes No

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

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

Ammonia scavenger drugs used during second pregnancy

- Unknown
- No scavengers used
- Buphenyl
- Sodium benzoate

Other medications/supplements used during second pregnancy

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

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

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

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

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

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

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

Patient Name _____

Date

Abnormal patient lab results during second delivery

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

Abnormal patient lab results during second delivery-other, specify

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

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

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

Patient death during or shortly after second delivery Unknown Yes No

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

Weeks gestation at time of second delivery - round to nearest week _____

Method of second delivery

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

Maternal inpatient days post- second delivery _____

Live delivery of second newborn Unknown Yes No

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

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

Patient Name _____

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

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

[Empty text box for health concerns]

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

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

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

[Empty text box for additional interventions]

Birth measurements for second newborn Unknown Head circumference Length Weight

Birth head circumference _____

Birth head circumference units cm in

Birth length _____

Birth length units cm in

Birth weight _____

Birth weight units lbs kg gm oz

APGAR score at 1 minute for second newborn

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

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

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

APGAR score at 5 minutes for second newborn

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

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

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

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

Patient Name _____

Date

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

Routine newborn screening done for second newborn

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

Describe abnormal newborn screen result for second newborn

Reason routine newborn screening was not done for second newborn

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

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

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

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

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

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

- Unknown
- Yes
- No

Patient Name _____

Date

Abnormal newborn exam findings for second newborn

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

Abnormal newborn exam findings for second newborn-other, specify _____

Newborn death at or shortly after delivery for second newborn Unknown Yes NoNewborn's death related to maternal disorder for second newborn Unknown Yes NoSecond newborn currently alive Unknown Yes No**Third Pregnancy**

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

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

- | | | | | | | |
|-------------------------------|---------------------------------|---------------------------|-------------------------|--------------------------|--------------------------|--------------------------|
| <input type="radio"/> Unknown | <input type="radio"/> Unplanned | <input type="radio"/> 1 | <input type="radio"/> 2 | <input type="radio"/> 3 | <input type="radio"/> 4 | <input type="radio"/> 5 |
| <input type="radio"/> 6 | <input type="radio"/> 7 | <input type="radio"/> 8 | <input type="radio"/> 9 | <input type="radio"/> 10 | <input type="radio"/> 11 | <input type="radio"/> 12 |
| <input type="radio"/> 13-24 | <input type="radio"/> 25-36 | <input type="radio"/> >36 | | | | |

History of infertility for third pregnancy

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

Cause(s) of infertility for third pregnancy

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

Cause(s) of male infertility for third pregnancy

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

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

Cause(s) of female infertility for third pregnancy

- | | |
|---|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Age |
| <input type="checkbox"/> Alcohol use | <input type="checkbox"/> Cancer/cancer treatment related |
| <input type="checkbox"/> Early menopause | <input type="checkbox"/> Fallopian tube damage/blockage |
| <input type="checkbox"/> Hyperprolactinemia | <input type="checkbox"/> Pelvic adhesions |
| <input type="checkbox"/> Polycystic ovary syndrome (PCOS) | <input type="checkbox"/> Premature ovarian insufficiency |

Patient Name _____

Date

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

Cause(s) of female infertility for third pregnancy-other, specify _____

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

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

- Unknown None
 Assisted reproductive technology Fertility drugs
 Surgery

Type of assisted reproductive technologies used during third pregnancy

- Assisted hatching ICSI IVF
 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 NoPreimplantation genetic diagnosis for disorder done for third pregnancy Unknown Yes NoPrenatal testing done for fetus for this disorder for third pregnancy Unknown Yes NoType of prenatal testing performed for third pregnancy Unknown Biochemical Molecular

Method of prenatal testing for third pregnancy

- Unknown Amniocentesis Chorionic villus sampling

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

- Unknown None
 Fetal echocardiogram Increased frequency prenatal ultrasounds
 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

Patient Name _____

Date Elective due to maternal well-being Spontaneous

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

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for third pregnancy _____

Prenatal care received during third pregnancy Unknown Yes No

Weeks gestation prenatal care started during third pregnancy _____

Treatment prescribed for patient's disorder during third pregnancy

 Unknown Metabolic diet Medications Biochemical lab monitoring Avoidance of fasting Other

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

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

Patient in good metabolic condition during second trimester of third pregnancy

 Unknown Pregnancy not sustained to second trimester Yes No

Patient in good metabolic condition during third trimester of third pregnancy

 Unknown Pregnancy not sustained to third trimester Yes No

Number of outpatient metabolic visits for patient during third pregnancy _____

Number of ED visits for management of disorder during third pregnancy

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

Number of hospitalizations for management of disorder during third pregnancy

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

Complications during third pregnancy

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

Complications related to disorder during third pregnancy- specify

 Unknown

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

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

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

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

Complications during third pregnancy-other, specify _____

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

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

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

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

Duration of longest inpatient hospitalization (in days) for any reason during third pregnancy _____

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

Patient Name _____

Date |||||

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

Laboratory studies done on patient during third pregnancy

- | | |
|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Ammonia - Abn high |
| <input type="checkbox"/> Ammonia - WNL | <input type="checkbox"/> Blood glucose - Abn low |
| <input type="checkbox"/> Blood glucose - WNL | <input type="checkbox"/> Blood Glucose - Abn high |
| <input type="checkbox"/> BNP - Abn high | <input type="checkbox"/> BNP - WNL |
| <input type="checkbox"/> CBC - Abn | <input type="checkbox"/> CBC - WNL |
| <input type="checkbox"/> C02 - Abn low | <input type="checkbox"/> C02 - WNL |
| <input type="checkbox"/> CK - Abn high | <input type="checkbox"/> CK - WNL |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn | <input type="checkbox"/> INR - WNL |
| <input type="checkbox"/> Liver function tests - Abn high | <input type="checkbox"/> Liver function tests - WNL |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn | <input type="checkbox"/> Plasma acylcarnitine profile - WNL |
| <input type="checkbox"/> Plasma amino acids - Abn | <input type="checkbox"/> Plasma amino acids - WNL |
| <input type="checkbox"/> Plasma carnitine levels - Abn | <input type="checkbox"/> Plasma carnitine levels - WNL |
| <input type="checkbox"/> Prealbumin - Abn low | <input type="checkbox"/> Prealbumin - WNL |
| <input type="checkbox"/> Transferrin - Abn | <input type="checkbox"/> Transferrin - WNL |
| <input type="checkbox"/> Urine acylcarnitines - Abn | <input type="checkbox"/> Urine acylcarnitines - WNL |
| <input type="checkbox"/> Urine acylglycines - Abn | <input type="checkbox"/> Urine acylglycines - WNL |
| <input type="checkbox"/> Urine ketones - Abn high | <input type="checkbox"/> Urine ketones - WNL |
| <input type="checkbox"/> Urine organic acids - Abn | <input type="checkbox"/> Urine organic acids - WNL |
| <input type="checkbox"/> Other | |

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

Additional interventions required during third pregnancy due to this metabolic condition

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

Patient Name _____

Date

- More intensive fetal monitoring Bedrest
 Other

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

Total maternal weight gain (in kg) during third pregnancy _____

Number of documented episodes of hyperammonemia during third pregnancy

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

Number of documented episodes of ketonuria during third pregnancy

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

Number of documented episodes of metabolic acidosis during third pregnancy

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

Treatment(s) for hyperammonemic episodes during third pregnancy

- Unknown No hyperammonemic episodes Hospital admission
 Protein restriction Ammonul Arginine

Maternal protein intake restricted during third pregnancy Unknown Yes No

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

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

Ammonia scavenger drugs used during third pregnancy

- Unknown No scavengers used Buphenyl Sodium benzoate

Other medications/supplements used during third pregnancy

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

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

Patient Name _____

Date

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

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

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

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

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

Abnormal patient lab results during third delivery

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

Abnormal patient lab results during third delivery-other, specify

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

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

Patient Name _____

Date

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

Patient death during or shortly after third delivery Unknown Yes No

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

Weeks gestation at time of third delivery - round to nearest week _____

Method of third delivery

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

Maternal inpatient days post- third delivery _____

Live delivery of third newborn Unknown Yes No

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

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

Birth measurements for third newborn Unknown Head circumference Length Weight

Birth head circumference _____

Birth head circumference units cm in

Birth length _____

Birth length units cm in

Birth weight _____

Birth weight units lbs kg gm oz

APGAR score at 1 minute for third newborn

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

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

Patient Name _____

Date

APGAR score at 5 minutes for third newborn

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

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

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

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

Routine newborn screening done for third newborn

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

Describe abnormal newborn screen result for third newborn

Reason routine newborn screening was not done for third newborn

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

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

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

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

Patient Name _____

Date

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

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

Unknown Yes No

Abnormal newborn exam findings for third newborn

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

Abnormal newborn exam findings for third newborn-other, specify _____

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

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

Third newborn currently alive Unknown Yes No

Fourth Pregnancy

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

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

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

History of infertility for fourth pregnancy

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

Cause(s) of infertility for fourth pregnancy

Unknown Male factor Female factor
 Combined male-female factor

Cause(s) of male infertility for fourth pregnancy

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

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

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

Cause(s) of female infertility for fourth pregnancy

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

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

Type of assisted reproductive technologies used during fourth pregnancy

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

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

Patient Name _____

Date

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

Fourth pregnancy terminated Unknown Yes No

Reason fourth pregnancy terminated

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

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

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for fourth pregnancy _____

Prenatal care received during fourth pregnancy Unknown Yes No

Weeks gestation prenatal care started during fourth pregnancy _____

Treatment prescribed for patient's disorder during fourth pregnancy

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

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

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

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

Patient in good metabolic condition during second trimester of fourth pregnancy

- Unknown Pregnancy not sustained to second trimester
- Yes No

Patient in good metabolic condition during third trimester of fourth pregnancy

- Unknown Pregnancy not sustained to third trimester
- Yes No

Number of outpatient metabolic visits for patient during fourth pregnancy _____

Number of ED visits for management of disorder during fourth pregnancy

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

Number of hospitalizations for management of disorder during fourth pregnancy

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

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

Complications during fourth pregnancy

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

Complications related to disorder during fourth pregnancy- specify

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

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

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

Complications during fourth pregnancy-other, specify_____

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

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

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

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

Patient Name _____

Date | |

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

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

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

Laboratory studies done on patient during fourth pregnancy

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

Patient Name _____

Date

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

Additional interventions required during fourth pregnancy due to this metabolic condition

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

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

Total maternal weight gain (in kg) during fourth pregnancy_____

Number of documented episodes of hyperammonemia during fourth pregnancy

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

Number of documented episodes of ketonuria during fourth pregnancy

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

Number of documented episodes of metabolic acidosis during fourth pregnancy

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

Treatment(s) for hyperammonemic episodes during fourth pregnancy

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

Maternal protein intake restricted during fourth pregnancy Unknown Yes No

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

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

Ammonia scavenger drugs used during fourth pregnancy

- Unknown No scavengers used Buphenyl Sodium benzoate

Other medications/supplements used during fourth pregnancy

Patient Name _____

Date

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

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

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

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

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

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

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

Abnormal patient lab results during fourth delivery

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

Abnormal patient lab results during fourth delivery-other, specify

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

- Unknown Ammonul Blood/blood product transfusion

Patient Name _____

Date

- Dialysis
- ICU monitoring
- Infusions
- Medications
- Resuscitation
- TPN
- Other

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

Patient death during or shortly after fourth delivery Unknown Yes No

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

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

Method of fourth delivery

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

Maternal inpatient days post- fourth delivery_____

Live delivery of fourth newborn Unknown Yes No

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

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

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

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

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

Patient Name _____

Date

Describe abnormal newborn screen result for fourth newborn

Reason routine newborn screening was not done for fourth newborn

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

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

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

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

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

Patient Name _____

Date Fourth newborn currently alive Unknown Yes No**Fifth Pregnancy**

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

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

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

History of infertility for fifth pregnancy

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

Cause(s) of infertility for fifth pregnancy

- Unknown Male factor Female factor
 Combined male-female factor

Cause(s) of male infertility for fifth pregnancy

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

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

Cause(s) of female infertility for fifth pregnancy

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

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

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

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

Patient Name _____

Date

- Unknown None
 Assisted reproductive technology Fertility drugs
 Surgery

Type of assisted reproductive technologies used during fifth pregnancy

- Assisted hatching ICSI IVF
 Surgical sperm aspiration Other

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

Partner tested for patient's disorder during fifth pregnancy

- Unknown Yes-biochemical Yes-molecular No

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

Method of prenatal testing for fifth pregnancy

- Unknown Amniocentesis Chorionic villus sampling

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

- Unknown None
 Fetal echocardiogram Increased frequency prenatal ultrasounds
 Other

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

Fifth pregnancy terminated Unknown Yes No

Reason fifth pregnancy terminated

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

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

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for fifth pregnancy _____

Prenatal care received during fifth pregnancy Unknown Yes No

Weeks gestation prenatal care started during fifth pregnancy _____

Patient Name _____

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

Treatment prescribed for patient's disorder during fifth pregnancy

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

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

Patient in good metabolic condition prior to fifth pregnancy Unknown Yes NoPatient in good metabolic condition during first trimester of fifth pregnancy Unknown Yes No

Patient in good metabolic condition during second trimester of fifth pregnancy

- Unknown Pregnancy not sustained to second trimester
 Yes No

Patient in good metabolic condition during third trimester of fifth pregnancy

- Unknown Pregnancy not sustained to third trimester
 Yes No

Number of outpatient metabolic visits for patient during fifth pregnancy _____

Number of ED visits for management of disorder during fifth pregnancy

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

Number of hospitalizations for management of disorder during fifth pregnancy

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

Complications during fifth pregnancy

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

Complications related to disorder during fifth pregnancy- specify

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

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

- Unknown
 Advanced maternal age (35+ years of age)

Patient Name _____

Date

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

Complications during fifth pregnancy-other, specify _____

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

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

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

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

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

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

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

Laboratory studies done on patient during fifth pregnancy

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

Patient Name _____

Date

- BNP - Abn high
- BNP - WNL
- CBC - Abn
- CBC - WNL
- CO2 - Abn low
- CO2 - 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 acylglycines - WNL
- Urine ketones - Abn high
- Urine ketones - WNL
- Urine organic acids - Abn
- Urine organic acids - WNL
- Other

Other laboratory studies done on patient during 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_____

Number of documented episodes of hyperammonemia during fifth pregnancy

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

Number of documented episodes of ketonuria during fifth pregnancy

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

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

Number of documented episodes of metabolic acidosis during fifth pregnancy

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

Treatment(s) for hyperammonemic episodes during fifth pregnancy

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

Maternal protein intake restricted during fifth pregnancy Unknown Yes No

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

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

Ammonia scavenger drugs used during fifth pregnancy

- Unknown No scavengers used Buphenyl Sodium benzoate

Other medications/supplements used during fifth pregnancy

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

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

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

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

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

Patient Name _____

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

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

[Empty text box for interventions]

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

[Empty text box for health concerns]

Abnormal patient lab results during fifth delivery

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

Abnormal patient lab results during fifth delivery-other, specify

[Empty text box for other lab results]

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

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

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

[Empty text box for other maternal interventions]

Patient death during or shortly after fifth delivery Unknown Yes No

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

Weeks gestation at time of fifth delivery - round to nearest week _____

Method of fifth delivery

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

Maternal inpatient days post- fifth delivery _____

Live delivery of fifth newborn Unknown Yes No

Patient Name _____

Date

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

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

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

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

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

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

Birth measurements for fifth newborn Unknown Head circumference Length Weight

Birth head circumference _____

Birth head circumference units cm in

Birth length _____

Birth length units cm in

Birth weight _____

Birth weight units lbs kg gm oz

APGAR score at 1 minute for fifth newborn

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

Patient Name _____

Date

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

APGAR score at 5 minutes for fifth newborn

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

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

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

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

Routine newborn screening done for fifth newborn

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

Describe abnormal newborn screen result for fifth newborn

Reason routine newborn screening was not done for fifth newborn

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

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

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

Patient Name _____

Date

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

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

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

- Unknown Yes No

Abnormal newborn exam findings for fifth newborn

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

Abnormal newborn exam findings for fifth newborn-other, specify _____

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

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

Fifth newborn currently alive Unknown Yes No

Sixth Pregnancy

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

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

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

History of infertility for sixth pregnancy

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

Cause(s) of infertility for sixth pregnancy

- Unknown Male factor Female factor
 Combined male-female factor

Cause(s) of male infertility for sixth pregnancy

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

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

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

Cause(s) of female infertility for sixth pregnancy

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

Cause(s) of female infertility for sixth pregnancy-other, specify _____

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

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

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

Type of assisted reproductive technologies used during sixth pregnancy

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

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

Partner tested for patient's disorder during sixth pregnancy

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

Partner also affected by disorder during sixth pregnancy Unknown Yes 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

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

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

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

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

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

Sixth pregnancy terminated Unknown Yes No

Reason sixth pregnancy terminated

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

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

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for sixth pregnancy _____

Prenatal care received during sixth pregnancy Unknown Yes No

Weeks gestation prenatal care started during sixth pregnancy _____

Treatment prescribed for patient's disorder during sixth pregnancy

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

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

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

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

Patient in good metabolic condition during second trimester of sixth pregnancy

- Unknown Pregnancy not sustained to second trimester
- Yes No

Patient in good metabolic condition during third trimester of sixth pregnancy

- Unknown Pregnancy not sustained to third trimester
- Yes No

Number of outpatient metabolic visits for patient during sixth pregnancy _____

Patient Name _____

Date

Number of ED visits for management of disorder during sixth pregnancy

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

Number of hospitalizations for management of disorder during sixth pregnancy

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

Complications during sixth pregnancy

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

Complications related to disorder during sixth pregnancy- specify

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

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

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

Complications during sixth pregnancy-other, specify _____

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

- Unknown 0 1 2 3 4 5

Patient Name _____

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

6 7 8 9 10 >10

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

Unknown 0 1 2 3 4 5

6 7 8 9 10 >10

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

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

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

Laboratory studies done on patient during sixth pregnancy

- | | |
|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Ammonia - Abn high |
| <input type="checkbox"/> Ammonia - WNL | <input type="checkbox"/> Blood glucose - Abn low |
| <input type="checkbox"/> Blood glucose - WNL | <input type="checkbox"/> Blood Glucose - Abn high |
| <input type="checkbox"/> BNP - Abn high | <input type="checkbox"/> BNP - WNL |
| <input type="checkbox"/> CBC - Abn | <input type="checkbox"/> CBC - WNL |
| <input type="checkbox"/> CO2 - Abn low | <input type="checkbox"/> CO2 - WNL |
| <input type="checkbox"/> CK - Abn high | <input type="checkbox"/> CK - WNL |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn | <input type="checkbox"/> INR - WNL |
| <input type="checkbox"/> Liver function tests - Abn high | <input type="checkbox"/> Liver function tests - WNL |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn | <input type="checkbox"/> Plasma acylcarnitine profile - WNL |
| <input type="checkbox"/> Plasma amino acids - Abn | <input type="checkbox"/> Plasma amino acids - WNL |
| <input type="checkbox"/> Plasma carnitine levels - Abn | <input type="checkbox"/> Plasma carnitine levels - WNL |
| <input type="checkbox"/> Prealbumin - Abn low | <input type="checkbox"/> Prealbumin - WNL |
| <input type="checkbox"/> Transferrin - Abn | <input type="checkbox"/> Transferrin - WNL |
| <input type="checkbox"/> Urine acylcarnitines - Abn | <input type="checkbox"/> Urine acylcarnitines - WNL |
| <input type="checkbox"/> Urine acylglycines - Abn | <input type="checkbox"/> Urine acylglycines - WNL |
| <input type="checkbox"/> Urine ketones - Abn high | <input type="checkbox"/> Urine ketones - WNL |
| <input type="checkbox"/> Urine organic acids - Abn | <input type="checkbox"/> Urine organic acids - WNL |

Patient Name _____

Date

Other

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

Additional interventions required during sixth pregnancy due to this metabolic condition

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

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

Total maternal weight gain (in kg) during sixth pregnancy _____

Number of documented episodes of hyperammonemia during sixth pregnancy

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

Number of documented episodes of ketonuria during sixth pregnancy

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

Number of documented episodes of metabolic acidosis during sixth pregnancy

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

Treatment(s) for hyperammonemic episodes during sixth pregnancy

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

Maternal protein intake restricted during sixth pregnancy Unknown Yes No

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

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

Ammonia scavenger drugs used during sixth pregnancy

- Unknown No scavengers used Buphenyl Sodium benzoate

Patient Name _____

Date

Other medications/supplements used during sixth pregnancy

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

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

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

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

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

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

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

Abnormal patient lab results during sixth delivery

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

Patient Name _____

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

Abnormal patient lab results during sixth delivery-other, specify

[Empty text box for abnormal patient lab results]

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

[Empty text box for additional maternal interventions]

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)

[Empty text box for acute health concerns]

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

[Empty text box for additional lab tests]

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

[Empty text box for health concerns]

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

Patient Name _____

Date

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

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

Birth measurements for sixth newborn Unknown Head circumference Length Weight

Birth head circumference _____

Birth head circumference units cm in

Birth length _____

Birth length units cm in

Birth weight _____

Birth weight units lbs kg gm oz

APGAR score at 1 minute for sixth newborn

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

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

APGAR score at 5 minutes for sixth newborn

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

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

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

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

Routine newborn screening done for sixth newborn

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

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

Describe abnormal newborn screen result for sixth newborn

Reason routine newborn screening was not done for sixth newborn

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

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

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

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

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

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

- Unknown
- Yes
- No

Abnormal newborn exam findings for sixth newborn

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

Abnormal newborn exam findings for sixth newborn-other, specify _____

Patient Name _____

Date Newborn death at or shortly after delivery for sixth newborn Unknown Yes NoNewborn's death related to maternal disorder for sixth newborn Unknown Yes NoSixth newborn currently alive Unknown Yes No**Seventh Pregnancy**

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

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

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

History of infertility for seventh pregnancy

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

Cause(s) of infertility for seventh pregnancy

- Unknown Male factor Female factor
 Combined male-female factor

Cause(s) of male infertility for seventh pregnancy

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

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

Cause(s) of female infertility for seventh pregnancy

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

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

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

Patient Name _____

Date

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

- Unknown None
 Assisted reproductive technology Fertility drugs
 Surgery

Type of assisted reproductive technologies used during seventh pregnancy

- Assisted hatching ICSI IVF
 Surgical sperm aspiration Other

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

Partner tested for patient's disorder during seventh pregnancy

- Unknown Yes-biochemical Yes-molecular No

Partner also affected by disorder during seventh pregnancy Unknown Yes No

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

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

Type of prenatal testing performed for seventh pregnancy Unknown Biochemical Molecular

Method of prenatal testing for seventh pregnancy

- Unknown Amniocentesis Chorionic villus sampling

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

- Unknown None
 Fetal echocardiogram Increased frequency prenatal ultrasounds
 Other

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

Seventh pregnancy terminated Unknown Yes No

Reason seventh pregnancy terminated

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

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

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

Patient Name _____

Date Prenatal care received during seventh pregnancy Unknown Yes No

Weeks gestation prenatal care started during seventh pregnancy _____

Treatment prescribed for patient's disorder during seventh pregnancy

Unknown Metabolic diet MedicationsBiochemical lab monitoring Avoidance of fasting Other

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

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

Patient in good metabolic condition during second trimester of seventh pregnancy

Unknown Pregnancy not sustained to second trimesterYes No

Patient in good metabolic condition during third trimester of seventh pregnancy

Unknown Pregnancy not sustained to third trimesterYes No

Number of outpatient metabolic visits for patient during seventh pregnancy _____

Number of ED visits for management of disorder during seventh pregnancy

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

Number of hospitalizations for management of disorder during seventh pregnancy

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

Complications during seventh pregnancy

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

Complications related to disorder during seventh pregnancy- specify

UnknownAcute fatty liver of pregnancy (AFLP)Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)Intrauterine growth restriction (AUGR)Mother affected with this conditionOther

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

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

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

Complications during seventh pregnancy-other, specify _____

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

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

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

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

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

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

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

Laboratory studies done on patient during seventh pregnancy

- Unknown Ammonia - Abn high

Patient Name _____

Date

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

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

Additional interventions required during seventh pregnancy due to this metabolic condition

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

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

Total maternal weight gain (in kg) during seventh pregnancy _____

Number of documented episodes of hyperammonemia during seventh pregnancy

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

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

8 9 10 >10 unknown

Number of documented episodes of ketonuria during seventh pregnancy

0 1 2 3 4 5 6 7

8 9 10 >10 unknown

Number of documented episodes of metabolic acidosis during seventh pregnancy

0 1 2 3 4 5 6 7

8 9 10 >10 unknown

Treatment(s) for hyperammonemic episodes during seventh pregnancy

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

Maternal protein intake restricted during seventh pregnancy Unknown Yes No

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

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

Ammonia scavenger drugs used during seventh pregnancy

- Unknown No scavengers used Buphenyl Sodium benzoate

Other medications/supplements used during seventh pregnancy

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

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

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

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

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

Patient Name _____

Date

- Altered anesthesia plan
- Other

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

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

Abnormal patient lab results during seventh delivery

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

Abnormal patient lab results during seventh delivery-other, specify

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

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

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

Patient death during or shortly after seventh delivery Unknown Yes No

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

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

Method of seventh delivery

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

Maternal inpatient days post- seventh delivery_____

Live delivery of seventh newborn Unknown Yes No

Patient Name _____

Date

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

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

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

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

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

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

Birth measurements for seventh newborn Unknown Head circumference Length Weight

Birth head circumference _____

Birth head circumference units cm in

Birth length _____

Birth length units cm in

Birth weight _____

Birth weight units lbs kg gm oz

APGAR score at 1 minute for seventh newborn

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

Patient Name _____

Date

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

APGAR score at 5 minutes for seventh newborn

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

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

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

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

Routine newborn screening done for seventh newborn

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

Describe abnormal newborn screen result for seventh newborn

Reason routine newborn screening was not done for seventh newborn

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

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

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

Patient Name _____

Date

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

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

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

- Unknown Yes No

Abnormal newborn exam findings for seventh newborn

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

Abnormal newborn exam findings for seventh newborn-other, specify _____

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

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

Seventh newborn currently alive Unknown Yes No

Eighth Pregnancy

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

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

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

History of infertility for eighth pregnancy

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

Cause(s) of infertility for eighth pregnancy

- Unknown Male factor Female factor
 Combined male-female factor

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

Cause(s) of male infertility for eighth pregnancy

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

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

Cause(s) of female infertility for eighth pregnancy

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

Cause(s) of female infertility for eighth pregnancy-other, specify _____

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

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

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

Type of assisted reproductive technologies used during eighth pregnancy

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

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

Partner tested for patient's disorder during eighth pregnancy

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

Partner also affected by disorder during eighth pregnancy Unknown Yes No

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

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

Type of prenatal testing performed for eighth pregnancy Unknown Biochemical Molecular

Method of prenatal testing for eighth pregnancy

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

- Unknown
 Amniocentesis
 Chorionic villus sampling

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

- Unknown
 None
 Fetal echocardiogram
 Increased frequency prenatal ultrasounds
 Other

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

Eighth pregnancy terminated Unknown Yes No

Reason eighth pregnancy terminated

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

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

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for eighth pregnancy _____

Prenatal care received during eighth pregnancy Unknown Yes No

Weeks gestation prenatal care started during eighth pregnancy _____

Treatment prescribed for patient's disorder during eighth pregnancy

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

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

Patient in good metabolic condition prior to eighth pregnancy Unknown Yes 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

Patient Name _____

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

Number of outpatient metabolic visits for patient during eighth pregnancy _____

Number of ED visits for management of disorder during eighth pregnancy

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

Number of hospitalizations for management of disorder during eighth pregnancy

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

Complications during eighth pregnancy

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

Complications related to disorder during eighth pregnancy- specify

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

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

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

Complications during eighth pregnancy-other, specify _____

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

Patient Name _____

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

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

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

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

Duration of longest inpatient hospitalization (in days) for any reason during eighth pregnancy _____

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

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

Laboratory studies done on patient during eighth pregnancy

- | | |
|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Ammonia - Abn high |
| <input type="checkbox"/> Ammonia - WNL | <input type="checkbox"/> Blood glucose - Abn low |
| <input type="checkbox"/> Blood glucose - WNL | <input type="checkbox"/> Blood Glucose - Abn high |
| <input type="checkbox"/> BNP - Abn high | <input type="checkbox"/> BNP - WNL |
| <input type="checkbox"/> CBC - Abn | <input type="checkbox"/> CBC - WNL |
| <input type="checkbox"/> CO2 - Abn low | <input type="checkbox"/> CO2 - WNL |
| <input type="checkbox"/> CK - Abn high | <input type="checkbox"/> CK - WNL |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn | <input type="checkbox"/> INR - WNL |
| <input type="checkbox"/> Liver function tests - Abn high | <input type="checkbox"/> Liver function tests - WNL |
| <input type="checkbox"/> Plasma acylcarnitine profile - Abn | <input type="checkbox"/> Plasma acylcarnitine profile - WNL |
| <input type="checkbox"/> Plasma amino acids - Abn | <input type="checkbox"/> Plasma amino acids - WNL |
| <input type="checkbox"/> Plasma carnitine levels - Abn | <input type="checkbox"/> Plasma carnitine levels - WNL |
| <input type="checkbox"/> Prealbumin - Abn low | <input type="checkbox"/> Prealbumin - WNL |
| <input type="checkbox"/> Transferrin - Abn | <input type="checkbox"/> Transferrin - WNL |
| <input type="checkbox"/> Urine acylcarnitines - Abn | <input type="checkbox"/> Urine acylcarnitines - WNL |
| <input type="checkbox"/> Urine acylglycines - Abn | <input type="checkbox"/> Urine acylglycines - WNL |
| <input type="checkbox"/> Urine ketones - Abn high | <input type="checkbox"/> Urine ketones - WNL |

Patient Name _____

Date

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

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

Additional interventions required during eighth pregnancy due to this metabolic condition

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

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

Total maternal weight gain (in kg) during eighth pregnancy _____

Number of documented episodes of hyperammonemia during eighth pregnancy

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

Number of documented episodes of ketonuria during eighth pregnancy

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

Number of documented episodes of metabolic acidosis during eighth pregnancy

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

Treatment(s) for hyperammonemic episodes during eighth pregnancy

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

Maternal protein intake restricted during eighth pregnancy Unknown Yes No

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

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

Ammonia scavenger drugs used during eighth pregnancy

- Unknown No scavengers used Buphenyl Sodium benzoate

Patient Name _____

Date

Other medications/supplements used during eighth pregnancy

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

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

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

Acute health concerns experienced by the patient during delivery for eighth delivery

Abnormal patient lab results during eighth delivery

- Unknown None Normal labs during delivery
- Elevated CK Elevated liver function tests Hyperammonemia
- Hypoglycemia Metabolic decompensation Other

Abnormal patient lab results during eighth delivery-other, specify

Additional maternal interventions during or after eighth delivery related to this disorder

- Unknown Ammonul Blood/blood product transfusion
- Dialysis ICU monitoring Infusions
- Medications Resuscitation TPN
- Other

Additional maternal interventions during or after eighth delivery related to this disorder-other, specify

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

Patient death during or shortly after eighth delivery Unknown Yes No

Disorder contributed to death of mother during eighth delivery Unknown Yes No

Weeks gestation at time of eighth delivery - round to nearest week_____

Method of eighth delivery

Unknown Caesarean section (scheduled or non-urgent)

Casesarean section (emergent) Vaginal

Maternal inpatient days post- eighth delivery_____

Live delivery of eighth newborn Unknown Yes No

Acute health concerns for eighth newborn related to maternal disorder (example: fetal distress secondary to maternal acute metabolic decompensation)

Additional lab tests and results done on eighth newborn specifically due to maternal disorder history

Health concerns for eighth newborn not known to be related to disorder

Additional interventions for eighth newborn during or shortly after delivery (other than labs)

Unknown Blood/blood product transfusion

Glucose infusion Medications

NICU/special care nursery monitoring Resuscitation

TPN Other

Additional interventions for eighth newborn during or shortly after delivery-other, specify

Birth measurements for eighth newborn Unknown Head circumference Length Weight

Birth head circumference_____

Birth head circumference units cm in

Birth length_____

Birth length units cm in

Patient Name _____

Date

Birth weight _____

Birth weight units lbs kg gm oz

APGAR score at 1 minute for eighth newborn

- Unknown 0 1 2 3 4 5
- 6 7 8 9 10

Health concern(s) with APGAR score at one minute < 8 for eighth newborn

APGAR score at 5 minutes for eighth newborn

- Unknown 0 1 2 3 4 5
- 6 7 8 9 10

Health concern(s) with APGAR score at five minutes < 8 for eighth newborn

Length of eighth newborn's stay in the hospital after birth

- Unknown <24 hours
- 24-28 hours 3-5 days
- 6-14 days >14 days
- N/A - baby was not born in a hospital baby is still in the hospital

Routine newborn screening done for eighth newborn

- Unknown Yes - normal for all screened disorders
- Yes -abnormal Yes - results pending
- No

Describe abnormal newborn screen result for eighth newborn

Reason routine newborn screening was not done for eighth newborn

- Unknown
- Died prior to collection of NBS
- Refused
- Transferred to another facility prior to NBS collection
- Transfused prior to collection of NBS

Patient Name _____

Date OtherReason routine newborn screening was not done for eighth newborn-other, specify

Additional testing (beyond newborn screening) done for the eighth baby after birth to rule out the mother's disorder

- Unknown Yes - biochemical normal
 Yes - biochemical abnormal Yes - molecular normal
 Yes - molecular abnormal affected Yes - molecular abnormal unaffected carrier
 No

Additional studies and results (examples: brain MRI, echocardiogram) done on eighth newborn to assess for effects of maternal disorder

Eighth newborn examined by a genetics professional prior to or shortly after discharge from the birth hospital

 Unknown Yes No

Abnormal newborn exam findings for eighth newborn

- Unknown None Congenital heart disease
 Dysmorphism Lethargy Microcephaly
 Other congenital anomalies Poor feeding Respiratory distress
 Seizure(s) Small for gestational age Other

Abnormal newborn exam findings for eighth newborn-other, specify _____

Newborn death at or shortly after delivery for eighth newborn Unknown Yes NoNewborn's death related to maternal disorder for eighth newborn Unknown Yes NoEighth newborn currently alive Unknown Yes No

Current Pregnancy

Patient is pregnant Unknown Yes No

Patient's age at time of current pregnancy (in years) _____

Length of time patient tried to become pregnant before current pregnancy (in months)

- Unknown Unplanned 1 2 3 4 5
 6 7 8 9 10 11 12
 13-24 25-36 >36

History of infertility for current pregnancy

 Unknown

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

- Yes- not known to be related to inborn error of metabolism
- Yes-related to inborn error of metabolism
- No

Cause(s) of infertility for current pregnancy

- Unknown Male factor Female factor
- Combined male-female factor

Cause(s) of male infertility for current pregnancy

- Unknown Abnormal sperm production or function
- Age Cancer/cancer treatment related
- Environmental Problems with sperm delivery
- Tobacco use Other

Cause(s) of male infertility for current pregnancy-other, specify _____

Cause(s) of female infertility for current pregnancy

- Unknown Age
- Alcohol use Cancer/cancer treatment related
- Early menopause Fallopian tube damage/blockage
- Hyperprolactinemia Pelvic adhesions
- Polycystic ovary syndrome (PCOS) Premature ovarian insufficiency
- Over-exercise Ovulation disorder
- Thyroid problems Tobacco use
- Uterine fibroid Weight related
- Other

Cause(s) of female infertility for current pregnancy-other, specify _____

Cause(s) of infertility for current pregnancy, combined male-female factor- specify

Types of fertility treatment(s) received prior to current pregnancy

- Unknown None
- Assisted reproductive technology Fertility drugs
- Surgery

Type of assisted reproductive technologies used during current pregnancy

- Assisted hatching ICSI IVF
- Surgical sperm aspiration Other

Type of assisted reproductive technologies used during current pregnancy-other, specify

Partner tested for patient's disorder during current pregnancy

Patient Name _____ Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

Unknown
 Yes-biochemical
 Yes-molecular
 No

Partner also affected by disorder during current pregnancy
 Unknown
 Yes
 No

Preimplantation genetic diagnosis for disorder done for current pregnancy
 Unknown
 Yes
 No

Prenatal testing done for fetus for this disorder for current pregnancy
 Unknown
 Yes
 No

Type of prenatal testing performed for current pregnancy
 Unknown
 Biochemical
 Molecular

Method of prenatal testing for current pregnancy

Unknown
 Amniocentesis
 Chorionic villus sampling

Additional prenatal testing performed on fetus as a result of parent's disorder for current pregnancy

Unknown
 None

Fetal echocardiogram
 Increased frequency prenatal ultrasounds

Other

Additional prenatal testing performed on fetus as a result of parent's disorder for current pregnancy-other, specify _____

Current pregnancy terminated
 Unknown
 Yes
 No

Reason current pregnancy terminated

- Elective due to fetus affected with disorder
- Elective due to other fetal well-being unrelated to disorder
- Elective for other reason
- Elective due to maternal well-being
- Spontaneous

Gestational age (in weeks) at time of current pregnancy termination - elective or spontaneous (round to the nearest week) _____

Amount of difference in weeks gestation between the estimated date of delivery by date and by most recent ultrasound for current pregnancy _____

Prenatal care received during current pregnancy
 Unknown
 Yes
 No

Weeks gestation prenatal care started during current pregnancy _____

Treatment prescribed for patient's disorder during current pregnancy

Unknown
 Metabolic diet
 Medications
 Biochemical lab monitoring
 Avoidance of fasting
 Other

Treatment prescribed for patient's disorder during current pregnancy-other, specify

Patient in good metabolic condition prior to current pregnancy
 Unknown
 Yes
 No

Patient in good metabolic condition during first trimester of current pregnancy

Patient Name _____

Date

Unknown Yes No

Patient in good metabolic condition during second trimester of current pregnancy

Unknown Pregnancy not sustained to second trimester
 Yes No

Patient in good metabolic condition during third trimester of current pregnancy

Unknown Pregnancy not sustained to third trimester
 Yes No

Number of outpatient metabolic visits for patient during current pregnancy _____

Number of ED visits for management of disorder during current pregnancy

Unknown 0 1 2 3 4 5
 6 7 8 9 10 >10

Number of hospitalizations for management of disorder during current pregnancy

Unknown 0 1 2 3 4 5
 6 7 8 9 10 >10

Complications during current pregnancy

Unknown Yes-related to disorder
 Yes- not known to be related to disorder No

Complications related to disorder during current pregnancy- specify

- Unknown
- Acute fatty liver of pregnancy (AFLP)
- Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)
- Intrauterine growth restriction (AUGR)
- Mother affected with this condition
- Other

Complications not known to be related to disorder during current pregnancy-specify

- Unknown
- Advanced maternal age (35+ years of age)
- Ectopic pregnancy
- Gestational diabetes
- Group B strep
- Hemolysis, Elevated liver enzymes, Low platelet count (HELLP Syndrome)
- Intrauterine growth restriction (AUGR)
- Inadequate prenatal care
- Maternal prenatal substance exposure
- Preeclampsia

Patient Name _____ Date <input type="text" value="M"/> <input type="text" value="M"/> <input type="text" value="D"/> <input type="text" value="D"/> <input type="text" value="Y"/> <input type="text" value="Y"/>
--

- Rh isoimmunization
- Toxemia
- Young maternal age (15 years of age + under)
- Preterm labor
- Other

Complications during current pregnancy-other, specify _____

Number of ED visits for complications during current pregnancy, unrelated to management of disorder

- Unknown 0 1 2 3 4 5
- 6 7 8 9 10 >10

Number of hospitalizations for complications during current pregnancy, unrelated to management of disorder

- Unknown 0 1 2 3 4 5
- 6 7 8 9 10 >10

Duration of longest inpatient hospitalization (in days) for any reason during current pregnancy _____

Highest value of primary metabolite of concern during current pregnancy (specify metabolite, value, and units of measure)

Lowest value of primary metabolite of concern during current pregnancy (specify metabolite, value, and units of measure)

Laboratory studies done on patient during current pregnancy

- | | |
|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Ammonia - Abn high |
| <input type="checkbox"/> Ammonia - WNL | <input type="checkbox"/> Blood glucose - Abn low |
| <input type="checkbox"/> Blood glucose - WNL | <input type="checkbox"/> Blood Glucose - Abn high |
| <input type="checkbox"/> BNP - Abn high | <input type="checkbox"/> BNP - WNL |
| <input type="checkbox"/> CBC - Abn | <input type="checkbox"/> CBC - WNL |
| <input type="checkbox"/> CO2 - Abn low | <input type="checkbox"/> CO2 - WNL |
| <input type="checkbox"/> CK - Abn high | <input type="checkbox"/> CK - WNL |
| <input type="checkbox"/> Glucose Tolerance Test (oral) - Abn | <input type="checkbox"/> Glucose Tolerance Test (oral) - WNL |
| <input type="checkbox"/> INR - Abn | <input type="checkbox"/> INR - WNL |
| <input type="checkbox"/> Liver function tests - Abn high | <input type="checkbox"/> Liver function tests - WNL |

Patient Name _____

Date |M|M| |D|D| |Y|Y|

- 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
- Plasma acylcarnitine profile - WNL
- Plasma amino acids - WNL
- Plasma carnitine levels - WNL
- Prealbumin - WNL
- Transferrin - WNL
- Urine acylcarnitines - WNL
- Urine acylglycines - WNL
- Urine ketones - WNL
- Urine organic acids - WNL

Other laboratory studies done on patient during 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
- More intensive fetal monitoring
- Other
- ED visits for hyperemesis/IV fluids
- TPN
- Additional medications
- Increased frequency of lab monitoring
- Bedrest

Additional interventions required during current pregnancy due to this metabolic condition-other, specify

Total maternal weight gain to date (in kg) during current pregnancy_____

Number of documented episodes of hyperammonemia during current pregnancy

- 0 1 2 3 4 5 6 7
- 8 9 10 >10 unknown

Number of documented episodes of ketonuria during current pregnancy

- 0 1 2 3 4 5 6 7
- 8 9 10 >10 unknown

Number of documented episodes of metabolic acidosis during current pregnancy

- 0 1 2 3 4 5 6 7
- 8 9 10 >10 unknown

Treatment(s) for hyperammonemic episodes during current pregnancy

- Unknown
- No hyperammonemic episodes
- Hospital admission

Patient Name _____

Date

Protein restriction

Ammonul

Arginine

Maternal protein intake restricted during current pregnancy Unknown Yes No

Number of grams of synthetic protein prescribed per day during current pregnancy (units = gm/day)

Number of grams of natural protein prescribed per day during current pregnancy (units = gm/day) _____

Ammonia scavenger drugs used during current pregnancy

Unknown

No scavengers used

Buphenyl

Sodium benzoate

Other medications/supplements used during current pregnancy

Additional interventions planned for labor/delivery related to patient's disorder for current pregnancy

Unknown

None

Additional maternal lab monitoring

Altered anesthesia plan

Change in delivery site

IV fluids

Letter to OB/MFM specialist

Planned C-section

Referral for high risk OB management Other

Additional interventions planned for labor/delivery related to patient's disorder for current

pregnancy-other, specify

Comments

Pregnancy comments

Patient Name _____

Date ***bKT - Dialysis***

Number of different episodes during which dialysis (any type) was used

 1 2 3 4 5 6 7 8 9 10 >10
First Dialysis Treatment

Type(s) of dialysis received during first episode

- Unknown APD/CCPD ECMO CAPD
 CVVH CVVD CVVHDF Hemodialysis
 Peritoneal dialysis

Reason for first episode of dialysis

- Unknown Hyperammonemia MSUD Organ failure Sepsis
 Other

Reason for first episode of dialysis-other, specify

Start date of first episode of dialysis treatment

Duration (in days) of first episode of dialysis treatment _____

Metabolite(s) of concern during first episode of dialysis

- Unknown None Ammonia Blood urea nitrogen
 Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine
 Serum creatinine Other

Metabolite(s) of concern during first episode of dialysis-other,specify _____

Peak value of ammonia during first episode of dialysis _____

Peak value of ammonia during first episode of dialysis units

 umol/L ug/dL ug/L ug/mL g/dL

Peak value of blood urea nitrogen during first episode of dialysis _____

Peak value of blood urea nitrogen during first episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during first episode of dialysis _____

Peak value of plasma alloisoleucine during first episode of dialysis units

 umol/dL umol/L mg/dL

Peak value of plasma isoleucine during first episode of dialysis _____

Peak value of plasma isoleucine during first episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during first episode of dialysis _____

Patient Name _____

Date Peak value of plasma leucine during first episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during first episode of dialysis _____

Peak value of plasma valine during first episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during first episode of dialysis _____

Peak value of serum creatinine during first episode of dialysis units umol/L mg/dL

Peak value of other metabolite during first episode of dialysis _____

Peak value of other metabolite during first episode of dialysis units _____

Reason first episode of dialysis treatment was stopped

 Unknown Acute episode resolved Treatment withdrawn Death Other

Reason first episode of dialysis treatment was stopped-other, specify _____

Second Dialysis Treatment

Type(s) of dialysis received during second episode

 Unknown APD/CCPD ECMO CAPD CVVH CVVD CVVHDF Hemodialysis Peritoneal dialysis

Reason for second episode of dialysis

 Unknown Hyperammonemia MSUD Organ failure Sepsis Other

Reason for second episode of dialysis-other, specify

Start date of second episode of dialysis treatment

Duration (in days) of second episode of dialysis treatment _____

Metabolite(s) of concern during second episode of dialysis

 Unknown None Ammonia Blood urea nitrogen Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine Serum creatinine Other

Metabolite(s) of concern during second episode of dialysis- other, specify _____

Peak value of ammonia during second episode of dialysis _____

Peak value of ammonia during second episode of dialysis units

 umol/L ug/dL ug/L ug/mL g/dL

Peak value of blood urea nitrogen during second episode of dialysis _____

Peak value of blood urea nitrogen during second episode of dialysis units mg/dL mmol/L

Patient Name _____

Date

Peak value of plasma alloisoleucine during second episode of dialysis _____

Peak value of plasma alloisoleucine during second episode of dialysis units

- umol/dL
- umol/L
- mg/dL

Peak value of plasma isoleucine during second episode of dialysis _____

Peak value of plasma isoleucine during second episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during second episode of dialysis _____

Peak value of plasma leucine during second episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during second episode of dialysis _____

Peak value of plasma valine during second episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during second episode of dialysis _____

Peak value of serum creatinine during second episode of dialysis units umol/L mg/dL

Peak value of other metabolite during second episode of dialysis _____

Peak value of other metabolite during second episode of dialysis units _____

Reason second episode of dialysis treatment was stopped

- Unknown
- Acute episode resolved
- Treatment withdrawn
- Death

Other

Reason second episode of dialysis treatment was stopped-other,specify _____

Third Dialysis Treatment

Type(s) of dialysis received during third episode

- Unknown
- APD/CCPD
- ECMO
- CAPD
- CVVH
- CVVD
- CVVHDF
- Hemodialysis
- Peritoneal dialysis

Reason for third episode of dialysis

- Unknown
- Hyperammonemia
- MSUD
- Organ failure
- Sepsis
- Other

Reason for third episode of dialysis-other, specify

Start date of third episode of dialysis treatment

Duration (in days) of third episode of dialysis treatment _____

Metabolite(s) of concern during third episode of dialysis

- Unknown
- None
- Ammonia
- Blood urea nitrogen
- Plasma alloisoleucine
- Plasma isoleucine
- Plasma leucine
- Plasma valine
- Serum creatinine
- Other

Patient Name _____

Date

Metabolite(s) of concern during third episode of dialysis-other, specify _____

Peak value of ammonia during third episode of dialysis _____

Peak value of ammonia during third episode of dialysis units

 umol/L ug/dL ug/L ug/mL g/dL

Peak value of blood urea nitrogen during third episode of dialysis _____

Peak value of blood urea nitrogen during third episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during third episode of dialysis _____

Peak value of plasma alloisoleucine during third episode of dialysis units

 umol/dL umol/L mg/dL

Peak value of plasma isoleucine during third episode of dialysis _____

Peak value of plasma isoleucine during third episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during third episode of dialysis _____

Peak value of plasma leucine during third episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during third episode of dialysis _____

Peak value of plasma valine during third episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during third episode of dialysis _____

Peak value of serum creatinine during third episode of dialysis units umol/L mg/dL

Peak value of other metabolite during third episode of dialysis _____

Peak value of other metabolite during third episode of dialysis units _____

Reason third episode of dialysis treatment was stopped

- Unknown Acute episode resolved Treatment withdrawn Death
 Other

Reason third episode of dialysis treatment was stopped-other, specify _____

Fourth Dialysis Treatment

Type(s) of dialysis received during fourth episode

- Unknown APD/CCPD ECMO CAPD
 CVVH CVVD CVVHDF Hemodialysis
 Peritoneal dialysis

Reason for fourth episode of dialysis

- Unknown Hyperammonemia MSUD Organ failure Sepsis
 Other

Reason for fourth episode of dialysis-other, specify

Patient Name _____

Date Start date of fourth episode of dialysis treatment

Duration (in days) of fourth episode of dialysis treatment _____

Metabolite(s) of concern during fourth episode of dialysis

- Unknown None Ammonia Blood urea nitrogen
 Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine
 Serum creatinine Other

Metabolite(s) of concern during fourth episode of dialysis-other, specify _____

Peak value of ammonia during fourth episode of dialysis _____

Peak value of ammonia during fourth episode of dialysis units

- umol/L ug/dL ug/L ug/mL g/dL

Peak value of blood urea nitrogen during fourth episode of dialysis _____

Peak value of blood urea nitrogen during fourth episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during fourth episode of dialysis _____

Peak value of plasma alloisoleucine during fourth episode of dialysis units

- umol/dL umol/L mg/dL

Peak value of plasma isoleucine during fourth episode of dialysis _____

Peak value of plasma isoleucine during fourth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during fourth episode of dialysis _____

Peak value of plasma leucine during fourth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during fourth episode of dialysis _____

Peak value of plasma valine during fourth episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during fourth episode of dialysis _____

Peak value of serum creatinine during fourth episode of dialysis units umol/L mg/dL

Peak value of other metabolite during fourth episode of dialysis _____

Peak value of other metabolite during fourth episode of dialysis units _____

Reason fourth episode of dialysis treatment was stopped

- Unknown Acute episode resolved Treatment withdrawn Death
 Other

Reason fourth episode of dialysis treatment was stopped-other, specify _____

Fifth Dialysis Treatment

Type(s) of dialysis received during fifth episode

- Unknown APD/CCPD ECMO CAPD
 CVVH CVVD CVVHDF Hemodialysis
 Peritoneal dialysis

Reason for fifth episode of dialysis

Patient Name _____

Date

- Unknown Hyperammonemia MSUD Organ failure Sepsis
- Other

Reason for fifth episode of dialysis-other, specify

Start date of fifth episode of dialysis treatment

Duration (in days) of fifth episode of dialysis treatment _____

Metabolite(s) of concern during fifth episode of dialysis

- Unknown None Ammonia Blood urea nitrogen
- Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine
- Serum creatinine Other

Metabolite(s) of concern during fifth episode of dialysis-other, specify _____

Peak value of ammonia during fifth episode of dialysis _____

Peak value of ammonia during fifth episode of dialysis units

- umol/L ug/dL ug/L ug/mL g/dL

Peak value of blood urea nitrogen during fifth episode of dialysis _____

Peak value of blood urea nitrogen during fifth episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during fifth episode of dialysis _____

Peak value of plasma alloisoleucine during fifth episode of dialysis units

- umol/dL umol/L mg/dL

Peak value of plasma isoleucine during fifth episode of dialysis _____

Peak value of plasma isoleucine during fifth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during fifth episode of dialysis _____

Peak value of plasma leucine during fifth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during fifth episode of dialysis _____

Peak value of plasma valine during fifth episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during fifth episode of dialysis _____

Peak value of serum creatinine during fifth episode of dialysis units umol/L mg/dL

Peak value of other metabolite during fifth episode of dialysis _____

Peak value of other metabolite during fifth episode of dialysis units _____

Reason fifth episode of dialysis treatment was stopped

- Unknown Acute episode resolved Treatment withdrawn Death
- Other

Reason fifth episode of dialysis treatment was stopped-other, specify _____

Patient Name _____

Date **Sixth Dialysis Treatment**

Type(s) of dialysis received during sixth episode

- Unknown APD/CCPD ECMO CAPD
 CVVH CVVD CVVHDF Hemodialysis
 Peritoneal dialysis

Reason for sixth episode of dialysis

- Unknown Hyperammonemia MSUD Organ failure Sepsis
 Other

Reason for sixth episode of dialysis-other, specify

Start date of sixth episode dialysis treatment

Duration (in days) of sixth episode of dialysis treatment _____

Metabolite(s) of concern during sixth episode of dialysis

- Unknown None Ammonia Blood urea nitrogen
 Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine
 Serum creatinine Other

Metabolite(s) of concern during sixth episode of dialysis-other, specify _____

Peak value of ammonia during sixth episode of dialysis _____

Peak value of ammonia during sixth episode of dialysis units

- umol/L ug/dL ug/L ug/mL g/dL

Peak value of blood urea nitrogen during sixth episode of dialysis _____

Peak value of blood urea nitrogen during sixth episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during sixth episode of dialysis _____

Peak value of plasma alloisoleucine during sixth episode of dialysis units

- umol/dL umol/L mg/dL

Peak value of plasma isoleucine during sixth episode of dialysis _____

Peak value of plasma isoleucine during sixth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during sixth episode of dialysis _____

Peak value of plasma leucine during sixth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during sixth episode of dialysis _____

Peak value of plasma valine during sixth episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during sixth episode of dialysis _____

Peak value of serum creatinine during sixth episode of dialysis units umol/L mg/dL

Patient Name _____

Date | |

Peak value of other metabolite during sixth episode of dialysis _____

Peak value of other metabolite during sixth episode of dialysis units _____

Reason sixth episode of dialysis treatment was stopped

- Unknown
- Acute episode resolved
- Treatment withdrawn
- Death
- Other

Reason sixth episode of dialysis treatment was stopped-other, specify _____

Seventh Dialysis Treatment

Type(s) of dialysis received during seventh episode

- Unknown
- APD/CCPD
- ECMO
- CAPD
- CVVH
- CVVD
- CVVHDF
- Hemodialysis
- Peritoneal dialysis

Reason for seventh episode of dialysis

- Unknown
- Hyperammonemia
- MSUD
- Organ failure
- Sepsis
- Other

Reason for seventh episode of dialysis -other, specify

Start date of seventh episode of dialysis treatment | |

Duration (in days) of seventh episode of dialysis treatment _____

Metabolite(s) of concern during seventh episode of dialysis

- Unknown
- None
- Ammonia
- Blood urea nitrogen
- Plasma alloisoleucine
- Plasma isoleucine
- Plasma leucine
- Plasma valine
- Serum creatinine
- Other

Metabolite(s) of concern during seventh episode of dialysis-other, specify _____

Peak value of ammonia during seventh episode of dialysis _____

Peak value of ammonia during seventh episode of dialysis units

- umol/L
- ug/dL
- ug/L
- ug/mL
- g/dL

Peak value of blood urea nitrogen during seventh episode of dialysis _____

Peak value of blood urea nitrogen during seventh episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during seventh episode of dialysis _____

Peak value of plasma alloisoleucine during seventh episode of dialysis units

- umol/dL
- umol/L
- mg/dL

Peak value of plasma isoleucine during seventh episode of dialysis _____

Peak value of plasma isoleucine during seventh episode of dialysis units

Patient Name _____

Date umol/dL umol/L mg/dL

Peak value of plasma leucine during seventh episode of dialysis _____

Peak value of plasma leucine during seventh episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during seventh episode of dialysis _____

Peak value of plasma valine during seventh episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during seventh episode of dialysis _____

Peak value of serum creatinine during seventh episode of dialysis units umol/L mg/dL

Peak value of other metabolite during seventh episode of dialysis _____

Peak value of other metabolite during seventh episode of dialysis units _____

Reason seventh episode of dialysis treatment was stopped

 Unknown Acute episode resolved Treatment withdrawn Death Other

Reason seventh episode of dialysis treatment was stopped-other, specify _____

Eighth Dialysis Treatment

Type(s) of dialysis received during eighth episode

 Unknown APD/CCPD ECMO CAPD CVVH CVVD CVVHDF Hemodialysis Peritoneal dialysis

Reason for eighth episode of dialysis

 Unknown Hyperammonemia MSUD Organ failure Sepsis Other

Reason for eighth episode of dialysis-other, specify

Start date of eighth episode of dialysis treatment

Duration (in days) of eighth episode of dialysis treatment _____

Metabolite(s) of concern during eighth episode of dialysis

 Unknown None Ammonia Blood urea nitrogen Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine Serum creatinine Other

Metabolite(s) of concern during eighth episode of dialysis-other, specify _____

Peak value of ammonia during eighth episode of dialysis _____

Peak value of ammonia during eighth episode of dialysis units

 umol/L ug/dL ug/L ug/mL g/dL

Patient Name _____
Date <input type="text" value="M"/> <input type="text" value="M"/> <input type="text" value="D"/> <input type="text" value="D"/> <input type="text" value="Y"/> <input type="text" value="Y"/>

Peak value of blood urea nitrogen during eighth episode of dialysis_____

Peak value of blood urea nitrogen during eighth episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during eighth episode of dialysis_____

Peak value of plasma alloisoleucine during eighth episode of dialysis units

umol/dL umol/L mg/dL

Peak value of plasma isoleucine during eighth episode of dialysis_____

Peak value of plasma isoleucine during eighth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during eighth episode of dialysis_____

Peak value of plasma leucine during eighth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during eighth episode of dialysis_____

Peak value of plasma valine during eighth episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during eighth episode of dialysis_____

Peak value of serum creatinine during eighth episode of dialysis units umol/L mg/dL

Peak value of other metabolite during eighth episode of dialysis_____

Peak value of other metabolite during eighth episode of dialysis units_____

Reason eighth episode of dialysis treatment was stopped

- Unknown Acute episode resolved Treatment withdrawn Death
- Other

Reason eighth episode of dialysis treatment was stopped-other, specify_____

Ninth Dialysis Treatment

Type(s) of dialysis received during ninth episode

- Unknown APD/CCPD ECMO CAPD
- CVVH CVVD CVVHDF Hemodialysis
- Peritoneal dialysis

Reason for ninth episode of dialysis

- Unknown Hyperammonemia MSUD Organ failure Sepsis
- Other

Reason for ninth episode of dialysis-other, specify

Start date of ninth episode of dialysis treatment | |

Duration (in days) of ninth episode of dialysis treatment_____

Metabolite(s) of concern during ninth episode of dialysis

- Unknown None Ammonia Blood urea nitrogen

Patient Name _____

Date Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine Serum creatinine Other

Metabolite(s) of concern during ninth episode of dialysis-other, specify _____

Peak value of ammonia during ninth episode of dialysis _____

Peak value of ammonia during ninth episode of dialysis units

 umol/L ug/dL ug/L ug/mL g/dL

Peak value of blood urea nitrogen during ninth episode of dialysis _____

Peak value of blood urea nitrogen during ninth episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during ninth episode of dialysis _____

Peak value of plasma alloisoleucine during ninth episode of dialysis units

 umol/dL umol/L mg/dL

Peak value of plasma isoleucine during ninth episode of dialysis _____

Peak value of plasma isoleucine during ninth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during ninth episode of dialysis _____

Peak value of plasma leucine during ninth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during ninth episode of dialysis _____

Peak value of plasma valine during ninth episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during ninth episode of dialysis _____

Peak value of serum creatinine during ninth episode of dialysis units umol/L mg/dL

Peak value of other metabolite during ninth episode of dialysis _____

Peak value of other metabolite during ninth episode of dialysis units _____

Reason ninth episode of dialysis treatment was stopped

 Unknown Acute episode resolved Treatment withdrawn Death Other

Reason ninth episode of dialysis treatment was stopped-other, specify _____

Tenth Dialysis Treatment

Type(s) of dialysis received during tenth episode

 Unknown APD/CCPD ECMO CAPD CVVH CVVD CVVHDF Hemodialysis Peritoneal dialysis

Reason for tenth episode of dialysis

 Unknown Hyperammonemia MSUD Organ failure Sepsis Other

Patient Name _____

Date

Reason for tenth episode of dialysis-other, specify

Start date of tenth episode of dialysis treatment

Duration (in days) of tenth episode of dialysis treatment _____

Metabolite(s) of concern during tenth episode of dialysis

- Unknown None Ammonia Blood urea nitrogen
- Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine
- Serum creatinine Other

Metabolite(s) of concern during tenth episode of dialysis-other, specify _____

Peak value of ammonia during tenth episode of dialysis _____

Peak value of ammonia during tenth episode of dialysis units

- umol/L ug/dL ug/L ug/mL g/dL

Peak value of blood urea nitrogen during tenth episode of dialysis _____

Peak value of blood urea nitrogen during tenth episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during tenth episode of dialysis _____

Peak value of plasma alloisoleucine during tenth episode of dialysis units

- umol/dL umol/L mg/dL

Peak value of plasma isoleucine during tenth episode of dialysis _____

Peak value of plasma isoleucine during tenth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during tenth episode of dialysis _____

Peak value of plasma leucine during tenth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during tenth episode of dialysis _____

Peak value of plasma valine during tenth episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during tenth episode of dialysis _____

Peak value of serum creatinine during tenth episode of dialysis units umol/L mg/dL

Peak value of other metabolite during tenth episode of dialysis _____

Peak value of other metabolite during tenth episode of dialysis units _____

Reason tenth episode of dialysis treatment was stopped

- Unknown Acute episode resolved Treatment withdrawn Death
- Other

Reason tenth episode of dialysis treatment was stopped-other, specify _____

Patient Name _____

Date

If >10 dialysis treatments, for each episode: enter start date, duration (days), and reason for stopping dialysis

Comments

Dialysis comments

Patient Name _____

Date ***bKT - Transplant***Number of organ transplants received 1 2 3 4 5 >5**First Transplant**

First organ received by transplant

- Unknown Bone marrow Heart Kidney Liver Lung
- Pancreas Stem Cell Other

First organ received by transplant, other- specify _____

Age (in days) at first transplant _____

Reason for first transplant

- Unknown Treatment of disorder Renal failure Liver failure
- Heart failure Other

Reason for first transplant, other- specify _____

Patient followed by Metabolism on an outpatient basis post first transplant Unknown Yes No

Number of outpatient metabolic visits in the last year post first transplant

- 1 2 3 4 5 6 7 8 9 10 >10

Metabolic labs monitored post first transplant Unknown Yes No

Metabolic lab monitoring post first transplant: Note date(s), test(s), and normal or abnormal result(s)

Known complications during the first transplant procedure Unknown Yes No

Known complications during the first transplant procedure- specify

- Clotting Death Major bleeding Other

Known complications during the first transplant procedure, other- specify _____

Known complications post first transplant Unknown Yes No

Known complications post first transplant- specify

- | | |
|--|---|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Cancer |
| <input type="checkbox"/> Cataract | <input type="checkbox"/> Clotting |
| <input type="checkbox"/> Death | <input type="checkbox"/> Delayed graft function |
| <input type="checkbox"/> Diabetes mellitus | <input type="checkbox"/> High cholesterol |
| <input type="checkbox"/> Hypertension | <input type="checkbox"/> Infection |
| <input type="checkbox"/> Major bleeding | <input type="checkbox"/> Osteoporosis |
| <input type="checkbox"/> Reappearance of heart disease | <input type="checkbox"/> Reappearance of kidney disease |

Patient Name _____

Date Reappearance of liver disease Rejection Transplant related renal impairment Other

Known complications post first transplant, other- specify _____

Second Transplant

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 >10Metabolic labs monitored post second transplant Unknown Yes No

Metabolic lab monitoring post second transplant: Note date(s), test(s), and normal or abnormal result(s)

--

Complications during the second transplant procedure Unknown Yes No

Complications during the second transplant procedure- specify

 Clotting Death Major bleeding Other

Complications during the second transplant procedure, other- specify _____

Complications post second transplant Unknown Yes No

Complications post second transplant- specify

<input type="checkbox"/> Unknown	<input type="checkbox"/> Cancer
<input type="checkbox"/> Cataract	<input type="checkbox"/> Clotting
<input type="checkbox"/> Death	<input type="checkbox"/> Delayed graft function
<input type="checkbox"/> Diabetes mellitus	<input type="checkbox"/> High cholesterol
<input type="checkbox"/> Hypertension	<input type="checkbox"/> Infection
<input type="checkbox"/> Major bleeding	<input type="checkbox"/> Osteoporosis
<input type="checkbox"/> Reappearance of heart disease	<input type="checkbox"/> Reappearance of kidney disease
<input type="checkbox"/> Reappearance of liver disease	<input type="checkbox"/> Rejection

Patient Name _____

Date Transplant related renal impairment Other

Complications post second transplant, other- specify _____

Third Transplant

Third organ received by transplant

Unknown Bone marrow Heart Kidney Liver Lung
 Pancreas Stem Cell Other

Third organ received by transplant, other- specify _____

Age (in days) at third transplant _____

Reason for third transplant

Unknown Treatment of disorder Renal failure Liver failure
 Heart failure Other

Reason for third transplant, other- specify _____

Patient followed by Metabolism on an outpatient basis post third transplant Unknown Yes No

Number of outpatient metabolic visits in the last year post transplant

1 2 3 4 5 6 7 8 9 10 >10

Metabolic labs monitored post third transplant Unknown Yes No

Metabolic lab monitoring post third transplant: Note date(s), test(s), and normal or abnormal result(s)

Complications during the third transplant procedure Unknown Yes No

Complications during the third transplant procedure- specify

Clotting Death Major bleeding Other

Complications during the third transplant procedure, other- specify _____

Complications post third transplant Unknown Yes No

Complications post third transplant- specify

Unknown Cancer
 Cataract Clotting
 Death Delayed graft function
 Diabetes mellitus High cholesterol
 Hypertension Infection
 Major bleeding Osteoporosis
 Reappearance of heart disease Reappearance of kidney disease
 Reappearance of liver disease Rejection
 Transplant related renal impairment Other

Patient Name _____

Date

Complications post third transplant, other- specify _____

Fourth Transplant

Fourth organ received by transplant

- Unknown Bone marrow Heart Kidney Liver Lung
 Pancreas Stem Cell Other

Fourth organ received by transplant, other- specify _____

Age (in days) at fourth transplant _____

Reason for fourth transplant

- Unknown Treatment of disorder Renal failure Liver failure
 Heart failure Other

Reason for fourth transplant, other- specify _____

Patient followed by Metabolism on an outpatient basis post fourth transplant Unknown Yes No

Number of outpatient metabolic visits in the last year post transplant

- 1 2 3 4 5 6 7 8 9 10 >10

Metabolic labs monitored post fourth transplant Unknown Yes No

Metabolic lab monitoring post fourth transplant: Note date(s), test(s), and normal or abnormal result(s)

Complications during the fourth transplant procedure Unknown Yes No

Complications during the fourth transplant procedure- specify

- Clotting Death Major bleeding Other

Complications during the fourth transplant procedure, other- specify _____

Complications post fourth transplant Unknown Yes No

Complications post fourth transplant- specify

- Unknown Cancer
 Cataract Clotting
 Death Delayed graft function
 Diabetes mellitus High cholesterol
 Hypertension Infection
 Major bleeding Osteoporosis
 Reappearance of heart disease Reappearance of kidney disease
 Reappearance of liver disease Rejection
 Transplant related renal impairment Other

Complications post fourth transplant, other- specify _____

Patient Name _____

Date **Fifth Transplant**

Fifth organ received by transplant

- Unknown Bone marrow Heart Kidney Liver Lung
 Pancreas Stem Cell Other

Fifth organ received by transplant, other- specify _____

Age (in days) at fifth transplant _____

Reason for fifth transplant

- Unknown Treatment of disorder Renal failure Liver failure
 Heart failure Other

Reason for fifth transplant, other- specify _____

Patient followed by Metabolism on an outpatient basis post fifth transplant Unknown Yes No

Number of outpatient metabolic visits in the last year post transplant

- 1 2 3 4 5 6 7 8 9 10 >10

Metabolic labs monitored post fifth transplant Unknown Yes No

Metabolic lab monitoring post fifth transplant: Note date(s), test(s), and normal or abnormal result(s)

Complications during the fifth transplant procedure Unknown Yes No

Complications during the fifth transplant procedure- specify

- Clotting Death Major bleeding Other

Complications during the fifth transplant procedure, other- specify _____

Complications post fifth transplant Unknown Yes No

Complications post fifth transplant- specify

- | | |
|--|---|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Cancer |
| <input type="checkbox"/> Cataract | <input type="checkbox"/> Clotting |
| <input type="checkbox"/> Death | <input type="checkbox"/> Delayed graft function |
| <input type="checkbox"/> Diabetes mellitus | <input type="checkbox"/> High cholesterol |
| <input type="checkbox"/> Hypertension | <input type="checkbox"/> Infection |
| <input type="checkbox"/> Major bleeding | <input type="checkbox"/> Osteoporosis |
| <input type="checkbox"/> Reappearance of heart disease | <input type="checkbox"/> Reappearance of kidney disease |
| <input type="checkbox"/> Reappearance of liver disease | <input type="checkbox"/> Rejection |
| <input type="checkbox"/> Transplant related renal impairment | <input type="checkbox"/> Other |

Complications post fifth transplant, other- specify _____

Patient Name _____

Date

Provide details for any other transplants not listed above

Comments

Transplant comments