

Patient Name _____

Date

LCHAD - Intake Demographics

NBSTRN ID _____

Version 2.1.2

Center name

 A B C D E F G H I J K L M N O P Q R S T U V OtherIntake date

Consent

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

Demographics Information

Patient last name _____

Patient first name _____

Date of birth

Age _____

Gestational age in weeks _____

Societal sex Unknown Male Female

Biological sex

- Not tested Unknown XX genotype/Female
- XY genotype/Male XXX Triple X syndrome XXY Klinefelter's syndrome
- XO Turner's syndrome XXXY syndrome XXYY syndrome
- Mosaic including XXXXY Penta X syndrome Other

Biological sex-other, specify _____

Biological mother's maiden name _____

Zip code _____

Condition

Patient condition category Fatty acid oxidation disorders

Specify fatty acid oxidation disorder diagnosis for the patient

 Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)

Patient disorder identification method

Patient Name _____

Date

- Unknown

 Abnormal newborn screen

 Abnormal labs
 Clinical presentation

 Family member with this condition

Family member with this condition

- Biological mother
 Biological father
 Full sibling

 Half sibling

 Other

Family member with this condition _____

Care and Other Studies

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)

Patient Name _____

Date

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

Patient education

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

Special education services received prior to intake Unknown Yes No

Age patient qualified for special education services

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

Ancestral Origin, Race and EthnicityAncestral 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

Patient Name _____

Date Romania Serbia Slovakia Slovenia Spain 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

Patient Name _____

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

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

Medical Coverage

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

LCHAD - 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 Borderline

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

Sibling 2: Specify the type of diagnostic tests performed

Unknown Biochemical Molecular Imaging Other

Sibling 2: Diagnostic tests performed-other, specify _____

Sibling 2: Deceased Unknown Yes No

Sibling 2: Age of death (in years) _____

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

Sibling 3: Year of birth _____

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

Sibling 3: NBSTRN ID for this study _____

Sibling 3: Method of diagnosis

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

Sibling 3: Method of diagnosis-other, specify _____

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

Sibling 3: Results of newborn screening for this condition

Unavailable Presumptive positive Negative/Normal Borderline

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

Sibling 3: Specify the type of diagnostic tests performed

Unknown Biochemical Molecular Imaging Other

Sibling 3: Diagnostic tests performed-other, specify _____

Sibling 3: Deceased Unknown Yes No

Patient Name _____

Date

Sibling 3: Age of death (in years) _____

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

Sibling 4: Year of birth _____

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

Sibling 4: NBSTRN ID for this study _____

Sibling 4: Method of diagnosis

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

Sibling 4: Method of diagnosis-other, specify _____

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

Sibling 4: Results of newborn screening for this condition

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

Sibling 4: Specify the type of diagnostic tests performed

 Unknown Biochemical Molecular Imaging Other

Sibling 4: Diagnostic tests performed-other, specify _____

Sibling 4: Deceased Unknown Yes No

Sibling 4: Age of death (in years) _____

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

Sibling 5: Year of birth _____

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

Sibling 5: NBSTRN ID for this study _____

Sibling 5: Method of diagnosis

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

Sibling 5: Method of diagnosis-other, specify _____

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

Patient Name _____

Date

Sibling 5: Results of newborn screening for this condition

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

Sibling 5: Specify the type of diagnostic tests performed

 Unknown Biochemical Molecular Imaging Other

Sibling 5: Diagnostic tests performed-other, specify _____

Sibling 5: Deceased Unknown Yes No

Sibling 5: Age of death (in years) _____

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

Sibling 6: Year of birth _____

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

Sibling 6: NBSTRN ID for this study _____

Sibling 6: Method of diagnosis

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

Sibling 6: Method of diagnosis-other, specify _____

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

Sibling 6: Results of newborn screening for this condition

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

Sibling 6: Specify the type of diagnostic tests performed

 Unknown Biochemical Molecular Imaging Other

Sibling 6: Diagnostic tests performed-other, specify _____

Sibling 6: Deceased Unknown Yes No

Sibling 6: Age of death (in years) _____

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

Sibling 7: Year of birth _____

Sibling 7: Affected with this condition Unknown Yes No

Patient Name _____

Date Sibling 7: Enrolled in this study Unknown Yes No

Sibling 7: NBSTRN ID for this study _____

Sibling 7: Method of diagnosis

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

Sibling 7: Method of diagnosis-other, specify _____

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

Sibling 7: Results of newborn screening for this condition

Unavailable Presumptive positive Negative/Normal 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

LCHAD - Intake Past Health History

Prenatal History

Prenatal diagnosis done for this condition Unknown Yes No

Form of prenatal diagnosis

- Unknown Amniocentesis Chorionic villus sampling (CVS)
 Fetal blood

Amniocentesis diagnosis Biochemical/enzyme DNA

Chorionic villus sampling (CVS) diagnosis Biochemical/enzyme DNA

Issues concerning mother's pregnancy with this patient

- Unknown None Pregnancy complications
 Assisted reproduction

Pregnancy complications/risk factors

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

Type of prenatal substance exposure

- Unknown Alcohol Tobacco Illicit drugs
 Harmful chemicals Known teratogens

Maternal treatment for affected fetus Unknown None Yes No

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 |

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- | | |
|---|---|
| <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 NoDate of last outpatient specialty visit

Days of age from birth until intervention for this condition _____

Days of age from birth until first seen by subspecialist _____

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.

Other History

Cardiomyopathy prior to intake Unknown No No imaging done Dilated HypertrophicCardiomyopathy-dilated Unknown Left Right BothCardiomyopathy-hypertrophic Unknown Left Right BothHospitalizations 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 _____

Patient Name _____

Date |||||

Comorbidities at time of intake

Prior testing

Echocardiogram prior to intake Unknown Yes No

Echocardiogram date |||||

Echocardiogram results Unknown Normal Abnormal

Echocardiogram comments

Electrocardiogram prior to intake Unknown Yes No

Electrocardiogram date |||||

Electrocardiogram results Unknown Normal Abnormal

Electrocardiogram comments

Neurological imaging prior to intake Unknown Yes No

Neurological imaging date |||||

Neurological imaging results Unknown Normal Abnormal

Neurological imaging comments

Patient has biospecimen stored Unknown Yes No

Type of patient biospecimen Blood Urine Other tissue

Type of patient biospecimen-other tissue, specify _____

Specify biospecimen location _____

Eye Exam

Eye exam performed prior to intake Unknown Yes No

Eye exam findings

Unknown Within normal limits

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

Evidence of pigmentary retinopathy Other

Eye exam findings-other, specify _____

Emergency Management

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

Name of web-based emergency alert program _____

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

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

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

Unknown

Yes

No

Comments

Intake past health history comments

Patient Name _____

Date

LCHAD - 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

3-Hydroxydecanoylcarnitine (C10:1-OH) on first newborn screen _____

3-Hydroxydecanoylcarnitine (C10:1-OH) on first newborn screen units umol/L

3-Hydroxydodecanoylcarnitine (C12:1-OH) on first newborn screen _____

3-Hydroxydodecanoylcarnitine (C12:1-OH) on first newborn screen units umol/L

3-Hydroxydodecanoylcarnitine (C12-OH) on first newborn screen _____

3-Hydroxydodecanoylcarnitine (C12-OH) on first newborn screen units umol/L

Tetradecanoylcarnitine (C14:1) on first newborn screen _____

Tetradecanoylcarnitine (C14:1) on first newborn screen units umol/L

3-Hydroxytetradecanoylcarnitine (C14:2-OH) on first newborn screen _____

3-Hydroxytetradecanoylcarnitine (C14:2-OH) on first newborn screen units umol/L

3-Hydroxytetradecanoylcarnitine (C14:1-OH) on first newborn screen _____

3-Hydroxytetradecanoylcarnitine (C14:1-OH) on first newborn screen units umol/L

3-Hydroxytetradecanoylcarnitine (C14-OH) on first newborn screen _____

3-Hydroxytetradecanoylcarnitine (C14-OH) on first newborn screen units umol/L

Palmitoleylcarnitine (C16:1) on first newborn screen _____

Palmitoleylcarnitine (C16:1) on first newborn screen units umol/L

Palmitoylcarnitine (C16) on first newborn screen _____

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

3-Hydroxypalmitoleylcarnitine (C16:1-OH) on first newborn screen _____

3-Hydroxypalmitoleylcarnitine (C16:1-OH) on first newborn screen units umol/L

3-Hydroxypalmitoylcarnitine (C16-OH) on first newborn screen _____

3-Hydroxypalmitoylcarnitine (C16-OH) on first newborn screen units umol/L

3-Hydroxypalmitoylcarnitine (C16-OH)/Palmitoylcarnitine (C16) on first newborn screen _____

Linoleoylcarnitine (C18:2) on first newborn screen _____

Linoleoylcarnitine (C18:2) on first newborn screen units umol/L

Patient Name _____

Date

Oleoylcarnitine (C18:1) on first newborn screen _____

Oleoylcarnitine (C18:1) on first newborn screen units umol/L

Stearoylcarnitine (C18) on first newborn screen _____

Stearoylcarnitine (C18) on first newborn screen units umol/L

3-Hydroxylinoleoylcarnitine (C18:2-OH) on first newborn screen _____

3-Hydroxylinoleoylcarnitine (C18:2-OH) on first newborn screen units umol/L

3-Hydroxyoleoylcarnitine (C18:1-OH) on first newborn screen _____

3-Hydroxyoleoylcarnitine (C18:1-OH) on first newborn screen units umol/L

3-Hydroxystearoylcarnitine (C18-OH) on first newborn screen _____

3-Hydroxystearoylcarnitine (C18-OH) 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

- | | | |
|---|--|--|
| <input type="checkbox"/> Unsatisfactory | <input type="checkbox"/> Borderline Result | <input type="checkbox"/> NICU Protocol |
| <input type="checkbox"/> TPN | <input type="checkbox"/> Transfused | <input type="checkbox"/> State Mandate |
| <input type="checkbox"/> NBS collected < 24 hours of age <input type="checkbox"/> Other | | |

Reason for second newborn screen-other, specify _____

3-Hydroxydecanoylcarnitine (C10:1-OH) on second newborn screen _____

3-Hydroxydecanoylcarnitine (C10:1-OH) on second newborn screen units umol/L

3-Hydroxydodecanoylcarnitine (C12:1-OH) on second newborn screen _____

3-Hydroxydodecanoylcarnitine (C12:1-OH) on second newborn screen units umol/L

3-Hydroxydodecanoylcarnitine (C12-OH) on second newborn screen _____

3-Hydroxydodecanoylcarnitine (C12-OH) on second newborn screen units umol/L

Tetradecanoylcarnitine (C14:1) on second newborn screen _____

Tetradecanoylcarnitine (C14:1) on second newborn screen units umol/L

3-Hydroxytetradecadienoylcarnitine (C14:2-OH) on second newborn screen _____

3-Hydroxytetradecadienoylcarnitine (C14:2-OH) on second newborn screen units umol/L

3-Hydroxytetradecanoylcarnitine (C14:1-OH) on second newborn screen _____

3-Hydroxytetradecanoylcarnitine (C14:1-OH) on second newborn screen units umol/L

3-Hydroxytetradecanoylcarnitine (C14-OH) on second newborn screen _____

3-Hydroxytetradecanoylcarnitine (C14-OH) on second newborn screen units umol/L

Patient Name _____

Date | |

Palmitoleylcarnitine (C16:1) on second newborn screen _____

Palmitoleylcarnitine (C16:1) on second newborn screen units umol/L

Palmitoylcarnitine (C16) on second newborn screen _____

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

3-Hydroxypalmitoleylcarnitine (C16:1-OH) on second newborn screen _____

3-Hydroxypalmitoleylcarnitine (C16:1-OH) on second newborn screen units umol/L

3-Hydroxypalmitoylcarnitine (C16-OH) on second newborn screen _____

3-Hydroxypalmitoylcarnitine (C16-OH) on second newborn screen units umol/L

3-Hydroxypalmitoylcarnitine (C16-OH)/Palmitoylcarnitine (C16) on second newborn screen _____

Linoleylcarnitine (C18:2) on second newborn screen _____

Linoleoylcarnitine (C18:2) on second newborn screen units umol/L

Oleoylcarnitine (C18:1) on second newborn screen _____

Oleoylcarnitine (C18:1) on second newborn screen units umol/L

Stearoylcarnitine (C18) on second newborn screen _____

Stearoylcarnitine (C18) on second newborn screen units umol/L

3-Hydroxylinoleoylcarnitine (C18:2-OH) on second newborn screen _____

3-Hydroxylinoleoylcarnitine (C18:2-OH) on second newborn screen units umol/L

3-Hydroxyoleoylcarnitine (C18:1-OH) on second newborn screen _____

3-Hydroxyoleoylcarnitine (C18:1-OH) on second newborn screen units umol/L

3-Hydroxystearoylcarnitine (C18-OH) on second newborn screen _____

3-Hydroxystearoylcarnitine (C18-OH) on second newborn screen units umol/L

Other result/report on second newborn screen

Other result/report on second newborn screen units _____

Date third newborn screen collected | |

Reason for third newborn screen

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

Reason for third newborn screen-other, specify _____

3-Hydroxydeceoylcarnitine (C10:1-OH) on third newborn screen _____

3-Hydroxydeceoylcarnitine (C10:1-OH) on third newborn screen units umol/L

3-Hydroxydodeceoylcarnitine (C12:1-OH) on third newborn screen _____

3-Hydroxydodeceoylcarnitine (C12:1-OH) on third newborn screen units umol/L

Patient Name _____

Date

3-Hydroxydodecanoylcarnitine (C12-OH) on third newborn screen _____

3-Hydroxydodecanoylcarnitine (C12-OH) on third newborn screen units umol/L

Tetradecenoylcarnitine (C14:1) on third newborn screen _____

Tetradecenoylcarnitine (C14:1) on third newborn screen units umol/L

3-Hydroxytetradecadienoylcarnitine (C14:2-OH) on third newborn screen _____

3-Hydroxytetradecadienoylcarnitine (C14:2-OH) on third newborn screen units umol/L

3-Hydroxytetradecenoylcarnitine (C14:1-OH) on third newborn screen _____

3-Hydroxytetradecenoylcarnitine (C14:1-OH) on third newborn screen units umol/L

3-Hydroxytetradecanoylcarnitine (C14-OH) on third newborn screen _____

3-Hydroxytetradecanoylcarnitine (C14-OH) on third newborn screen units umol/L

Palmitoleylcarnitine (C16:1) on third newborn screen _____

Palmitoleylcarnitine (C16:1) on third newborn screen units umol/L

Palmitoylcarnitine (C16) on third newborn screen _____

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

3-Hydroxypalmitoleylcarnitine (C16:1-OH) on third newborn screen _____

3-Hydroxypalmitoleylcarnitine (C16:1-OH) on third newborn screen units umol/L

3-Hydroxypalmitoylcarnitine (C16-OH) on third newborn screen _____

3-Hydroxypalmitoylcarnitine (C16-OH) on third newborn screen units umol/L

3-Hydroxypalmitoylcarnitine (C16-OH)/Palmitoylcarnitine (C16) on third newborn screen _____

Linoleoylcarnitine (C18:2) on third newborn screen _____

Linoleoylcarnitine (C18:2) on third newborn screen units umol/L

Oleoylcarnitine (C18:1) on third newborn screen _____

Oleoylcarnitine (C18:1) on third newborn screen units umol/L

Stearoylcarnitine (C18) on third newborn screen _____

Stearoylcarnitine (C18) on third newborn screen units umol/L

3-Hydroxylinoleoylcarnitine (C18:2-OH) on third newborn screen _____

3-Hydroxylinoleoylcarnitine (C18:2-OH) on third newborn screen units umol/L

3-Hydroxyoleoylcarnitine (C18:1-OH) on third newborn screen _____

3-Hydroxyoleoylcarnitine (C18:1-OH) on third newborn screen units umol/L

3-Hydroxystearoylcarnitine (C18-OH) on third newborn screen _____

3-Hydroxystearoylcarnitine (C18-OH) on third newborn screen units umol/L

Other result/report on third newborn screen

--

Patient Name _____

Date

Other result/report on third newborn screen units _____

Newborn Hearing Screen

Newborn hearing screen performed Unknown Yes NoR Ear: Equipment Type Unknown DPOAE TEOAE ABR AABR ALGO OtherRight ear: Screening test results Pass Refer Not TestedL Ear: Equipment Type Unknown DPOAE TEOAE ABR AABR ALGO OtherLeft ear: Screening test results Pass Refer Not Tested**Recommendation** Repeat hearing screen Referral for diagnostic testing Risk factor monitoring only Refused further action No further action requiredDate 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 OtherLeft 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 ***LCHAD - 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 |M|M| |D|D| |Y|Y|

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

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

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

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

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

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

Diagnostic Testing

Diagnostic labs performed

- Unknown
- None
- Blood spot acylcarnitine profile
- Fatty acid oxidation probe assay
- Long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay
- Plasma acylcarnitine profile
- Plasma carnitine
- Plasma fatty acid profile
- Urine organic acids
- Western blot
- Other

Blood spot acylcarnitine profile Unknown Within normal limits Abnormal

Blood spot acylcarnitine profile significance Diagnostic Non-diagnostic

Blood spot acylcarnitine profile comments

Blood spot acylcarnitine profile reference range _____

Fatty acid oxidation probe assay Unknown Within normal limits Abnormal

Fatty acid oxidation probe assay significance Diagnostic Non-diagnostic

Fatty acid oxidation probe value _____

Fatty acid oxidation probe units nmol/mg protein mU/mg

Fatty acid oxidation probe reference range _____

Number of times long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay done 1 2

Long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay

Unknown Within normal limits Abnormal

Long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay significance

Diagnostic Non-diagnostic

Long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay value _____

Long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay units U/mL mU/mg protein

Long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay reference range

Patient Name _____

Date

Long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay medium

- Unknown Fibroblasts Liver Lymphocytes Red blood cells
- Other

Long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay medium, other- specify

Second long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay

- Unknown Within normal limits Abnormal

Second long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay significance

- Diagnostic Non-diagnostic

Second long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay value _____

Second long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay units

- U/mL mU/mg protein

Second long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay reference range

Second long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay medium

- Unknown Fibroblasts Liver Lymphocytes Red blood cells
- Other

Second long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay medium, other- specify

Plasma acylcarnitine profile Unknown Within normal limits Abnormal

Plasma acylcarnitine profile significance Diagnostic Non-diagnostic

Plasma acylcarnitine profile 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

Patient Name _____

Date |||||

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 _____

Plasma fatty acid profile, comprehensive (C8-C26) Unknown Within normal limits Abnormal

Plasma fatty acid profile, comprehensive (C8-C26) significance Diagnostic Non-diagnostic

Plasma fatty acid profile comments

Urine organic acids Unknown Within normal limits Abnormal

Urine organic acids significance Diagnostic Non-diagnostic

Urine organic acid comments

Western blot < 20% >=20% < 50% >=50% Results unavailable

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 _____

Patient Name _____

Date

Other diagnostic lab reference range _____

Genetic Testing

Type of genetic/genomic testing

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

Reason genotyping was done

- Unknown Diagnosis confirmation Modifier genes Pharmacogenetics
 Other

Reason genotyping was done-other, specify _____

Reason genotyping was not done

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

Reason genotyping was not done-other, specify _____

Type of genetic/genomic testing-other, specify _____

Gene(s) associated with LCHAD HADHA Other

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

HADHA: Specify allele 1 c.1528G>C (p.E474Q) Other

HADHA: Specify allele 1-other, specify _____

HADHA: Specify allele 2 c.1528G>C (p.E474Q) Other

HADHA: Specify allele 2-other, specify _____

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

LCHAD - Visit Demographics And History

Visit Date Date of last outpatient speciality visit

Consent

Patient consent valid Unknown Yes No

Obtain new consent prior to completing data entry.

Care and Other Studies

Providers seen at this visit

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

Providers seen at this visit, other- specify _____

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

Location of visit-other, specify _____

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

Miles from home to primary care _____

Miles from home to specialty care _____

Name of primary care center _____

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

Identify the research study _____

Clinicaltrials.gov identifier _____

Education

Education status has changed since the last visit Unknown Yes No

Complete education questions on the Demographics form.

Medical Coverage

Medical coverage at visit

- Unknown
 None
 Commercial/private
 Medicaid

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

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

Medical coverage at visit-Patient assistance program, specify _____

Medical coverage at visit-Other, specify _____

Family History

Patient has new biological sibling since last visit Unknown Yes No

Complete sibling history questions on the Family History form.

Comments

Visit demographics and history comments

Patient Name _____

Date

LCHAD - 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

Highest level of CK since last visit _____

Highest level of CK since last visit units U/L umol/L mg/dLDocumented hyperammonemia since last visit Unknown Yes No

Highest level of ammonia since last visit _____

Highest level of ammonia since last visit units umol/L ug/dL

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

Patient Name _____

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

Direct hospital admission Other

Patient was admitted to the hospital as a result of sick visit 1 Unknown Yes No

Name of hospital for sick visit 1 _____

ICD-9 codes for sick visit 1 known Yes No

ICD-9 codes for sick visit 1 _____

Diagnosis for sick visit 1 _____

Number of inpatient days for sick visit 1 _____

Number of ICU days for sick visit 1 _____

Date of sick visit 2 |M|M| |D|D| |Y|Y|

Reason for sick visit 2 Unknown Condition related Condition unrelated

Sick visit 2 was a condition exacerbation Unknown Yes No

Location for sick visit 2

Unknown Emergency department Retail clinic

Primary care Specialty center Urgent care

Direct hospital admission Other

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

Name of hospital for sick visit 2 _____

ICD-9 codes for sick visit 2 known Yes No

ICD-9 codes for sick visit 2 _____

Diagnosis for sick visit 2 _____

Number of inpatient days for sick visit 2 _____

Number of ICU days for sick visit 2 _____

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

Reason for sick visit 3 Unknown Condition related Condition unrelated

Sick visit 3 was a condition exacerbation Unknown Yes No

Location for sick visit 3

Unknown Emergency department Retail clinic

Primary care Specialty center Urgent care

Direct hospital admission Other

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

Name of hospital for sick visit 3 _____

ICD-9 codes for sick visit 3 known Yes No

ICD-9 codes for sick visit 3 _____

Diagnosis for sick visit 3 _____

Number of inpatient days for sick visit 3 _____

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

Number of ICU days for sick visit 3 _____

Date of sick visit 4

Reason for sick visit 4 Unknown Condition related Condition unrelated

Sick visit 4 was a condition exacerbation Unknown Yes No

Location for sick visit 4

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

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

Name of hospital for sick visit 4 _____

ICD-9 codes for sick visit 4 known Yes No

ICD-9 codes for sick visit 4 _____

Diagnosis for sick visit 4 _____

Number of inpatient days for sick visit 4 _____

Number of ICU days for sick visit 4 _____

Date of sick visit 5

Reason for sick visit 5 Unknown Condition related Condition unrelated

Sick visit 5 was a condition exacerbation Unknown Yes No

Location for sick visit 5

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

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

Name of hospital for sick visit 5 _____

ICD-9 codes for sick visit 5 known Yes No

ICD-9 codes for sick visit 5 _____

Diagnosis for sick visit 5 _____

Number of inpatient days for sick visit 5 _____

Number of ICU days for sick visit 5 _____

Date of sick visit 6

Reason for sick visit 6 Unknown Condition related Condition unrelated

Sick visit 6 was a condition exacerbation Unknown Yes No

Location for sick visit 6

- Unknown Emergency department Retail clinic
- Primary care Specialty center Urgent care

Patient Name _____

Date Direct hospital admission OtherPatient was admitted to the hospital as a result of sick visit 6 Unknown Yes No

Name of hospital for sick visit 6 _____

ICD-9 codes for sick visit 6 known Yes No

ICD-9 codes for sick visit 6 _____

Diagnosis for sick visit 6 _____

Number of inpatient days for sick visit 6 _____

Number of ICU days for sick visit 6 _____

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

Location for sick visit 7

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

Name of hospital for sick visit 7 _____

ICD-9 codes for sick visit 7 known Yes No

ICD-9 codes for sick visit 7 _____

Diagnosis for sick visit 7 _____

Number of inpatient days for sick visit 7 _____

Number of ICU days for sick visit 7 _____

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

Location for sick visit 8

 Unknown Emergency department Primary care Specialty center Urgent care Direct hospital admission OtherPatient was admitted to the hospital as a result of sick visit 8 Unknown Yes No

Name of hospital for sick visit 8 _____

ICD-9 codes for sick visit 8 known Yes No

ICD-9 codes for sick visit 8 _____

Diagnosis for sick visit 8 _____

Number of inpatient days for sick visit 8 _____

Patient Name _____

Date

Number of ICU days for sick visit 8 _____

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

Location for sick visit 9

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

Name of hospital for sick visit 9 _____

ICD-9 codes for sick visit 9 known Yes No

ICD-9 codes for sick visit 9 _____

Diagnosis for sick visit 9 _____

Number of inpatient days for sick visit 9 _____

Number of ICU days for sick visit 9 _____

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

Location for sick visit 10

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

Name of hospital for sick visit 10 _____

ICD-9 codes for sick visit 10 known Yes No

ICD-9 codes for sick visit 10 _____

Diagnosis for sick visit 10 _____

Number of inpatient days for sick visit 10 _____

Number of ICU days for sick visit 10 _____

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

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

Surgical precautions taken

Anesthesia complications Unknown Yes No

Anesthesia complications-specify

Surgeries since last visit Unknown Yes No

Surgical procedure(s)_____

Surgery complications Unknown Yes No

Surgery complications-specify

Pregnancy

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

Please complete the pregnancy form.

Patient has become pregnant since the last visit Unknown Yes No

Please complete the pregnancy form.

Dialysis

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

Please complete the dialysis form.

Transplants

Transplant since last visit

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

Please complete the transplant form.

Other Procedures

Major medical procedure since last visit Unknown Yes No

Description of major medical procedure_____

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"/>

Comments

Visit health history comments

Patient Name _____

Date

LCHAD - Visit Findings

Visit Measurements

Measurements taken at visit

 Unknown None Blood pressure Head circumference Height/length Weight

Blood pressure-systolic _____

Blood pressure-diastolic _____

Head circumference _____

Head circumference units cm in

Height/length _____

Height/length units cm inHow height/length measured Unknown Supine Standing

Weight _____

Weight units lbs kg gm oz

Body mass index _____

Visit findings Unknown Within normal limits Evidence of peripheral neuropathy Other

Visit findings-other, specify _____

Comments

Visit findings comments

Patient Name _____

Date ***LCHAD - 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

Developmental status Typical Atypical

Severity of atypical development Unknown Mild delay Moderate delay Severe delay

Developmental milestones that were achieved in a typical order and timeframe

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

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

Referred for further developmental assessment

- Unknown Yes
 No Previously referred
 Family declined further assessments

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

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

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

Neuropsychometric evaluation performed since last visit Unknown Yes No

Overall neuropsychometric impression Above average Average Below average In progress

Patient has mental health concerns Unknown Yes No

Referred for further mental health assessment

- Unknown Yes
 No Previously referred
 Family declined further assessments

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

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

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

Behavioral concerns Unknown Yes No

Patient Name _____

Date |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

LCHAD - Visit Lab Studies

Biochemical Labs

Biochemical labs associated with this visit

- Unknown
- None
- Blood spot acylcarnitine profile
- Creatinine kinase
- Fatty acid oxidation probe assay
- Long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay
- Plasma acylcarnitine profile
- Plasma carnitine
- Plasma essential fatty acid profile
- Triglycerides
- Urine myoglobin
- Urine organic acids
- Western blot
- Other

Blood spot acylcarnitine profile

- Within normal limits Abnormal In progress Results unavailable

Blood spot acylcarnitine profile comments

Creatinine kinase Within normal limits Abnormal In progress Results unavailable

Creatinine kinase value _____

Creatinine kinase units U/L umol/L mg/dL

Creatinine kinase reference range _____

Please complete information about the fatty acid oxidation probe assay on the Intake Initial Testing form.

Please complete information about the long-chain-3-hydroxyacyl-CoA dehydrogenase enzyme assay on the Intake Initial Testing form.

Plasma acylcarnitine profile Within normal limits Abnormal In progress Results unavailable

Patient Name _____

Date

Plasma acylcarnitine profile comments

Plasma carnitine levels

 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 _____

Plasma essential fatty acid profile

 Unknown Alpha linolenic acid (C18:3w3) Docosahexaenoic acid (DHA)

 EPA (C20:5w3) Linoleic acid (C18:2w6) Triene/tetraene ratio

 Total fatty acids Other
Alpha linolenic acid Within normal limits Abnormal In progress Results unavailable

Alpha linolenic acid value _____

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

Alpha linoleic acid reference range _____

Docosahexaenoic acid (DHA) acid

 Within normal limits Abnormal In progress Results unavailable

Docosahexaenoic acid (DHA) acid value _____

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

Patient Name _____

Date

Docosahexaenoic acid (DHA) acid reference range _____

EPA Within normal limits Abnormal In progress Results unavailable

EPA value _____

EPA units nmol/L ng/mL nmol/mL

EPA reference range _____

Linoleic acid Within normal limits Abnormal In progress Results unavailable

Linoleic acid value _____

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

Linoleic acid reference range _____

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

Triene/tetraene ratio value _____

Triene/tetraene ratio reference range _____

Total fatty acids Within normal limits Abnormal In progress Results unavailable

Total fatty acids comments _____

Other plasma essential fatty acid _____

Other plasma essential fatty acid result

Within normal limits Abnormal In progress Results unavailable

Other plasma essential fatty acid value _____

Other plasma essential fatty acid units _____

Other plasma essential fatty acid reference range _____

Other plasma essential fatty acid comments

Triglyceride condition Fasting Non-fasting

Triglycerides Within normal limits Abnormal In progress Results unavailable

Triglyceride value _____

Triglyceride units mmol/L mg/dL

Triglyceride reference range _____

Urine myoglobin Within normal limits Abnormal In progress Results unavailable

Urine myoglobin value _____

Urine myoglobin units nmol/L ug/L

Urine myoglobin reference range _____

Urine organic acids Within normal limits Abnormal In progress Results unavailable

Patient Name _____

Date

Urine organic acid comments

Western blot Unknown < 20% >=20% < 50% >=50%

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 _____

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

Patient Name _____

Date

Anion gap reference range _____

Ammonia Within normal limits Abnormal In progress Results unavailable

Ammonia value _____

Ammonia units umol/L ug/dL

Ammonia reference range _____

Calcium Within normal limits Abnormal In progress Results unavailable

Calcium value _____

Calcium units mmol/L mg/dL

Calcium reference range _____

Chloride Within normal limits Abnormal In progress Results unavailable

Chloride value _____

Chloride units mmol/L mg/L

Chloride reference range _____

Carbon dioxide Within normal limits Abnormal In progress Results unavailable

Carbon dioxide value _____

Carbon dioxide units mEq/L mmol/L

Carbon dioxide reference range _____

Glucose Within normal limits Abnormal In progress Results unavailable

Glucose value _____

Glucose units mmol/L mg/dL

Glucose reference range _____

Potassium Within normal limits Abnormal In progress Results unavailable

Potassium value _____

Potassium units mEq/L mmol/L

Potassium reference range _____

Lactate dehydrogenase Within normal limits Abnormal In progress Results unavailable

Lactate dehydrogenase value _____

Lactate dehydrogenase units U/L

Lactate dehydrogenase reference range _____

Magnesium Within normal limits Abnormal In progress Results unavailable

Magnesium value _____

Magnesium units mmol/L mg/dL

Magnesium reference range _____

Sodium Within normal limits Abnormal In progress Results unavailable

Sodium value _____

Patient Name _____

Date

Sodium units mEq/L mmol/L

Sodium reference range _____

Phosphorous Within normal limits Abnormal In progress Results unavailable

Phosphorous value _____

Phosphorous units mmol/L mg/dL

Phosphorous reference range _____

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

Total Cholesterol (fasting) value _____

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

Total Cholesterol (fasting) reference range _____

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

Total Cholesterol (random) value _____

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

Total Cholesterol (random) reference range _____

Uric acid Within normal limits Abnormal In progress Results unavailable

Uric acid value _____

Uric acid units umol/L mg/dL

Uric acid reference range _____

Chemistry labs-other, specify _____

Other chemistry labs Within normal limits Abnormal In progress Results unavailable

Other chemistry lab comments

Other chemistry lab value _____

Other chemistry lab units _____

Other chemistry lab reference range _____

Hematology Labs

Hematology labs associated with this visit

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

Fibrinogen Within normal limits Abnormal In progress Results unavailable

Patient Name _____

Date

Fibrinogen value _____

Fibrinogen units umol/L mg/dL

Fibrinogen reference range _____

Hematocrit Within normal limits Abnormal In progress Results unavailable

Hematocrit value _____

Hematocrit units % Proportion of total hemoglobin

Hematocrit reference range _____

Hemoglobin Within normal limits Abnormal In progress Results unavailable

Hemoglobin value _____

Hemoglobin units g/dL g/L

Hemoglobin reference range _____

International Normalized Ratio (INR)

 Within normal limits Abnormal In progress Results unavailable

International Normalized Ratio (INR) value _____

International Normalized Ratio (INR) reference range _____

Partial Thromboplastin Time (PTT)

 Within normal limits Abnormal In progress Results unavailable

Partial Thromboplastin Time (PTT) value _____

Partial Thromboplastin Time (PTT) units seconds

Partial Thromboplastin Time (PTT) reference range _____

Prothrombin time test Within normal limits Abnormal In progress Results unavailable

Prothrombin time test value _____

Prothrombin time test units seconds

Prothrombin time test reference range _____

Peripheral blood smear Within normal limits Abnormal In progress Results unavailable

Peripheral blood smear value _____

Peripheral blood smear units platelet/RBC/WBC count

Peripheral blood smear reference range _____

Platelet count Within normal limits Abnormal In progress Results unavailable

Platelet count value _____

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

Platelet count reference range _____

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

Red blood cell count value _____

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

Patient Name _____

Date

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

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

Albumin Within normal limits Abnormal In progress Results unavailable

Albumin value _____

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

Albumin reference range _____

Alkaline phosphatase Within normal limits Abnormal In progress Results unavailable

Alkaline phosphatase value _____

Alkaline phosphatase units U/L ukat/L

Alkaline phosphatase reference range _____

Alanine aminotransferase Within normal limits Abnormal In progress Results unavailable

Alanine aminotransferase value _____

Alanine aminotransferase units U/L ukat/L

Alanine aminotransferase reference range _____

Aspartate aminotransferase Within normal limits Abnormal In progress Results unavailable

Aspartate aminotransferase value _____

Patient Name _____

Date

Aspartate aminotransferase units U/L ukat/L

Aspartate aminotransferase reference range _____

Direct bilirubin Within normal limits Abnormal In progress Results unavailable

Direct bilirubin value _____

Direct bilirubin units

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

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

Direct bilirubin reference range _____

Gamma-glutamyl transpeptidase

Within normal limits Abnormal In progress Results unavailable

Gamma-glutamyl transpeptidase value _____

Gamma-glutamyl transpeptidase units IU/L U/L

Gamma-glutamyl transpeptidase reference range _____

Globulin Within normal limits Abnormal In progress Results unavailable

Globulin value _____

Globulin units g/dL g/L

Globulin reference range _____

Prealbumin Within normal limits Abnormal In progress Results unavailable

Prealbumin value _____

Prealbumin units mg/dL mg/L

Prealbumin reference range _____

Total bilirubin Within normal limits Abnormal In progress Results unavailable

Total bilirubin value _____

Total bilirubin units umol/L mg/dL

Total bilirubin reference range _____

Total protein Within normal limits Abnormal In progress Results unavailable

Total protein value _____

Total protein units g/dL g/L

Total protein reference range _____

Liver labs-other, specify _____

Other liver labs Within normal limits Abnormal In progress Results unavailable

Other liver lab comments

Patient Name _____

Date

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

Other renal lab comments

Other renal lab value _____

Other renal lab units _____

Other renal lab reference range _____

Patient Name _____

Date **Miscellaneous Labs**

Miscellaneous labs associated with this visit

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

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

B-type natriuretic peptide value _____

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

B-type natriuretic peptide reference range _____

Creatine phosphokinase Within normal limits Abnormal In progress Results unavailable

Creatine phosphokinase value _____

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

Creatine phosphokinase reference range _____

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

C-reactive protein value _____

C-reactive protein units nmol/L mg/dL

C-reactive protein reference range _____

Erythrocyte sedimentation rate Within normal limits Abnormal In progress Results unavailable

Erythrocyte sedimentation rate value _____

Erythrocyte sedimentation rate units mm/h

Erythrocyte sedimentation rate reference range _____

Ferritin Within normal limits Abnormal In progress Results unavailable

Ferritin value _____

Ferritin units pmol/L ng/mL

Ferritin reference range _____

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

Thyroid-stimulating hormone value _____

Thyroid-stimulating hormone units mIU/L

Thyroid-stimulating hormone reference range _____

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

Thyroxine (free) value _____

Thyroxine (free) units pmol/L ng/mL

Thyroxine (free) reference range _____

Patient Name _____

Date

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

Thyroxine (total) value _____

Thyroxine (total) units pmol/L ng/mL

Thyroxine (total) reference range _____

Transferrin Within normal limits Abnormal In progress Results unavailable

Transferrin value _____

Transferrin units mg/dL g/L

Transferrin reference range _____

Troponin Within normal limits Abnormal In progress Results unavailable

Troponin value _____

Troponin units ng/mL ug/L

Troponin reference range _____

Urinalysis Within normal limits Abnormal In progress Results unavailable

Urinalysis comments

Zinc Within normal limits Abnormal In progress Results unavailable

Zinc value _____

Zinc units umol/L ug/dL

Zinc reference range _____

Miscellaneous labs-other, specify _____

Other miscellaneous labs Within normal limits Abnormal In progress Results unavailable

Other miscellaneous lab comments

Other miscellaneous lab value _____

Other miscellaneous lab units _____

Other miscellaneous lab reference range _____

Comments

Visit lab studies comments

Patient Name _____

Date

LCHAD - Visit Studies Other

Home Monitoring

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

Type of home monitoring

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

Glucose

Frequency of glucose home monitoring Unknown Only when symptomatic Routinely

Specify routine frequency _____

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

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

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

 mmol/L mg/dL

Branched Chain Amino Acids

Type of branched chain amino acid home monitoring

- Unknown Alloisoleucine Isoleucine Leucine Valine

Sample type of branched chain amino acid home monitoring

- Unknown Filter paper blood Plasma

Frequency of branched chain amino acid home monitoring

- Unknown Only when symptomatic Routinely

Specify routine frequency _____

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

Alloisoleucine

Recommended therapeutic alloisoleucine goal _____

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

Average alloisoleucine on home monitoring samples _____

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

Average percent above recommended alloisoleucine therapeutic goal _____

Patient Name _____

Date

Average percent below recommended alloisoleucine therapeutic goal _____

Isoleucine

Recommended therapeutic isoleucine goal _____

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

Average isoleucine on home monitoring samples _____

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

Average percent above recommended isoleucine therapeutic goal _____

Average percent below recommended isoleucine therapeutic goal _____

Leucine

Recommended therapeutic leucine goal _____

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

Highest leucine value on home monitoring samples _____

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

Average leucine on home monitoring samples _____

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

Average percent above recommended leucine therapeutic goal _____

Average percent below recommended leucine therapeutic goal _____

Valine

Recommended therapeutic valine goal _____

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

Average valine on home monitoring samples _____

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

Average percent above recommended valine therapeutic goal _____

Average percent below recommended valine therapeutic goal _____

PhenylalanineSample type of phenylalanine home monitoring Unknown Filter paper blood PlasmaFrequency of phenylalanine home monitoring Unknown Only when symptomatic Routinely

Specify routine frequency _____

Recommended therapeutic phenylalanine goal _____

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

Highest phenylalanine value on home monitoring samples _____

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

Average phenylalanine on home monitoring samples _____

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

Average percent above recommended phenylalanine therapeutic goal _____

Patient Name _____

Date

Average percent below recommended phenylalanine therapeutic goal _____

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

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

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

Cardiomyopathy since last visit Unknown Yes No

Cardiomyopathy has worsened/progressed since last visit Unknown Yes No

Current treatments for cardiomyopathy

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

Patient Name _____

Date

Musculoskeletal findings of note

Neurological imaging

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

Evidence of abnormal myelination on CNS imaging Unknown Yes No

Neurological findings of note

Renal/Pelvic/Genital imaging

- Unknown
- 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 _____

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

Other imaging studies date

Other imaging studies explanation

Eye Exam

Eye exam associated with this visit Unknown Yes No

Eye exam findings

- Unknown Within normal limits
- Evidence of pigmentary retinopathy Other

Eye exam findings-other, specify_____

Comments

Visit studies-other comments

Patient Name _____

Date

LCHAD - 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-carnitine

L-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 ***LCHAD - 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

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_____

Cornstarch

- Cornstarch recommended/prescribed Unknown Yes No
- Cornstarch route recommended/prescribed Unknown Feeding tube Oral
- Cornstarch dose recommended/prescribed_____
- Cornstarch dose recommended/prescribed units g
- Cornstarch frequency recommended/prescribed Unknown Once/day at bedtime PRN Other
- Cornstarch frequency recommended/prescribed-other, specify_____
- Cornstarch taken as recommended/prescribed Unknown Yes No
- Actual cornsarch dose reported_____
- Actual cornsarch dose reported units g
- Actual cornstarch frequency reported Unknown Once/day at bedtime PRN Other
- Actual cornstarch frequency reported-other, specify_____
- Reason cornstarch is not taken as recommended/prescribed_____

Method of payment for cornstarch

- 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

Patient Name _____

Date Other**MCT Supplement**MCT supplement recommended/prescribed Unknown Yes No

MCT supplement formulation recommended/prescribed

 Unknown Metabolic formula Oil Powder

Total dose of MCT (grams) provided by the recommended/prescribed supplement per day _____

MCT supplement taken as recommended/prescribed Unknown Yes No

Actual total dose of MCT (grams) taken daily _____

Reason MCT supplement is not taken as recommended/prescribed _____

Method of payment for MCT supplement

 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**Essential fatty acids**Essential fatty acids recommended/prescribed Unknown Yes NoEssential fatty acids route recommended/prescribed Unknown Feeding tube Oral

Essential fatty acids dose recommended/prescribed _____

Essential fatty acids dose recommended/prescribed units g mg

Essential fatty acids frequency recommended/prescribed

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

Essential fatty acids frequency recommended/prescribed-other, specify _____

Essential fatty acids taken as recommended/prescribed Unknown Yes No

Actual essential fatty acids dose reported _____

Actual essential fatty acids dose reported units g mg

Actual essential fatty acids frequency reported

Patient Name _____

Date

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

Actual essential fatty acids frequency reported-other, specify _____

Reason essential fatty acids are not taken as recommended/prescribed _____

Method of payment for essential fatty acids

- 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

Fat Restricted DietFat restricted diet recommended/prescribed Unknown Yes No

Percent of total kcal per 24 hours from fat recommended/prescribed in fat restricted diet _____

Fat grams per day recommended/prescribed in fat restricted diet _____

Patient/primary caregiver reports adherence to fat restricted diet Unknown Yes No

Actual frequency of adherence to fat restricted diet

- 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 fat restricted diet _____

Feeding IntervalsSpecific information given to avoid fasting Unknown Yes No

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

- 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)

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 NIGHT (in hours)

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

Comments

Nutrition comments

Patient Name _____

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

Reason for inactive status

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

Date of death

Age of death (in years) _____

Moved-specify

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

Patient Name _____

Date

Specify other non-IBEMC participating center _____

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

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

Specify reason subject initiated withdrawal, if known

Specify reason investigator initiated withdrawal, if known

Second date of study status change

Condition follow-up status Active Inactive

Reason for inactive status

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

Date of death

Age of death (in years) _____

Moved-specify

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

Specify other IBEMC participating center

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

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

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

Specify other non-IBEMC participating center _____

Specify reason for follow-up refusal

Specify reason follow-up deemed unnecessary, if known

Specify reason for study withdrawal

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

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

Specify reason subject initiated withdrawal, if known

Specify reason investigator initiated withdrawal, if known

Third date of study status change

Condition follow-up status Active Inactive

Reason for inactive status

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

Date of death

Age of death (in years) _____

Moved-specify

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

Patient Name _____

Date

- 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 withdrawl, if known

Specify reason investigator initiated withdrawal, if known

Comments

Study status comments

Patient Name _____

Date

LCHAD - Pregnancy

Patient has had one or more pregnancies Unknown Yes No

Number of pregnancies _____

Number of term pregnancies _____

Number of preterm pregnancies _____

Number of pregnancies ending in abortion/miscarriage _____

Number of pregnancies resulting in live birth _____

Patient has biological children Unknown Yes No

Number of biological children _____

First Pregnancy

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

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

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

History of infertility for first pregnancy

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

Cause(s) of infertility for first pregnancy

- Unknown Male factor Female factor
 Combined male-female factor

Cause(s) of male infertility for first pregnancy

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

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

Cause(s) of female infertility for first pregnancy

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

Patient Name _____ Date M M D D Y Y

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

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

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

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

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

Type of assisted reproductive technologies used during first pregnancy

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

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

Partner tested for patient's disorder during first pregnancy

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

Partner also affected by disorder during first pregnancy Unknown Yes No

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

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

Type of prenatal testing performed for first pregnancy Unknown Biochemical Molecular

Method of prenatal testing for first pregnancy

- Unknown
- Amniocentesis
- Chorionic villus sampling

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

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

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

First pregnancy terminated Unknown Yes No

Reason first pregnancy terminated

- Elective due to fetus affected with disorder

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

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

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

- 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 _____

First pregnancy complicated by maternal HELLP syndrome Unknown Yes No

First pregnancy complicated by acute fatty liver of pregnancy (AFLP) Unknown Yes No

Number of documented episodes of hypoglycemia during first pregnancy

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

Lowest maternal blood glucose value documented during first pregnancy _____

Lowest maternal blood glucose value documented during first pregnancy units mmol/L mg/dL

Number of documented episodes of rhabdomyolysis during first pregnancy

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

Highest maternal CK value documented during first pregnancy _____

Highest maternal CK value documented during first pregnancy units U/L umol/L mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during first pregnancy

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

Longest permitted fasting interval (in hours) during the NIGHT recommended during first pregnancy

- Unknown 1 2 3 4 5 6
 7 8 9 10 11 13

L-carnitine supplementation used on a regular basis during first pregnancy

- Unknown Yes No
 L-carnitine not prescribed

Patient adhered to a fat-restricted diet during first pregnancy

- Unknown Yes No
 Fat-restriction not prescribed

Patient took a metabolic formula regularly during first pregnancy

- Unknown Yes
 No Metabolic formula not prescribed

Evidence of cardiac hypertrophy during first pregnancy

- Unknown Yes
 No Echocardiogram not performed during pregnancy

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

Evidence of any cardiac dysfunction during first pregnancy

- Unknown Yes
- No Cardiac testing not performed during pregnancy

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

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

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

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

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

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

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

Abnormal patient lab results during first delivery

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

Patient Name _____

Date

Abnormal patient lab results during first delivery-other, specify

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

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

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

Patient death during or shortly after first delivery Unknown Yes No

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

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

Method of first delivery

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

Maternal inpatient days post- first delivery_____

Live delivery of first newborn Unknown Yes No

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

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

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

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

Patient Name _____

Date

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

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

Birth measurements for first newborn Unknown Head circumference Length Weight

Birth head circumference _____

Birth head circumference units cm in

Birth length _____

Birth length units cm in

Birth weight _____

Birth weight units lbs kg gm oz

APGAR score at 1 minute for first newborn

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

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

APGAR score at 5 minutes for first newborn

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

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

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

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

Routine newborn screening done for first newborn

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

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

Describe abnormal newborn screen result for first newborn

Reason routine newborn screening was not done for first newborn

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

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

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

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

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

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

- Unknown
- Yes
- No

Abnormal newborn exam findings for first newborn

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

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

Patient Name _____

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

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

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

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

History of infertility for second pregnancy

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

Cause(s) of infertility for second pregnancy

- Unknown Male factor Female factor
 Combined male-female factor

Cause(s) of male infertility for second pregnancy

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

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

Cause(s) of female infertility for second pregnancy

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

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

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

Patient Name _____

Date

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

- Unknown None
 Assisted reproductive technology Fertility drugs
 Surgery

Type of assisted reproductive technologies used during second pregnancy

- Assisted hatching ICSI IVF
 Surgical sperm aspiration Other

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

Partner tested for patient's disorder during second pregnancy

- Unknown Yes-biochemical Yes-molecular No

Partner also affected by disorder during second pregnancy Unknown Yes No

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

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

Type of prenatal testing performed for second pregnancy Unknown Biochemical Molecular

Method of prenatal testing for second pregnancy

- Unknown Amniocentesis Chorionic villus sampling

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

- Unknown None
 Fetal echocardiogram Increased frequency prenatal ultrasounds
 Other

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

Second pregnancy terminated Unknown Yes No

Reason second pregnancy terminated

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

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

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

Patient Name _____

Date Prenatal care received during second pregnancy Unknown Yes No

Weeks gestation prenatal care started during second pregnancy _____

Treatment prescribed for patient's disorder during second pregnancy

 Unknown Metabolic diet Medications Biochemical lab monitoring Avoidance of fasting Other

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

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

Patient in good metabolic condition during second trimester of second pregnancy

 Unknown Pregnancy not sustained to second trimester Yes No

Patient in good metabolic condition during third trimester of second pregnancy

 Unknown Pregnancy not sustained to third trimester Yes No

Number of outpatient metabolic visits for patient during second pregnancy _____

Number of ED visits for management of disorder during second pregnancy

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

Number of hospitalizations for management of disorder during second pregnancy

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

Complications during second pregnancy

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

Complications related to disorder during second pregnancy- specify

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

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

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

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

Complications during second pregnancy-other, specify _____

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

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

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

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

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

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

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

Laboratory studies done on patient during second pregnancy

- Unknown Ammonia - Abn high

Patient Name _____

Date

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

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

Additional interventions required during second pregnancy due to this metabolic condition

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

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

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

Second pregnancy complicated by maternal HELLP syndrome Unknown Yes NoSecond pregnancy complicated by acute fatty liver of pregnancy (AFLP) Unknown Yes No

Patient Name _____

Date

Number of documented episodes of hypoglycemia during second pregnancy

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

Lowest maternal blood glucose value documented during second pregnancy _____

Lowest maternal blood glucose value documented during second pregnancy units mmol/L mg/dL

Number of documented episodes of rhabdomyolysis during second pregnancy

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

Highest maternal CK value documented during second pregnancy _____

Highest maternal CK value documented during second pregnancy units U/L umol/L mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during second pregnancy

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

Longest permitted fasting interval (in hours) during the NIGHT recommended during second pregnancy

- Unknown 1 2 3 4 5 6
 7 8 9 10 11 13

L-carnitine supplementation used on a regular basis during second pregnancy

- Unknown Yes No
 L-carnitine not prescribed

Patient adhered to a fat-restricted diet during second pregnancy

- Unknown Yes No
 Fat-restriction not prescribed

Patient took a metabolic formula regularly during second pregnancy

- Unknown Yes
 No Metabolic formula not prescribed

Evidence of cardiac hypertrophy during second pregnancy

- Unknown Yes
 No Echocardiogram not performed during pregnancy

Evidence of any cardiac dysfunction during second pregnancy

- Unknown Yes
 No Cardiac testing not performed during 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

Patient Name _____

Date

- Letter to OB/MFM specialist
- Planned C-section
- Referral for high risk OB management
- Other

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

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

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

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

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

Abnormal patient lab results during second delivery

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

Abnormal patient lab results during second delivery-other, specify

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

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

Patient Name _____

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

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

[Empty text box for maternal interventions]

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)

[Empty text box for acute health concerns]

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

[Empty text box for lab tests and results]

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

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 second newborn

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

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

APGAR score at 5 minutes for second newborn

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

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

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

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

Routine newborn screening done for second newborn

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

Describe abnormal newborn screen result for second newborn

Reason routine newborn screening was not done for second newborn

- 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 second newborn-other, specify

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

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

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

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

- Unknown Yes No

Abnormal newborn exam findings for second newborn

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

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

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

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

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

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

Patient Name _____

Date

13-24 25-36 >36

History of infertility for third pregnancy

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

Cause(s) of infertility for third pregnancy

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

Cause(s) of male infertility for third pregnancy

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

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

Cause(s) of female infertility for third pregnancy

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

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

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

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

- Unknown None
- Assisted reproductive technology Fertility drugs
- Surgery

Type of assisted reproductive technologies used during third pregnancy

- Assisted hatching ICSI IVF
- Surgical sperm aspiration Other

Patient Name _____

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

Method of prenatal testing for third pregnancy

 Unknown Amniocentesis Chorionic villus sampling

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

 Unknown None Fetal echocardiogram Increased frequency prenatal ultrasounds OtherAdditional prenatal testing performed on fetus as a result of parent's disorder for third pregnancy-other,
specify _____Third pregnancy terminated Unknown Yes No

Reason third pregnancy terminated

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

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

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

Prenatal care received during third pregnancy Unknown Yes No

Weeks gestation prenatal care started during third pregnancy _____

Treatment prescribed for patient's disorder during third pregnancy

 Unknown Metabolic diet Medications
 Biochemical lab monitoring Avoidance of fasting Other

Patient Name _____

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

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

--

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

Patient in good metabolic condition during second trimester of third pregnancy

UnknownPregnancy not sustained to second trimesterYesNo

Patient in good metabolic condition during third trimester of third pregnancy

UnknownPregnancy not sustained to third trimesterYesNo

Number of outpatient metabolic visits for patient during third pregnancy _____

Number of ED visits for management of disorder during third pregnancy

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

Number of hospitalizations for management of disorder during third pregnancy

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

Complications during third pregnancy

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

Complications related to disorder during third pregnancy- specify

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

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

UnknownAdvanced maternal age (35+ years of age)Ectopic pregnancyGestational diabetesGroup B strep

Patient Name _____

Date

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

Complications during third pregnancy-other, specify _____

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

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

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

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

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

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

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

Laboratory studies done on patient during third pregnancy

- | | |
|--|---|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Ammonia - Abn high |
| <input type="checkbox"/> Ammonia - WNL | <input type="checkbox"/> Blood glucose - Abn low |
| <input type="checkbox"/> Blood glucose - WNL | <input type="checkbox"/> Blood Glucose - Abn high |
| <input type="checkbox"/> BNP - Abn high | <input type="checkbox"/> BNP - WNL |
| <input type="checkbox"/> CBC - Abn | <input type="checkbox"/> CBC - WNL |
| <input type="checkbox"/> C02 - Abn low | <input type="checkbox"/> C02 - WNL |

Patient Name _____

Date

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

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

Additional interventions required during third pregnancy due to this metabolic condition

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

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

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

Third pregnancy complicated by maternal HELLP syndrome Unknown Yes No

Third pregnancy complicated by acute fatty liver of pregnancy (AFLP) Unknown Yes No

Number of documented episodes of hypoglycemia during third pregnancy

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

Lowest maternal blood glucose value documented during third pregnancy _____

Lowest maternal blood glucose value documented during third pregnancy units mmol/L mg/dL

Patient Name _____

Date |||||

Number of documented episodes of rhabdomyolysis during third pregnancy

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

Highest maternal CK value documented during third pregnancy _____

Highest maternal CK value documented during third pregnancy units U/L umol/L mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during third pregnancy

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

Longest permitted fasting interval (in hours) during the NIGHT recommended during third pregnancy

- Unknown 1 2 3 4 5 6
7 8 9 10 11 13

L-carnitine supplementation used on a regular basis during third pregnancy

- Unknown Yes No
L-carnitine not prescribed

Patient adhered to a fat-restricted diet during third pregnancy

- Unknown Yes No
Fat-restriction not prescribed

Patient took a metabolic formula regularly during third pregnancy

- Unknown Yes
No Metabolic formula not prescribed

Evidence of cardiac hypertrophy during third pregnancy

- Unknown Yes
No Echocardiogram not performed during pregnancy

Evidence of any cardiac dysfunction during third pregnancy

- Unknown Yes
No Cardiac testing not performed during 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

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

Cause(s) of female infertility for fourth pregnancy

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

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

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

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

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

Type of assisted reproductive technologies used during fourth pregnancy

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

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

Partner tested for patient's disorder during fourth pregnancy

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

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

Method of prenatal testing for fourth pregnancy

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

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

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

Patient Name _____

Date

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

Fourth pregnancy terminated Unknown Yes No

Reason fourth pregnancy terminated

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

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

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

Prenatal care received during fourth pregnancy Unknown Yes No

Weeks gestation prenatal care started during fourth pregnancy _____

Treatment prescribed for patient's disorder during fourth pregnancy

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

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

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

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

Patient in good metabolic condition during second trimester of fourth pregnancy

- Unknown Pregnancy not sustained to second trimester
 Yes No

Patient in good metabolic condition during third trimester of fourth pregnancy

- Unknown Pregnancy not sustained to third trimester
 Yes No

Number of outpatient metabolic visits for patient during fourth pregnancy _____

Number of ED visits for management of disorder during fourth pregnancy

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

Number of hospitalizations for management of disorder during fourth pregnancy

Patient Name _____

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

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

Patient Name _____

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

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

Additional interventions required during fourth pregnancy due to this metabolic condition

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

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

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

Fourth pregnancy complicated by maternal HELLP syndrome Unknown Yes NoFourth pregnancy complicated by acute fatty liver of pregnancy (AFLP) Unknown Yes No

Number of documented episodes of hypoglycemia during fourth pregnancy

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

Lowest maternal blood glucose value documented during fourth pregnancy _____

Lowest maternal blood glucose value documented during fourth pregnancy units mmol/L mg/dL

Number of documented episodes of rhabdomyolysis during fourth pregnancy

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

Highest maternal CK value documented during fourth pregnancy _____

Highest maternal CK value documented during fourth pregnancy units U/L umol/L mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during fourth pregnancy

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

Longest permitted fasting interval (in hours) during the NIGHT recommended during fourth pregnancy

- Unknown 1 2 3 4 5 6
 7 8 9 10 11 13

L-carnitine supplementation used on a regular basis during fourth pregnancy

- Unknown Yes No
 L-carnitine not prescribed

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

Patient adhered to a fat-restricted diet during fourth pregnancy

- Unknown Yes No
- Fat-restriction not prescribed

Patient took a metabolic formula regularly during fourth pregnancy

- Unknown Yes
- No Metabolic formula not prescribed

Evidence of cardiac hypertrophy during fourth pregnancy

- Unknown Yes
- No Echocardiogram not performed during pregnancy

Evidence of any cardiac dysfunction during fourth pregnancy

- Unknown Yes
- No Cardiac testing not performed during pregnancy

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

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

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

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

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

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

Patient Name _____

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

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

[Empty text box for acute health concerns]

Abnormal patient lab results during fourth delivery

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

Abnormal patient lab results during fourth delivery-other, specify

[Empty text box for abnormal lab results other]

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

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

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

[Empty text box for additional maternal interventions other]

Patient death during or shortly after fourth delivery Unknown Yes No

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

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

Method of fourth delivery

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

Maternal inpatient days post- fourth delivery_____

Live delivery of fourth newborn Unknown Yes No

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

[Empty text box for acute health concerns for newborn]

Patient Name _____

Date

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

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

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

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

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

Birth measurements for fourth newborn Unknown Head circumference Length Weight

Birth head circumference _____

Birth head circumference units cm in

Birth length _____

Birth length units cm in

Birth weight _____

Birth weight units lbs kg gm oz

APGAR score at 1 minute for fourth newborn

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

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

APGAR score at 5 minutes for fourth newborn

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

Patient Name _____

Date

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

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

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

Routine newborn screening done for fourth newborn

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

Describe abnormal newborn screen result for fourth newborn

Reason routine newborn screening was not done for fourth newborn

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

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

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

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

Patient Name _____

Date

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

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

Unknown Yes No

Abnormal newborn exam findings for fourth newborn

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

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

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

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

Fourth newborn currently alive Unknown Yes No

Fifth Pregnancy

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

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

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

History of infertility for fifth pregnancy

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

Cause(s) of infertility for fifth pregnancy

Unknown Male factor Female factor
 Combined male-female factor

Cause(s) of male infertility for fifth pregnancy

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

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

- Tobacco use
- Other

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

Cause(s) of female infertility for fifth pregnancy

- 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

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

Type of assisted reproductive technologies used during fifth pregnancy

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

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

Partner tested for patient's disorder during fifth pregnancy

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

Partner also affected by disorder during fifth pregnancy Unknown Yes No

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

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

Type of prenatal testing performed for fifth pregnancy Unknown Biochemical Molecular

Method of prenatal testing for fifth pregnancy

- Unknown
- Amniocentesis
- Chorionic villus sampling

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

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

Patient Name _____

Date

Other

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

Fifth pregnancy terminated Unknown Yes No

Reason fifth pregnancy terminated

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

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

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

Prenatal care received during fifth pregnancy Unknown Yes No

Weeks gestation prenatal care started during fifth pregnancy _____

Treatment prescribed for patient's disorder during fifth pregnancy

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

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

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

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

Patient in good metabolic condition during second trimester of fifth pregnancy

- Unknown Pregnancy not sustained to second trimester
- Yes No

Patient in good metabolic condition during third trimester of fifth pregnancy

- Unknown Pregnancy not sustained to third trimester
- Yes No

Number of outpatient metabolic visits for patient during fifth pregnancy _____

Number of ED visits for management of disorder during fifth pregnancy

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

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

Number of hospitalizations for management of disorder during fifth pregnancy

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

Complications during fifth pregnancy

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

Complications related to disorder during fifth pregnancy- specify

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

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

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

Complications during fifth pregnancy-other, specify _____

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

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

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

- Unknown 0 1 2 3 4 5

Patient Name _____

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

6
 7
 8
 9
 10
 >10

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

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

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

Laboratory studies done on patient during fifth pregnancy

- | | |
|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Ammonia - Abn high |
| <input type="checkbox"/> Ammonia - WNL | <input type="checkbox"/> Blood glucose - Abn low |
| <input type="checkbox"/> Blood glucose - WNL | <input type="checkbox"/> Blood Glucose - Abn high |
| <input type="checkbox"/> BNP - Abn high | <input type="checkbox"/> BNP - WNL |
| <input type="checkbox"/> CBC - Abn | <input type="checkbox"/> CBC - WNL |
| <input type="checkbox"/> CO 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 |M|M| |D|D| |Y|Y|

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

Additional interventions required during fifth pregnancy due to this metabolic condition

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

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

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

Fifth pregnancy complicated by maternal HELLP syndrome Unknown Yes NoFifth pregnancy complicated by acute fatty liver of pregnancy (AFLP) Unknown Yes No

Number of documented episodes of hypoglycemia during fifth pregnancy

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

Lowest maternal blood glucose value documented during fifth pregnancy _____

Lowest maternal blood glucose value documented during fifth pregnancy units mmol/L mg/dL

Number of documented episodes of rhabdomyolysis during fifth pregnancy

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

Highest maternal CK value documented during fifth pregnancy _____

Highest maternal CK value documented during fifth pregnancy units U/L umol/L mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during fifth pregnancy

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

Longest permitted fasting interval (in hours) during the NIGHT recommended during fifth pregnancy

- Unknown 1 2 3 4 5 6
 7 8 9 10 11 13

L-carnitine supplementation used on a regular basis during fifth pregnancy

- Unknown Yes No
 L-carnitine not prescribed

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

Patient adhered to a fat-restricted diet during fifth pregnancy

- Unknown Yes No
- Fat-restriction not prescribed

Patient took a metabolic formula regularly during fifth pregnancy

- Unknown Yes
- No Metabolic formula not prescribed

Evidence of cardiac hypertrophy during fifth pregnancy

- Unknown Yes
- No Echocardiogram not performed during pregnancy

Evidence of any cardiac dysfunction during fifth pregnancy

- Unknown Yes
- No Cardiac testing not performed during pregnancy

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

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

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

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

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

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

Patient Name _____

Date

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

Abnormal patient lab results during fifth delivery

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

Abnormal patient lab results during fifth delivery-other, specify

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

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

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

Patient death during or shortly after fifth delivery Unknown Yes No

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

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

Method of fifth delivery

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

Maternal inpatient days post- fifth delivery _____

Live delivery of fifth newborn Unknown Yes No

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

Patient Name _____

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

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

[Empty text box for lab tests and results]

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

[Empty text box for health concerns]

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

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

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

[Empty text box for additional interventions]

Birth measurements for fifth newborn Unknown Head circumference Length Weight

Birth head circumference _____

Birth head circumference units cm in

Birth length _____

Birth length units cm in

Birth weight _____

Birth weight units lbs kg gm oz

APGAR score at 1 minute for fifth newborn

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

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

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

APGAR score at 5 minutes for fifth newborn

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

Patient Name _____

Date

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

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

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

Routine newborn screening done for fifth newborn

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

Describe abnormal newborn screen result for fifth newborn

Reason routine newborn screening was not done for fifth newborn

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

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

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

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

Patient Name _____

Date

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

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

Unknown Yes No

Abnormal newborn exam findings for fifth newborn

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

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

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

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

Fifth newborn currently alive Unknown Yes No

Sixth Pregnancy

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

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

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

History of infertility for sixth pregnancy

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

Cause(s) of infertility for sixth pregnancy

Unknown Male factor Female factor
 Combined male-female factor

Cause(s) of male infertility for sixth pregnancy

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

Patient Name _____

Date

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

Cause(s) of female infertility for sixth pregnancy

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

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

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

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

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

Type of assisted reproductive technologies used during sixth pregnancy

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

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

Partner tested for patient's disorder during sixth pregnancy

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

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

Method of prenatal testing for sixth pregnancy

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

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

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

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

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

Sixth pregnancy terminated Unknown Yes No

Reason sixth pregnancy terminated

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

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

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

Prenatal care received during sixth pregnancy Unknown Yes No

Weeks gestation prenatal care started during sixth pregnancy _____

Treatment prescribed for patient's disorder during sixth pregnancy

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

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

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

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

Patient in good metabolic condition during second trimester of sixth pregnancy

- Unknown Pregnancy not sustained to second trimester
- Yes No

Patient in good metabolic condition during third trimester of sixth pregnancy

- Unknown Pregnancy not sustained to third trimester
- Yes No

Number of outpatient metabolic visits for patient during sixth pregnancy _____

Number of ED visits for management of disorder during sixth pregnancy

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

Number of hospitalizations for management of disorder during sixth pregnancy

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

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

Complications during sixth pregnancy

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

Complications related to disorder during sixth pregnancy- specify

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

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

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

Complications during sixth pregnancy-other, specify_____

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

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

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

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

Patient Name _____

Date

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

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

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

Laboratory studies done on patient during sixth pregnancy

- | | |
|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Ammonia - Abn high |
| <input type="checkbox"/> Ammonia - WNL | <input type="checkbox"/> Blood glucose - Abn low |
| <input type="checkbox"/> Blood glucose - WNL | <input type="checkbox"/> Blood Glucose - Abn high |
| <input type="checkbox"/> BNP - Abn high | <input type="checkbox"/> BNP - WNL |
| <input type="checkbox"/> CBC - Abn | <input type="checkbox"/> CBC - WNL |
| <input type="checkbox"/> 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 |M|M| |D|D| |Y|Y|

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

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

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

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

Sixth pregnancy complicated by maternal HELLP syndrome Unknown Yes NoSixth pregnancy complicated by acute fatty liver of pregnancy (AFLP) Unknown Yes No

Number of documented episodes of hypoglycemia during sixth pregnancy

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

Lowest maternal blood glucose value documented during sixth pregnancy _____

Lowest maternal blood glucose value documented during sixth pregnancy units mmol/L mg/dL

Number of documented episodes of rhabdomyolysis during sixth pregnancy

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

Highest maternal CK value documented during sixth pregnancy _____

Highest maternal CK value documented during sixth pregnancy units U/L umol/L mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during sixth pregnancy

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

Longest permitted fasting interval (in hours) during the NIGHT recommended during sixth pregnancy

- Unknown 1 2 3 4 5 6
 7 8 9 10 11 13

L-carnitine supplementation used on a regular basis during sixth pregnancy

- Unknown Yes No
 L-carnitine not prescribed

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

Patient adhered to a fat-restricted diet during sixth pregnancy

- Unknown Yes No
- Fat-restriction not prescribed

Patient took a metabolic formula regularly during sixth pregnancy

- Unknown Yes
- No Metabolic formula not prescribed

Evidence of cardiac hypertrophy during sixth pregnancy

- Unknown Yes
- No Echocardiogram not performed during pregnancy

Evidence of any cardiac dysfunction during sixth pregnancy

- Unknown Yes
- No Cardiac testing not performed during 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

Patient Name _____

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

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

[Empty text box for acute health concerns]

Abnormal patient lab results during sixth delivery

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

Abnormal patient lab results during sixth delivery-other, specify

[Empty text box for abnormal lab results other]

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 other]

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 for sixth newborn]

Patient Name _____

Date

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

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

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

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

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

Birth measurements for sixth newborn Unknown Head circumference Length Weight

Birth head circumference _____

Birth head circumference units cm in

Birth length _____

Birth length units cm in

Birth weight _____

Birth weight units lbs kg gm oz

APGAR score at 1 minute for sixth newborn

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

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

APGAR score at 5 minutes for sixth newborn

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

Patient Name _____

Date |||||

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

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

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

Routine newborn screening done for sixth newborn

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

Describe abnormal newborn screen result for sixth newborn

Reason routine newborn screening was not done for sixth newborn

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

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

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

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

Patient Name _____

Date

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

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

Unknown Yes No

Abnormal newborn exam findings for sixth newborn

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

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

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

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

Sixth newborn currently alive Unknown Yes No

Seventh Pregnancy

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

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

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

History of infertility for seventh pregnancy

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

Cause(s) of infertility for seventh pregnancy

Unknown Male factor Female factor
 Combined male-female factor

Cause(s) of male infertility for seventh pregnancy

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

Patient Name _____

Date

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

Cause(s) of female infertility for seventh pregnancy

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

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

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

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

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

Type of assisted reproductive technologies used during seventh pregnancy

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

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

Partner tested for patient's disorder during seventh pregnancy

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

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

Method of prenatal testing for seventh pregnancy

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

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

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

Patient Name _____

Date

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

Seventh pregnancy terminated Unknown Yes No

Reason seventh pregnancy terminated

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

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

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

Prenatal care received during seventh pregnancy Unknown Yes No

Weeks gestation prenatal care started during seventh pregnancy _____

Treatment prescribed for patient's disorder during seventh pregnancy

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

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

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

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

Patient in good metabolic condition during second trimester of seventh pregnancy

- Unknown Pregnancy not sustained to second trimester
- Yes No

Patient in good metabolic condition during third trimester of seventh pregnancy

- Unknown Pregnancy not sustained to third trimester
- Yes No

Number of outpatient metabolic visits for patient during seventh pregnancy _____

Number of ED visits for management of disorder during seventh pregnancy

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

Number of hospitalizations for management of disorder during seventh pregnancy

Patient Name _____

Date

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

Complications during seventh pregnancy

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

Complications related to disorder during seventh pregnancy- specify

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

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

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

Complications during seventh pregnancy-other, specify _____

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

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

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

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

Patient Name _____

Date | |

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

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

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

Laboratory studies done on patient during seventh pregnancy

- | | |
|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Ammonia - Abn high |
| <input type="checkbox"/> Ammonia - WNL | <input type="checkbox"/> Blood glucose - Abn low |
| <input type="checkbox"/> Blood glucose - WNL | <input type="checkbox"/> Blood Glucose - Abn high |
| <input type="checkbox"/> BNP - Abn high | <input type="checkbox"/> BNP - WNL |
| <input type="checkbox"/> CBC - Abn | <input type="checkbox"/> CBC - WNL |
| <input type="checkbox"/> CO 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 |M|M| |D|D| |Y|Y|

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

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

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

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

Seventh pregnancy complicated by maternal HELLP syndrome Unknown Yes NoSeventh pregnancy complicated by acute fatty liver of pregnancy (AFLP) Unknown Yes No

Number of documented episodes of hypoglycemia during seventh pregnancy

- | | | | | | | |
|-------------------------------|-------------------------|-------------------------|-------------------------|--------------------------|---------------------------|-------------------------|
| <input type="radio"/> Unknown | <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 | |

Lowest maternal blood glucose value documented during seventh pregnancy _____

Lowest maternal blood glucose value documented during seventh pregnancy units mmol/L mg/dL

Number of documented episodes of rhabdomyolysis during seventh pregnancy

- | | | | | | | |
|-------------------------------|-------------------------|-------------------------|-------------------------|--------------------------|---------------------------|-------------------------|
| <input type="radio"/> Unknown | <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 | |

Highest maternal CK value documented during seventh pregnancy _____

Highest maternal CK value documented during seventh pregnancy units U/L umol/L mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during seventh pregnancy

- | | | | | | | |
|-------------------------------|-------------------------|-------------------------|--------------------------|--------------------------|--------------------------|-------------------------|
| <input type="radio"/> Unknown | <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 | |

Longest permitted fasting interval (in hours) during the NIGHT recommended during seventh pregnancy

- | | | | | | | |
|-------------------------------|-------------------------|-------------------------|--------------------------|--------------------------|--------------------------|-------------------------|
| <input type="radio"/> Unknown | <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"/> 13 | |

L-carnitine supplementation used on a regular basis during seventh pregnancy

- | | | |
|--|---------------------------|--------------------------|
| <input type="radio"/> Unknown | <input type="radio"/> Yes | <input type="radio"/> No |
| <input type="radio"/> L-carnitine not prescribed | | |

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

Patient adhered to a fat-restricted diet during seventh pregnancy

- Unknown Yes No
- Fat-restriction not prescribed

Patient took a metabolic formula regularly during seventh pregnancy

- Unknown Yes
- No Metabolic formula not prescribed

Evidence of cardiac hypertrophy during seventh pregnancy

- Unknown Yes
- No Echocardiogram not performed during pregnancy

Evidence of any cardiac dysfunction during seventh pregnancy

- Unknown Yes
- No Cardiac testing not performed during pregnancy

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

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

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

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

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

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

Patient Name _____

Date | |

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

Abnormal patient lab results during seventh delivery

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

Abnormal patient lab results during seventh delivery-other, specify

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

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

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

Patient death during or shortly after seventh delivery Unknown Yes No

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

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

Method of seventh delivery

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

Maternal inpatient days post- seventh delivery_____

Live delivery of seventh newborn Unknown Yes No

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

Patient Name _____

Date

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

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

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

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

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

Birth measurements for seventh newborn Unknown Head circumference Length Weight

Birth head circumference _____

Birth head circumference units cm in

Birth length _____

Birth length units cm in

Birth weight _____

Birth weight units lbs kg gm oz

APGAR score at 1 minute for seventh newborn

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

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

APGAR score at 5 minutes for seventh newborn

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

Patient Name _____

Date |||||

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

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

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

Routine newborn screening done for seventh newborn

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

Describe abnormal newborn screen result for seventh newborn

Reason routine newborn screening was not done for seventh newborn

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

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

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

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

Patient Name _____

Date

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

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

Unknown Yes No

Abnormal newborn exam findings for seventh newborn

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

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

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

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

Seventh newborn currently alive Unknown Yes No

Eighth Pregnancy

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

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

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

History of infertility for eighth pregnancy

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

Cause(s) of infertility for eighth pregnancy

Unknown Male factor Female factor
 Combined male-female factor

Cause(s) of male infertility for eighth pregnancy

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

Patient Name _____

Date

- Tobacco use Other

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

Cause(s) of female infertility for eighth pregnancy

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

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

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

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

- Unknown None
 Assisted reproductive technology Fertility drugs
 Surgery

Type of assisted reproductive technologies used during eighth pregnancy

- Assisted hatching ICSI IVF
 Surgical sperm aspiration Other

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

Partner tested for patient's disorder during eighth pregnancy

- Unknown Yes-biochemical Yes-molecular No

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

Method of prenatal testing for eighth pregnancy

- Unknown Amniocentesis Chorionic villus sampling

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

- Unknown None
 Fetal echocardiogram Increased frequency prenatal ultrasounds

Patient Name _____

Date

Other

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

Eighth pregnancy terminated Unknown Yes No

Reason eighth pregnancy terminated

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

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

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

Prenatal care received during eighth pregnancy Unknown Yes No

Weeks gestation prenatal care started during eighth pregnancy _____

Treatment prescribed for patient's disorder during eighth pregnancy

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

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

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

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

Patient in good metabolic condition during second trimester of eighth pregnancy

- Unknown Pregnancy not sustained to second trimester
- Yes No

Patient in good metabolic condition during third trimester of eighth pregnancy

- Unknown Pregnancy not sustained to third trimester
- Yes No

Number of outpatient metabolic visits for patient during eighth pregnancy _____

Number of ED visits for management of disorder during eighth pregnancy

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

Patient Name _____

Date

Number of hospitalizations for management of disorder during eighth pregnancy

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

Complications during eighth pregnancy

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

Complications related to disorder during eighth pregnancy- specify

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

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

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

Complications during eighth pregnancy-other, specify _____

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

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

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

- Unknown 0 1 2 3 4 5

Patient Name _____

Date

6
 7
 8
 9
 10
 >10

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

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

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

Laboratory studies done on patient during eighth pregnancy

- | | |
|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Ammonia - Abn high |
| <input type="checkbox"/> Ammonia - WNL | <input type="checkbox"/> Blood glucose - Abn low |
| <input type="checkbox"/> Blood glucose - WNL | <input type="checkbox"/> Blood Glucose - Abn high |
| <input type="checkbox"/> BNP - Abn high | <input type="checkbox"/> BNP - WNL |
| <input type="checkbox"/> CBC - Abn | <input type="checkbox"/> CBC - WNL |
| <input type="checkbox"/> 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 eighth pregnancy: describe test(s) and result(s)

Additional interventions required during eighth pregnancy due to this metabolic condition

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

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

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

Eighth pregnancy complicated by maternal HELLP syndrome Unknown Yes NoEighth pregnancy complicated by acute fatty liver of pregnancy (AFLP) Unknown Yes No

Number of documented episodes of hypoglycemia during eighth pregnancy

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

Lowest maternal blood glucose value documented during eighth pregnancy _____

Lowest maternal blood glucose value documented during eighth pregnancy units mmol/L mg/dL

Number of documented episodes of rhabdomyolysis during eighth pregnancy

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

Highest maternal CK value documented during eighth pregnancy _____

Highest maternal CK value documented during eighth pregnancy units U/L umol/L mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during eighth pregnancy

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

Longest permitted fasting interval (in hours) during the NIGHT recommended during eighth pregnancy

- Unknown 1 2 3 4 5 6
 7 8 9 10 11 13

L-carnitine supplementation used on a regular basis during eighth pregnancy

- Unknown Yes No
 L-carnitine not prescribed

Patient Name _____

Date

Patient adhered to a fat-restricted diet during eighth pregnancy

- Unknown
- Yes
- No
- Fat-restriction not prescribed

Patient took a metabolic formula regularly during eighth pregnancy

- Unknown
- Yes
- No
- Metabolic formula not prescribed

Evidence of cardiac hypertrophy during eighth pregnancy

- Unknown
- Yes
- No
- Echocardiogram not performed during pregnancy

Evidence of any cardiac dysfunction during eighth pregnancy

- Unknown
- Yes
- No
- Cardiac testing not performed during pregnancy

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

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

Patient Name _____

Date

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

Abnormal patient lab results during eighth delivery

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

Abnormal patient lab results during eighth delivery-other, specify

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

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

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

Patient death during or shortly after eighth delivery Unknown Yes No

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

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

Method of eighth delivery

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

Maternal inpatient days post- eighth delivery_____

Live delivery of eighth newborn Unknown Yes No

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

Patient Name _____

Date

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

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

Patient Name _____

Date

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
 Other

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

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

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

Patient Name _____

Date

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

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

Unknown Yes No

Abnormal newborn exam findings for eighth newborn

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

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

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

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

Eighth newborn currently alive Unknown Yes No

Current Pregnancy

Patient is pregnant Unknown Yes No

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

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

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

History of infertility for current pregnancy

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

Cause(s) of infertility for current pregnancy

Unknown Male factor Female factor
 Combined male-female factor

Cause(s) of male infertility for current pregnancy

Unknown Abnormal sperm production or function
 Age Cancer/cancer treatment related

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

- Environmental
- Tobacco use
- Problems with sperm delivery
- Other

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

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

Type of assisted reproductive technologies used during current pregnancy

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

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

Partner tested for patient's disorder during current pregnancy

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

Partner also affected by disorder during current pregnancy Unknown Yes No

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

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

Type of prenatal testing performed for current pregnancy Unknown Biochemical Molecular

Method of prenatal testing for current pregnancy

- Unknown
- Amniocentesis
- Chorionic villus sampling

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

- Unknown
- None

Patient Name _____

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

Fetal echocardiogram

Increased frequency prenatal ultrasounds

Other

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

Current pregnancy terminated Unknown Yes No

Reason current pregnancy terminated

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

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

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

Prenatal care received during current pregnancy Unknown Yes No

Weeks gestation prenatal care started during current pregnancy _____

Treatment prescribed for patient's disorder during current pregnancy

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

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

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

Patient in good metabolic condition during first trimester of current pregnancy

- Unknown Yes No

Patient in good metabolic condition during second trimester of current pregnancy

- Unknown Pregnancy not sustained to second trimester
- Yes No

Patient in good metabolic condition during third trimester of current pregnancy

- Unknown Pregnancy not sustained to third trimester
- Yes No

Number of outpatient metabolic visits for patient during current pregnancy _____

Number of ED visits for management of disorder during current pregnancy _____

Patient Name _____

Date

- 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
 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

Patient Name _____

Date

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 |
| <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 current pregnancy: describe test(s) and result(s)

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

Total maternal weight gain to date (in kg) during current pregnancy _____

Current pregnancy complicated by maternal HELLP syndrome Unknown Yes No

Current pregnancy complicated by acute fatty liver of pregnancy (AFLP) Unknown Yes No

Number of documented episodes of hypoglycemia during current pregnancy

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

Lowest maternal blood glucose value documented during current pregnancy _____

Lowest maternal blood glucose value documented during current pregnancy units mmol/L mg/dL

Number of documented episodes of rhabdomyolysis during current pregnancy

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

Highest maternal CK value documented during current pregnancy _____

Highest maternal CK value documented during current pregnancy units U/L umol/L mg/dL

Longest permitted fasting interval (in hours) during the DAY recommended during current pregnancy

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

Longest permitted fasting interval (in hours) during the NIGHT recommended during current pregnancy

- Unknown 1 2 3 4 5 6
- 7 8 9 10 11 13

L-carnitine supplementation used on a regular basis during current pregnancy

- Unknown Yes No

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

L-carnitine not prescribed

Patient adhered to a fat-restricted diet during current pregnancy

Unknown Yes No

Fat-restriction not prescribed

Patient took a metabolic formula regularly during current pregnancy

Unknown Yes
 No Metabolic formula not prescribed

Evidence of cardiac hypertrophy during current pregnancy

Unknown Yes
 No Echocardiogram not performed during pregnancy

Evidence of any cardiac dysfunction during current pregnancy

Unknown Yes
 No Cardiac testing not performed during 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 **LCHAD - 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 allosioleucine Plasma isoleucine Plasma leucine Plasma valine
- Serum creatinine Other

Metabolite(s) of concern during second episode of dialysis- other, specify _____

Peak value of ammonia during second episode of dialysis _____

Peak value of ammonia during second episode of dialysis units

- umol/L ug/dL ug/L ug/mL g/dL

Peak value of blood urea nitrogen during second episode of dialysis _____

Peak value of blood urea nitrogen during second episode of dialysis units mg/dL mmol/L

Patient Name _____

Date

Peak value of plasma alloisoleucine during second episode of dialysis _____

Peak value of plasma alloisoleucine during second episode of dialysis units

 umol/dL umol/L mg/dL

Peak value of plasma isoleucine during second episode of dialysis _____

Peak value of plasma isoleucine during second episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during second episode of dialysis _____

Peak value of plasma leucine during second episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during second episode of dialysis _____

Peak value of plasma valine during second episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during second episode of dialysis _____

Peak value of serum creatinine during second episode of dialysis units umol/L mg/dL

Peak value of other metabolite during second episode of dialysis _____

Peak value of other metabolite during second episode of dialysis units _____

Reason second episode of dialysis treatment was stopped

 Unknown Acute episode resolved Treatment withdrawn Death Other

Reason second episode of dialysis treatment was stopped-other, specify _____

Third Dialysis Treatment

Type(s) of dialysis received during third episode

 Unknown APD/CCPD ECMO CAPD CVVH CVVD CVVHDF Hemodialysis Peritoneal dialysis

Reason for third episode of dialysis

 Unknown Hyperammonemia MSUD Organ failure Sepsis Other

Reason for third episode of dialysis-other, specify

Start date of third episode of dialysis treatment

Duration (in days) of third episode of dialysis treatment _____

Metabolite(s) of concern during third episode of dialysis

 Unknown None Ammonia Blood urea nitrogen Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine Serum creatinine Other

Patient Name _____

Date

Metabolite(s) of concern during third episode of dialysis-other, specify _____

Peak value of ammonia during third episode of dialysis _____

Peak value of ammonia during third episode of dialysis units

 umol/L ug/dL ug/L ug/mL g/dL

Peak value of blood urea nitrogen during third episode of dialysis _____

Peak value of blood urea nitrogen during third episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during third episode of dialysis _____

Peak value of plasma alloisoleucine during third episode of dialysis units

 umol/dL umol/L mg/dL

Peak value of plasma isoleucine during third episode of dialysis _____

Peak value of plasma isoleucine during third episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during third episode of dialysis _____

Peak value of plasma leucine during third episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during third episode of dialysis _____

Peak value of plasma valine during third episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during third episode of dialysis _____

Peak value of serum creatinine during third episode of dialysis units umol/L mg/dL

Peak value of other metabolite during third episode of dialysis _____

Peak value of other metabolite during third episode of dialysis units _____

Reason third episode of dialysis treatment was stopped

 Unknown Acute episode resolved Treatment withdrawn Death
 Other

Reason third episode of dialysis treatment was stopped-other, specify _____

Fourth Dialysis Treatment

Type(s) of dialysis received during fourth episode

 Unknown APD/CCPD ECMO CAPD
 CVVH CVVD CVVHDF Hemodialysis
 Peritoneal dialysis

Reason for fourth episode of dialysis

 Unknown Hyperammonemia MSUD Organ failure Sepsis
 Other

Reason for fourth episode of dialysis-other, specify

Patient Name _____

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

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

Duration (in days) of fourth episode of dialysis treatment _____

Metabolite(s) of concern during fourth episode of dialysis

- Unknown None Ammonia Blood urea nitrogen
 Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine
 Serum creatinine Other

Metabolite(s) of concern during fourth episode of dialysis-other, specify _____

Peak value of ammonia during fourth episode of dialysis _____

Peak value of ammonia during fourth episode of dialysis units

- umol/L ug/dL ug/L ug/mL g/dL

Peak value of blood urea nitrogen during fourth episode of dialysis _____

Peak value of blood urea nitrogen during fourth episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during fourth episode of dialysis _____

Peak value of plasma alloisoleucine during fourth episode of dialysis units

- umol/dL umol/L mg/dL

Peak value of plasma isoleucine during fourth episode of dialysis _____

Peak value of plasma isoleucine during fourth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during fourth episode of dialysis _____

Peak value of plasma leucine during fourth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during fourth episode of dialysis _____

Peak value of plasma valine during fourth episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during fourth episode of dialysis _____

Peak value of serum creatinine during fourth episode of dialysis units umol/L mg/dL

Peak value of other metabolite during fourth episode of dialysis _____

Peak value of other metabolite during fourth episode of dialysis units _____

Reason fourth episode of dialysis treatment was stopped

- Unknown Acute episode resolved Treatment withdrawn Death
 Other

Reason fourth episode of dialysis treatment was stopped-other, specify _____

Fifth Dialysis Treatment

Type(s) of dialysis received during fifth episode

- Unknown APD/CCPD ECMO CAPD
 CVVH CVVD CVVHDF Hemodialysis
 Peritoneal dialysis

Reason for fifth episode of dialysis

Patient Name _____

Date

- Unknown Hyperammonemia MSUD Organ failure Sepsis
- Other

Reason for fifth episode of dialysis-other, specify

Start date of fifth episode of dialysis treatment

Duration (in days) of fifth episode of dialysis treatment _____

Metabolite(s) of concern during fifth episode of dialysis

- Unknown None Ammonia Blood urea nitrogen
- Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine
- Serum creatinine Other

Metabolite(s) of concern during fifth episode of dialysis-other, specify _____

Peak value of ammonia during fifth episode of dialysis _____

Peak value of ammonia during fifth episode of dialysis units

- umol/L ug/dL ug/L ug/mL g/dL

Peak value of blood urea nitrogen during fifth episode of dialysis _____

Peak value of blood urea nitrogen during fifth episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during fifth episode of dialysis _____

Peak value of plasma alloisoleucine during fifth episode of dialysis units

- umol/dL umol/L mg/dL

Peak value of plasma isoleucine during fifth episode of dialysis _____

Peak value of plasma isoleucine during fifth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during fifth episode of dialysis _____

Peak value of plasma leucine during fifth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during fifth episode of dialysis _____

Peak value of plasma valine during fifth episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during fifth episode of dialysis _____

Peak value of serum creatinine during fifth episode of dialysis units umol/L mg/dL

Peak value of other metabolite during fifth episode of dialysis _____

Peak value of other metabolite during fifth episode of dialysis units _____

Reason fifth episode of dialysis treatment was stopped

- Unknown Acute episode resolved Treatment withdrawn Death
- Other

Reason fifth episode of dialysis treatment was stopped-other, specify _____

Patient Name _____

Date **Sixth Dialysis Treatment**

Type(s) of dialysis received during sixth episode

- Unknown APD/CCPD ECMO CAPD
 CVVH CVVD CVVHDF Hemodialysis
 Peritoneal dialysis

Reason for sixth episode of dialysis

- Unknown Hyperammonemia MSUD Organ failure Sepsis
 Other

Reason for sixth episode of dialysis-other, specify

Start date of sixth episode dialysis treatment

Duration (in days) of sixth episode of dialysis treatment _____

Metabolite(s) of concern during sixth episode of dialysis

- Unknown None Ammonia Blood urea nitrogen
 Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine
 Serum creatinine Other

Metabolite(s) of concern during sixth episode of dialysis-other, specify _____

Peak value of ammonia during sixth episode of dialysis _____

Peak value of ammonia during sixth episode of dialysis units

- umol/L ug/dL ug/L ug/mL g/dL

Peak value of blood urea nitrogen during sixth episode of dialysis _____

Peak value of blood urea nitrogen during sixth episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during sixth episode of dialysis _____

Peak value of plasma alloisoleucine during sixth episode of dialysis units

- umol/dL umol/L mg/dL

Peak value of plasma isoleucine during sixth episode of dialysis _____

Peak value of plasma isoleucine during sixth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during sixth episode of dialysis _____

Peak value of plasma leucine during sixth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during sixth episode of dialysis _____

Peak value of plasma valine during sixth episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during sixth episode of dialysis _____

Peak value of serum creatinine during sixth episode of dialysis units umol/L mg/dL

Patient Name _____

Date

Peak value of other metabolite during sixth episode of dialysis _____

Peak value of other metabolite during sixth episode of dialysis units _____

Reason sixth episode of dialysis treatment was stopped

- Unknown
- Acute episode resolved
- Treatment withdrawn
- Death
- Other

Reason sixth episode of dialysis treatment was stopped-other, specify _____

Seventh Dialysis Treatment

Type(s) of dialysis received during seventh episode

- Unknown
- APD/CCPD
- ECMO
- CAPD
- CVVH
- CVVD
- CVVHDF
- Hemodialysis
- Peritoneal dialysis

Reason for seventh episode of dialysis

- Unknown
- Hyperammonemia
- MSUD
- Organ failure
- Sepsis
- Other

Reason for seventh episode of dialysis -other, specify

Start date of seventh episode of dialysis treatment

Duration (in days) of seventh episode of dialysis treatment _____

Metabolite(s) of concern during seventh episode of dialysis

- Unknown
- None
- Ammonia
- Blood urea nitrogen
- Plasma alloisoleucine
- Plasma isoleucine
- Plasma leucine
- Plasma valine
- Serum creatinine
- Other

Metabolite(s) of concern during seventh episode of dialysis-other, specify _____

Peak value of ammonia during seventh episode of dialysis _____

Peak value of ammonia during seventh episode of dialysis units

- umol/L
- ug/dL
- ug/L
- ug/mL
- g/dL

Peak value of blood urea nitrogen during seventh episode of dialysis _____

Peak value of blood urea nitrogen during seventh episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during seventh episode of dialysis _____

Peak value of plasma alloisoleucine during seventh episode of dialysis units

- umol/dL
- umol/L
- mg/dL

Peak value of plasma isoleucine during seventh episode of dialysis _____

Peak value of plasma isoleucine during seventh episode of dialysis units

Patient Name _____

Date

umol/dL umol/L mg/dL

Peak value of plasma leucine during seventh episode of dialysis _____

Peak value of plasma leucine during seventh episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during seventh episode of dialysis _____

Peak value of plasma valine during seventh episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during seventh episode of dialysis _____

Peak value of serum creatinine during seventh episode of dialysis units umol/L mg/dL

Peak value of other metabolite during seventh episode of dialysis _____

Peak value of other metabolite during seventh episode of dialysis units _____

Reason seventh episode of dialysis treatment was stopped

- Unknown Acute episode resolved Treatment withdrawn Death
- Other

Reason seventh episode of dialysis treatment was stopped-other, specify _____

Eighth Dialysis Treatment

Type(s) of dialysis received during eighth episode

- Unknown APD/CCPD ECMO CAPD
- CVVH CVVD CVVHDF Hemodialysis
- Peritoneal dialysis

Reason for eighth episode of dialysis

- Unknown Hyperammonemia MSUD Organ failure Sepsis
- Other

Reason for eighth episode of dialysis-other, specify

Start date of eighth episode of dialysis treatment

Duration (in days) of eighth episode of dialysis treatment _____

Metabolite(s) of concern during eighth episode of dialysis

- Unknown None Ammonia Blood urea nitrogen
- Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine
- Serum creatinine Other

Metabolite(s) of concern during eighth episode of dialysis-other, specify _____

Peak value of ammonia during eighth episode of dialysis _____

Peak value of ammonia during eighth episode of dialysis units

umol/L ug/dL ug/L ug/mL g/dL

Patient Name _____

Date

Peak value of blood urea nitrogen during eighth episode of dialysis _____

Peak value of blood urea nitrogen during eighth episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during eighth episode of dialysis _____

Peak value of plasma alloisoleucine during eighth episode of dialysis units

umol/dL

umol/L

mg/dL

Peak value of plasma isoleucine during eighth episode of dialysis _____

Peak value of plasma isoleucine during eighth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during eighth episode of dialysis _____

Peak value of plasma leucine during eighth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during eighth episode of dialysis _____

Peak value of plasma valine during eighth episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during eighth episode of dialysis _____

Peak value of serum creatinine during eighth episode of dialysis units umol/L mg/dL

Peak value of other metabolite during eighth episode of dialysis _____

Peak value of other metabolite during eighth episode of dialysis units _____

Reason eighth episode of dialysis treatment was stopped

Unknown

Acute episode resolved Treatment withdrawn Death

Other

Reason eighth episode of dialysis treatment was stopped-other, specify _____

Ninth Dialysis Treatment

Type(s) of dialysis received during ninth episode

Unknown

APD/CCPD

ECMO

CAPD

CVVH

CVVD

CVVHDF

Hemodialysis

Peritoneal dialysis

Reason for ninth episode of dialysis

Unknown

Hyperammonemia MSUD

Organ failure

Sepsis

Other

Reason for ninth episode of dialysis-other, specify

Start date of ninth episode of dialysis treatment

Duration (in days) of ninth episode of dialysis treatment _____

Metabolite(s) of concern during ninth episode of dialysis

Unknown

None

Ammonia

Blood urea nitrogen

Patient Name _____

Date Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine Serum creatinine Other

Metabolite(s) of concern during ninth episode of dialysis-other, specify _____

Peak value of ammonia during ninth episode of dialysis _____

Peak value of ammonia during ninth episode of dialysis units

 umol/L ug/dL ug/L ug/mL g/dL

Peak value of blood urea nitrogen during ninth episode of dialysis _____

Peak value of blood urea nitrogen during ninth episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during ninth episode of dialysis _____

Peak value of plasma alloisoleucine during ninth episode of dialysis units

 umol/dL umol/L mg/dL

Peak value of plasma isoleucine during ninth episode of dialysis _____

Peak value of plasma isoleucine during ninth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during ninth episode of dialysis _____

Peak value of plasma leucine during ninth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during ninth episode of dialysis _____

Peak value of plasma valine during ninth episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during ninth episode of dialysis _____

Peak value of serum creatinine during ninth episode of dialysis units umol/L mg/dL

Peak value of other metabolite during ninth episode of dialysis _____

Peak value of other metabolite during ninth episode of dialysis units _____

Reason ninth episode of dialysis treatment was stopped

 Unknown Acute episode resolved Treatment withdrawn Death Other

Reason ninth episode of dialysis treatment was stopped-other, specify _____

Tenth Dialysis Treatment

Type(s) of dialysis received during tenth episode

 Unknown APD/CCPD ECMO CAPD CVVH CVVD CVVHDF Hemodialysis Peritoneal dialysis

Reason for tenth episode of dialysis

 Unknown Hyperammonemia MSUD Organ failure Sepsis Other

Patient Name _____

Date

Reason for tenth episode of dialysis-other, specify

Start date of tenth episode of dialysis treatment

Duration (in days) of tenth episode of dialysis treatment _____

Metabolite(s) of concern during tenth episode of dialysis

- Unknown None Ammonia Blood urea nitrogen
- Plasma alloisoleucine Plasma isoleucine Plasma leucine Plasma valine
- Serum creatinine Other

Metabolite(s) of concern during tenth episode of dialysis-other, specify _____

Peak value of ammonia during tenth episode of dialysis _____

Peak value of ammonia during tenth episode of dialysis units

- umol/L ug/dL ug/L ug/mL g/dL

Peak value of blood urea nitrogen during tenth episode of dialysis _____

Peak value of blood urea nitrogen during tenth episode of dialysis units mg/dL mmol/L

Peak value of plasma alloisoleucine during tenth episode of dialysis _____

Peak value of plasma alloisoleucine during tenth episode of dialysis units

- umol/dL umol/L mg/dL

Peak value of plasma isoleucine during tenth episode of dialysis _____

Peak value of plasma isoleucine during tenth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma leucine during tenth episode of dialysis _____

Peak value of plasma leucine during tenth episode of dialysis units umol/dL umol/L mg/dL

Peak value of plasma valine during tenth episode of dialysis _____

Peak value of plasma valine during tenth episode of dialysis units umol/dL umol/L mg/dL

Peak value of serum creatinine during tenth episode of dialysis _____

Peak value of serum creatinine during tenth episode of dialysis units umol/L mg/dL

Peak value of other metabolite during tenth episode of dialysis _____

Peak value of other metabolite during tenth episode of dialysis units _____

Reason tenth episode of dialysis treatment was stopped

- Unknown Acute episode resolved Treatment withdrawn Death
- Other

Reason tenth episode of dialysis treatment was stopped-other, specify _____

Patient Name _____

Date

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

Comments

Dialysis comments

Patient Name _____

Date **LCHAD - 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)

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Complications during the second transplant procedure Unknown Yes No

Complications during the second transplant procedure- specify

 Clotting Death Major bleeding Other

Complications during the second transplant procedure, other- specify _____

Complications post second transplant Unknown Yes No

Complications post second transplant- specify

<input type="checkbox"/> Unknown	<input type="checkbox"/> Cancer
<input type="checkbox"/> Cataract	<input type="checkbox"/> Clotting
<input type="checkbox"/> Death	<input type="checkbox"/> Delayed graft function
<input type="checkbox"/> Diabetes mellitus	<input type="checkbox"/> High cholesterol
<input type="checkbox"/> Hypertension	<input type="checkbox"/> Infection
<input type="checkbox"/> Major bleeding	<input type="checkbox"/> Osteoporosis
<input type="checkbox"/> Reappearance of heart disease	<input type="checkbox"/> Reappearance of kidney disease
<input type="checkbox"/> Reappearance of liver disease	<input type="checkbox"/> Rejection

Patient Name _____

Date Transplant related renal impairment Other

Complications post second transplant, other- specify _____

Third Transplant

Third organ received by transplant

Unknown Bone marrow Heart Kidney Liver Lung
 Pancreas Stem Cell Other

Third organ received by transplant, other- specify _____

Age (in days) at third transplant _____

Reason for third transplant

Unknown Treatment of disorder Renal failure Liver failure
 Heart failure Other

Reason for third transplant, other- specify _____

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

Number of outpatient metabolic visits in the last year post transplant

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

Metabolic labs monitored post third transplant Unknown Yes No

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

Complications during the third transplant procedure Unknown Yes No

Complications during the third transplant procedure- specify

Clotting Death Major bleeding Other

Complications during the third transplant procedure, other- specify _____

Complications post third transplant Unknown Yes No

Complications post third transplant- specify

Unknown Cancer
 Cataract Clotting
 Death Delayed graft function
 Diabetes mellitus High cholesterol
 Hypertension Infection
 Major bleeding Osteoporosis
 Reappearance of heart disease Reappearance of kidney disease
 Reappearance of liver disease Rejection
 Transplant related renal impairment Other

Patient Name _____

Date

Complications post third transplant, other- specify _____

Fourth Transplant

Fourth organ received by transplant

- Unknown Bone marrow Heart Kidney Liver Lung
 Pancreas Stem Cell Other

Fourth organ received by transplant, other- specify _____

Age (in days) at fourth transplant _____

Reason for fourth transplant

- Unknown Treatment of disorder Renal failure Liver failure
 Heart failure Other

Reason for fourth transplant, other- specify _____

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

Number of outpatient metabolic visits in the last year post transplant

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

Metabolic labs monitored post fourth transplant Unknown Yes No

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

Complications during the fourth transplant procedure Unknown Yes No

Complications during the fourth transplant procedure- specify

- Clotting Death Major bleeding Other

Complications during the fourth transplant procedure, other- specify _____

Complications post fourth transplant Unknown Yes No

Complications post fourth transplant- specify

- Unknown Cancer
 Cataract Clotting
 Death Delayed graft function
 Diabetes mellitus High cholesterol
 Hypertension Infection
 Major bleeding Osteoporosis
 Reappearance of heart disease Reappearance of kidney disease
 Reappearance of liver disease Rejection
 Transplant related renal impairment Other

Complications post fourth transplant, other- specify _____

Patient Name _____

Date **Fifth Transplant**

Fifth organ received by transplant

- Unknown Bone marrow Heart Kidney Liver Lung
 Pancreas Stem Cell Other

Fifth organ received by transplant, other- specify _____

Age (in days) at fifth transplant _____

Reason for fifth transplant

- Unknown Treatment of disorder Renal failure Liver failure
 Heart failure Other

Reason for fifth transplant, other- specify _____

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

Number of outpatient metabolic visits in the last year post transplant

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

Metabolic labs monitored post fifth transplant Unknown Yes No

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

Complications during the fifth transplant procedure Unknown Yes No

Complications during the fifth transplant procedure- specify

- Clotting Death Major bleeding Other

Complications during the fifth transplant procedure, other- specify _____

Complications post fifth transplant Unknown Yes No

Complications post fifth transplant- specify

- Unknown Cancer
 Cataract Clotting
 Death Delayed graft function
 Diabetes mellitus High cholesterol
 Hypertension Infection
 Major bleeding Osteoporosis
 Reappearance of heart disease Reappearance of kidney disease
 Reappearance of liver disease Rejection
 Transplant related renal impairment Other

Complications post fifth transplant, other- specify _____

Patient Name _____

Date

Provide details for any other transplants not listed above

Comments

Transplant comments