GALC - Demographics

NBSTRN ID_____

Intake date |M|M| |D|D| |Y|Y|

Consent

Consent obtained ○Yes ○No ○IRB Exempt

Assent obtained ○Yes ○No ○Not applicable

Type of assent ○Written ○Verbal

Permission to recontact ○Unknown ○Yes ○No

Demographics Information

Patient last name_____

Patient first name_____

Date of birth |M|M| |D|D| |Y|Y|

Gestational Age (weeks)_____

Gestational Age (days)_____

Societal sex ○Unknown ○Male ○Female ○Ambiguous

Biological mother's maiden name_____

Condition

Patient condition category □SACHDNC candidate disorders

Specify SACHDNC candidate disorder diagnosis for the patient □Krabbe disease

Patient disorder identification method

□Unknown □Abnormal newborn screen □Abnormal labs

□Clinical presentation □Family member with this condition □Missed by newborn sceening

Miles from home to specialty care_____

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

Specify medical home-other, specify_____

Patient is in other research studies ○Unknown ○Yes ○No

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

Research study-other, specify_____

Clinicaltrials.gov identifier_____

Education

Maternal education

○Unknown

(Continued on next page.)

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

Paternal education

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

Ancestral Origin, Race and Ethnicity

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

Ancestral Origin-Africa

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

Ancestral Origin-Africa-Other, specify_____

Ancestral Origin-Asia

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

Ancestral Origin-Asia-Other, specify_____

Ancestral Origin-Europe

☐Unknown ☐Austria ☐Belgium ☐Bulgaria ☐Croatia
☐Czech Republic ☐Denmark ☐Finland ☐France ☐Germany
☐Greece ☐Hungary ☐Iceland ☐Ireland ☐Italy

(Continued on next page.)

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

Ancestral Origin-Europe-Italy ☐Unknown ☐Sicily

Ancestral Origin-Europe-Romania ☐Unknown ☐Transylvania

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

Ancestral Origin-Europe-Other, specify_____

Ancestral Origin-North America

☐Unknown ☐Aleutian Islands ☐Canada ☐Dominican Republic ☐Honduas
☐Mexico ☐Puerto Rico ☐United States ☐Other

Ancestral Origin-North America-Canada ☐Unknown ☐French Canadian

Ancestral Origin-North America-Other, specify_____

Ancestral Origin-South America ☐Unknown ☐Colombia ☐Venezuela ☐Other

Ancestral Origin-South America-Other, specify_____

Ancestral Origin-Oceania ☐Unknown ☐Australia ☐Other

Ancestral Origin-Oceania-Other, specify_____

Ancestral Origin-Other ☐Unknown ☐Amish ☐Arabic ☐Hutterite ☐Mennonite ☐Jewish ☐Other

Ancestral Origin-Other-Jewish ☐Unknown ☐Ashkenazic ☐Sephardic

Ancestral Origin-Other, specify_____

Race

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

Race-White, specify ○Ashkenazi Jewish ○Non-Ashkenazi Jewish

Patient is Hispanic or Latino ○Not reported ○Yes ○No

Socioeconomics

Maternal 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

Medical Coverage

Maternal medical coverage at time of delivery

☐Unknown
☐None
☐Commercial/private

(Continued on next page.)

- ☐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

Comments

Intake demographics comments

Patient Name_____

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

GALC - Enzymemutation Analysis

Newborn Screening

Newborn screening performed ○Unknown ○Yes ○No

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

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

First newborn screen taken 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 ○Pilot study ○Quality improvement

GALC enzyme activity on first newborn screen (umol/L/hr)_____

GALC (% daily mean) on first newborn screen_____

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

Reason for second newborn screen

☐Unsatisfactory ☐Borderline Result ☐NICU Protocol

☐TPN ☐Transfused ☐State Mandate

☐NBS collected < 24 hours of age ☐Other

Reason for second newborn screen-other, specify_____

GALC enzyme activity on second newborn screen (umol/L/hr)_____

GALC (% daily mean) on second newborn screen_____

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

Reason for third newborn screen

☐Unsatisfactory ☐Borderline Result ☐NICU Protocol ☐TPN ☐Transfused

☐State Mandate ☐Other

Reason for third newborn screen-other, specify_____

GALC enzyme activity on third newborn screen (umol/L/hr)_____

GALC (% daily mean) on third newborn screen_____

Enzymatic Confirmatory Testing

Date of enzymatic confirmatory testing |M|M| |D|D| |Y|Y|

Lab where enzymatic confirmation was done

○Icahn School of Medicine ○Thomas Jefferson University ○Mayo Clinic

○Greenwood ○Emory ○Duke

○Other

Lab where enzymatic confirmation was done-other, specify_____

Confirmatory testing GALC result_____

Confirmatory testing GALC normal reference range_____

Newborn Hearing Screen

Newborn hearing screen performed ○Unknown ○Yes ○No

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

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

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

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

Recommendation

○Repeat hearing screen ○Referral for diagnostic testing ○Risk factor monitoring only

○Refused further action ○No further action required

Date of Audiological Diagnostic Evaluation |M|M| |D|D| |Y|Y|

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

Right ear: Diagnosis: Degree of hearing loss

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

Right ear: Diagnosis: Type of hearing loss

☐Sensorineural ☐Conductive

☐Mixed ☐Auditory neuropathy/Auditory dys-synchrony

☐Other

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

Left ear: Diagnosis: Degree of hearing loss

○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

Patient status at time of NBS reporting to specialty center ○Unknown ○Well ○Symptomatic ○Deceased

Patient symptoms at time of initial contact

☐Unknown	☐None	☐Alopecia
☐Apnea	☐Arrhythmia	☐Ataxia
☐Athetosis	☐Autistic-like features	☐Body odor
☐Brain abnormalities	☐Brain malformations	☐Candidiasis
☐Cardiomyopathy	☐Cataract(s)	☐Cerebral edema
☐Chorea	☐Cirrhosis	☐Clonus
☐Cognitive impairment	☐Coma	☐Confusion
☐Conjunctivitis	☐Contracture(s)-musculoskeletal	☐Corneal erosion

(Continued on next page.)

☐Dehydration	☐Dermatitis	☐Developmental delay(s)
☐Disorientation	☐Drooling/hypersalivation	☐Dysarthria
☐Dysmetria	☐Dysmorphism	☐Dysphagia
☐Dystonia	☐Eczema	☐Edema
☐Failure to thrive	☐Fatigue	☐Flapping tremor
☐Fluctuating level of alertness	☐Gait abnormality (other than ataxia)	☐Genital abnormalities
☐Headache	☐Hearing loss	☐Hepatic encephalopathy
☐Hepatomegaly	☐Hyperreflexia	☐Hypertension
☐Hypertonia	☐Hyporeflexia	☐Hypothermia
☐Hypotonia	☐Increased intracranial pressure	☐Infection/sepsis
☐Irritability	☐Jaundice	☐Keratosis
☐Learning disability	☐Lethargy	☐Liver failure-acute
☐Loss of consciousness	☐Loss of developmental milestone(s)	☐Macrocephaly
☐Malignant hyperthermia	☐Microcephaly	☐Multiorgan failure
☐Myopathy	☐Nystagmus	☐Opisthotonos
☐Optic nerve atrophy	☐Pancreatitis	☐Peripheral neuropathy
☐Photophobia	☐Polycystic kidney(s)	☐Poor feeding
☐Poor growth	☐Profuse sweating	☐Renal dysplasia
☐Renal failure-acute	☐Retinal hemorrhage	☐Rickets
☐Rigidity	☐Scotomas	☐Seizure
☐Slurred speech	☐Spasticity	☐Splenomegaly
☐Stereotyped movements	☐Stomatitis	☐Stridor
☐Stroke	☐Subdural hemorrhage	☐Sudden death
☐Syncope	☐Tachycardia	☐Tachypnea
☐Tremors	☐Trichorrhexis nodosa	☐Vision loss
☐Vomiting	☐Other	

Patient symptoms at time of initial contact-other, specify_____

Mutation 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		

Mutation 1_____

Mutation 2_____

Polymorphisms_____

Most appropriate Krabbe diagnosis

○ Early Infantile ○ Late Infantile/Early Childhood ○ Late Childhood/Juvenile

○ Unknown Significance

Mutation is de novo ○ Unknown ○ Yes ○ No

Pseudodeficiency alleles_____

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

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

Phase of mutation(s) ○ Unknown ○ Maternal ○ Paternal

Comments

Enzyme mutation analysis comments

Patient Name_____

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

GALC - Initial Medical History

Date medical history performed |M|M| |D|D| |Y|Y|

Prenatal History

Prenatal diagnosis done for this condition ○Unknown ○Yes ○No

Issues concerning mother's pregnancy with this patient

☐Unknown ☐None ☐Pregnancy complications ☐Assisted reproduction

Pregnancy complications/risk factors

☐Unknown

☐Acute fatty liver of pregnancy (AFLP)

☐Advanced maternal age (35+ years of age)

☐Ectopic pregnancy

☐Gestational diabetes

☐Group B strep

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

☐Intrauterine growth restriction (AUGR)

☐Inadequate prenatal care

☐Maternal prenatal substance exposure

☐Mother affected with this condition

☐Preeclampsia

☐Rh isoimmunization

☐Toxemia

☐Young maternal age (15 years of age + under)

☐Preterm labor

☐Other

Type of prenatal substance exposure

☐Unknown ☐Alcohol ☐Tobacco ☐Illicit drugs ☐Harmful chemicals

☐Known teratogens

Maternal treatment for affected fetus ○Unknown ○None ○Yes ○No

Type of maternal treatment for affected fetus

Pregnancy complication/risk factor-other, specify_____

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

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

Type(s) of assisted reproductive technology-other, specify_____

Medical issues related to prematurity

☐None ☐Intracranial hemorrhage ☐Necrotizing enterocolitis
☐Respiratory distress ☐Retinopathy of prematurity ☐Total parenteral nutrition (TPN)
☐Sepsis ☐Blood transfusions ☐Other

Intracranial hemorrhage grade ○Unknown ○1 ○2 ○3

Necrotizing enterocolitis comments

Type of respiratory support required for respiratory distress

☐Unknown ☐None ☐Oxygen ☐CPAP ☐Ventilator ☐Surfactant ☐Other

Medical issues related to prematurity-other, specify

Neonatal History

Patient's birth was a result of multiple gestation pregnancy

○Unknown ○No-single birth ○Yes-twins (identical)
○Yes-twins (fraternal) ○Yes-other higher order multiple ○Yes-Other, specify

Specify other number of multiples_____

Birth Measurements

Birth measurements ☐Unknown ☐Head circumference ☐Length ☐Weight

Birth head circumference_____

Birth head circumference units ○cm ○in

Birth length_____

Birth length units ○cm ○in

Birth weight_____

Birth weight units ○lbs ○kg ○gm ○oz

1 minute APGAR score ○Unknown ○Not done ○0 ○1 ○2 ○3 ○4 ○5 ○6 ○7 ○8 ○9 ○10

5 minute APGAR score ○Unknown ○Not done ○0 ○1 ○2 ○3 ○4 ○5 ○6 ○7 ○8 ○9 ○10

10 minute APGAR score ○Unknown ○Not done ○0 ○1 ○2 ○3 ○4 ○5 ○6 ○7 ○8 ○9 ○10

Nutrition

Type of neonatal nutrition

☐Unknown ☐TPN ☐Breast milk ☐Elemental formula

☐Human milk fortifier ☐Intralipid ☐Regular formula ☐Non-Lactose formula

☐Metabolic formula ☐Other

Type of neonatal nutrition-other, specify_____

Family History

Consanguinity ○Unknown ○Yes ○No

Type of consanguinity ○First cousins or closer ○Other

Pedigree obtained ○Yes ○No

Family members with confirmed Krabbe ○Unknown ○Yes ○No

Relationship of family members with confirmed Krabbe to proband

☐Mother ☐Father ☐Sister ☐Brother ☐Grandmother ☐Grandfather ☐Aunt

☐Uncle ☐First cousin ☐Half-sister ☐Half-brother ☐Other

Relationship of family members with confirmed Krabbe to proband-Other, specify_____

Family member(s) with symptoms suggestive of Krabbe ○Unknown ○Yes ○No

Relationship of family members with suggested Krabbe to proband

☐Mother ☐Father ☐Sister ☐Brother ☐Grandmother ☐Grandfather ☐Aunt

☐Uncle ☐First cousin ☐Half-sister ☐Half-brother ☐Other

Relationship of family members with suggested Krabbe to proband-Other, specify_____

Family history/ancestry comments

Family Demographics

Maternal race

☐Not reported ☐American Indian/Alaskan Native

☐Asian ☐Black or African American

☐Native Hawaiian or Other Pacific Islander ☐White

Maternal race-White ○Ashkenazi Jewish ○Non-Ashkenazi Jewish

Mother is Hispanic or Latino ○Not reported ○Yes ○No

Paternal race

(Continued on next page.)

☐Not reported ☐American Indian/Alaskan Native

☐Asian ☐Black or African American

☐Native Hawaiian or Other Pacific Islander ☐White

Paternal race-White ○Ashkenazi Jewish ○Non-Ashkenazi Jewish

Father is Hispanic or Latino ○Not reported ○Yes ○No

Hospitalizations prior to intake ○Unknown ○Yes ○No

Number of hospitalizations prior to intake related to this condition_____

Number of hospitalizations prior to intake not related to this condition_____

Genetic counseling provided ○Unknown ○Yes ○No

Provider of genetic counseling

☐Unknown ☐Dietitian ☐Genetic counselor ☐Neuropsychologist ☐Nurse

☐Nurse practitioner ☐Physician ☐Physician assistant ☐Other

Provider of genentic counseling, other- specify_____

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

Initial medical history comments

GALC - Followup Medical History

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

Age at visit CALCULATED Condition follow-up status ○Active ○Inactive

Reason for inactive status

○Unknown ○Deceased ○Lost to follow-up

○Moved ○Refused follow-up ○Follow-up deemed unnecessary

○Subject withdrawal from study

Date of death |M|M| |D|D| |Y|Y|

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

Miles from home to specialty care_____

Patient has enrolled in a research study since the last visit ○Unknown ○Yes ○No

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

Identify the research study_____

Clinicaltrials.gov identifier_____

Care Coordination

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

Number of missed subspecialty visits_____

Missed phone calls since last visit ○Unknown ○Yes ○No

Number of missed phone calls_____

Missed school or work since last visit ○Unknown ○Yes ○No

Number of missed school or work days_____

Other health services currently received ○Unknown ○Yes ○No

Specify other current health services

☐Unknown ☐Allergy

☐Audiology ☐Behavioral/Developmental

☐Cardiology ☐Dentistry

☐Dermatology ☐Dietitian

☐Endocrinology ☐Gastroenterology

☐Genetic Counseling ☐Hematology

☐Home health care ☐Nephrology

☐Neurology ☐Neuropsychology

☐Neurosurgery ☐Occupational therapy

☐Oncology ☐Ophthalmology

☐Orthopedics ☐Otolaryngology

(Continued on next page.)

☐Physical medicine and rehabilitation (PM&R) ☐Physical therapy
☐Primary care provider ☐Psychiatry
☐Psychology ☐Public health nursing
☐Pulmonology ☐Respiratory therapy
☐Speech-Language therapy ☐Surgery
☐Transplant ☐Other

Specify other current health services-other, specify_____

Preventive care status

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

Medical coverage at visit

☐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

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_____

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

(Continued on next page.)

☐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

☐Speech or language impairments ☐Traumatic brain injury

☐Other health disabilities

Special education, other- specify_____

Procedures

Anesthesia since last visit ○Unknown ○Yes ○No

Type of anesthesia since last visit ○General ○Regional ○Local

Constitutional Changes

Constitutional changes since last visit

☐Unknown ☐None ☐Patient fatigues easily ☐Patient requires daily naps

☐Patient has early satiety ☐Patient is small for age

Approximate age of onset for fatigue

○Unknown ○0 ○1 ○2 ○3 ○4 ○5 ○6

○7 ○8 ○9 ○10 ○11 ○12 ○13 ○14

○15 ○16 ○17 ○18

Approximate age of onset for early satiety

○Unknown ○0 ○1 ○2 ○3 ○4 ○5 ○6

○7 ○8 ○9 ○10 ○11 ○12 ○13 ○14

○15 ○16 ○17 ○18

Hematologic Changes

Hematologic changes since last visit

☐Unknown ☐None ☐Patient bruises easily ☐Patient bleeds easily

☐Nosebleeds ☐Bleeding from gums ☐Heavy menstrual bleeding ☐Transfusions required

Approximate age of onset for bruising easily

○Unknown ○0 ○1 ○2 ○3 ○4 ○5 ○6

○7 ○8 ○9 ○10 ○11 ○12 ○13 ○14

(Continued on next page.)

○15 ○16 ○17 ○18

Approximate age of onset for easy bleeding

○Unknown ○0 ○1 ○2 ○3 ○4 ○5 ○6
○7 ○8 ○9 ○10 ○11 ○12 ○13 ○14
○15 ○16 ○17 ○18

Approximate age of onset for nosebleed

○Unknown ○0 ○1 ○2 ○3 ○4 ○5 ○6
○7 ○8 ○9 ○10 ○11 ○12 ○13 ○14
○15 ○16 ○17 ○18

Approximate age of onset for bleeding from gums

○Unknown ○0 ○1 ○2 ○3 ○4 ○5 ○6
○7 ○8 ○9 ○10 ○11 ○12 ○13 ○14
○15 ○16 ○17 ○18

Approximate age of onset for heavy menstrual bleeding

○Unknown ○0 ○1 ○2 ○3 ○4 ○5 ○6
○7 ○8 ○9 ○10 ○11 ○12 ○13 ○14
○15 ○16 ○17 ○18

Number of transfusions ○1 ○2 ○3 or more

Date of transfusion 1 |M|M| |D|D| |Y|Y|
Date of transfusion 2 |M|M| |D|D| |Y|Y|
Date of transfusion 3 |M|M| |D|D| |Y|Y|

Circumstances under which the patient has had blood transfusions

GI Changes

GI changes since last visit

☐Unknown ☐None ☐Frequent abdominal pain
☐Patient uses gastrostomy tube ☐GERD ☐Liver disease
☐Frequent diarrhea

Average number of daily bowel movements ○0 ○1 ○2 ○3 ○4 ○5 or more

Approximate age of onset for frequent abdominal pain

○Unknown ○0 ○1 ○2 ○3 ○4 ○5 ○6
○7 ○8 ○9 ○10 ○11 ○12 ○13 ○14
○15 ○16 ○17 ○18

Approximate age at which gastrostomy tube was inserted

○Unknown ○0 ○1 ○2 ○3 ○4 ○5 ○6
○7 ○8 ○9 ○10 ○11 ○12 ○13 ○14
○15 ○16 ○17 ○18

Approximate age of onset for GERD

○Unknown ○0 ○1 ○2 ○3 ○4 ○5 ○6
○7 ○8 ○9 ○10 ○11 ○12 ○13 ○14
○15 ○16 ○17 ○18

Type of liver disease ○Unknown ○Jaundice ○Cirrhosis ○Infectious liver disease ○Liver failure

Approximate age of onset for liver disease

○Unknown ○0 ○1 ○2 ○3 ○4 ○5 ○6
○7 ○8 ○9 ○10 ○11 ○12 ○13 ○14
○15 ○16 ○17 ○18

Neurological changes since last visit

☐Unknown ☐None ☐Seizures ☐Peripheral neuropathy
☐Vision loss ☐Other

Neurological changes since last visit-Other, specify_____

Psychiatric Changes

Psychiatric changes since last visit ☐Unknown ☐None ☐Depression ☐Anxiety ☐Other

Describe the patient's depression

Patient is on medication for depression ○Unknown ○Yes ○No

Patient required hospitalization for depression ○Unknown ○Yes ○No

Patient is suicidal ○Unknown ○Yes ○No

Approximate age of onset for depression

○Unknown ○0 ○1 ○2 ○3 ○4 ○5 ○6
○7 ○8 ○9 ○10 ○11 ○12 ○13 ○14
○15 ○16 ○17 ○18

Describe the impact of the patient's depression on the family

Approximate age of onset for anxiety

(Continued on next page.)

○Unknown ○0 ○1 ○2 ○3 ○4 ○5 ○6
○7 ○8 ○9 ○10 ○11 ○12 ○13 ○14
○15 ○16 ○17 ○18

Describe any other psychiatric problems

Developmental Assessment

Developmental assessment done at this visit ○Unknown ○Yes ○No

Standardized developmental screening tool(s) used ○Unknown ○Yes ○No

Developmental status ○Typical ○Atypical

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

Developmental milestones that were achieved in a typical order and timeframe

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

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

_____

Communication ○Normal ○Delayed

Delayed communication ☐No speech ☐Articulation difficulties ☐Uses assistive devices

Gait abnormalities

☐Walks independently ☐Walks with assistive ambulation device
☐Needs orthotics ☐Wheelchair bound

Referred for further developmental assessment

○Unknown ○Yes
○No ○Previously referred
○Family declined further assessments

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

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

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

_____

Progressive loss of developmental milestones since last visit ○Unknown ○Yes ○No

Transplants Since Last Visit

Organ transplant since last visit ○Unknown ○Yes ○No

Transplant organ ○Unknown ○Bone marrow ○Stem Cell ○Liver ○Kidney ○Heart ○Other

Transplant organ-other, specify_____

Date of organ transplant M M D D Y Y

Reason for transplant

○Unknown ○Liver failure ○Condition-related treatment

○Condition-related organ failure ○Other

Reason for transplant-other, specify_____

Other

Immunization status

○Unknown ○Not up to date ○Up to date via report

○Up to date via clinical confirmation ○Immunizations declined

Comments

Followup medical history comments

GALC - Physical Exam

Date of Physical Exam M M D D Y Y

Vital Signs

Vital signs taken at visit

☐Unknown ☐None ☐Blood pressure ☐Head circumference ☐Height/length
☐Pulse ☐Respiratory rate ☐Weight

Blood pressure-systolic_____

Blood pressure-diastolic_____

Head circumference_____

Head circumference units ○cm ○in

Head circumference percentile (GA appropriate)_____

Height/length_____

Height/length units ○cm ○in

How height/length measured ○Unknown ○Supine ○Standing

Height/length percentile (GA appropriate)_____

Pulse_____

Respiratory Rate_____

Weight_____

Weight units ○lbs ○kg ○gm ○oz

Weight percentile (GA appropriate)_____

Abnormal vital sign findings ○Unknown ○Yes ○No

Describe any abnormal vital sign findings

Constitutional

Constitutional exam findings ○Unknown ○Not done ○Normal ○Abnormal

Constitutional abnormality ☐Unknown ☐Small for age ☐Sweating ☐Other

Constitutional abnormality-other, specify

Physical Exam

Exam findings ○Unknown ○Not done ○Normal ○Abnormal

Abnormal exam findings ☐Irritability ☐Temperature instability ☐Increased tone/arching ☐Other

Abnormal exam findings-Other, specify_____

Describe all abnormal exam findings

Presence of hernias ☐None ☐Inguinal ☐Umbilical ☐Epigastric ☐Incisional

HEENT Exam

HEENT Exam Findings ○Unknown ○Not done ○Normal ○Abnormal

Describe all abnormal HEENT exam findings

Neck Exam

Neck exam findings ○Unknown ○Not done ○Normal ○Abnormal

Describe all abnormal neck findings ☐Back extremeties ☐Drooling ☐Suck ☐Microglossia ☐Other

Describe all abnormal neck findings-Other, specify

Lung Exam

Lung exam findings ○Unknown ○Not done ○Normal ○Abnormal

Describe all lung abnormalities

Chest Exam

Chest exam findings ○Unknown ○Not done ○Normal ○Abnormal

Abnormal chest exam findings ☐Chest wall ☐Other

Heart exam findings ○Unknown ○Not done ○Normal ○Abnormal

Describe all abnormal heart exam findings

Abdomen Exam

Abdomen exam findings ○Unknown ○Not done ○Normal ○Abnormal

Method of assessment for abnormal abdomen exam findings ☐Clinical exam ☐MRI ☐Ultrasound ☐CT scan

Describe all abnormal abdomen exam findings

Extremity Exam

Extremity exam findings ○Unknown ○Not done ○Normal ○Abnormal

Describe all abnormal extremity exam findings

Neurological Exam

Neurological exam findings ○Unknown ○Not done ○Normal ○Abnormal

Abnormal neurological exam findings

☐Unknown ☐Hypotonia ☐Poor head control
☐Hypertonia ☐Tacheomalacia ☐Seizures (via abnormal EEG)
☐Abnormal reflexes ☐Myoclonic jerks ☐Other

Abnormal reflexes

☐Increased reflexes ☐Clonus ☐Moro ☐Walking/stepping ☐Rooting
☐Sucking ☐Babinski

Abnormal neurological exam findings-other, specify

Describe all abnormal neurological exam findings

Skin Exam

Skin exam findings ○ Unknown ○ Not done ○ Normal ○ Abnormal

Describe all abnormal skin exam findings

Genitourinary Exam

Genitourinary Exam Findings ○ Unknown ○ Not done ○ Normal ○ Abnormal

Describe all abnormal genitourinary exam findings

Comments

Physical exam comments

GALC - Consultations

Consultations

☐Unknown ☐None ☐Six-minute walk test ☐Opthalmologic exam ☐Echocardiogram
☐Electrocardiogram ☐Stress test ☐24 hour Holter ☐Other

Six-minute Walk Test

Six-minute walk test date |M|M| |D|D| |Y|Y|

Device used

○None ○Straight cane ○Wide-based cnae ○One crutch ○Two crutches
○Standard walker ○Rolling walker ○Othotics ○Other

Abnormalities found in the six-minute walk test ○Unknown ○Yes ○No

Abnormalities found in the six-minute walk test-specify

Resting heart rate_____

Heart rate after six-minute walk test_____

O2 set_____

Number of meters walked_____

Opthalmologic Exam

Ophthalmologic exam date |M|M| |D|D| |Y|Y|

Age at time of ophthalmologic exam_____

Abnormal opthalmologic exam findings

☐Unknown ☐None
☐Cherry red spot on left retina ☐Cherry red spot on right retina
☐Macular halo on left eye ☐Macular halo on right eye
☐Corneal abnormalities ☐Abnormalities in other eye structures (iris, lens, retina)
☐Ptosis ☐Astigmatous
☐Opticatrophy

Abnormalities to the structures of the eye-specify

Vision in the patient's right eye

(Continued on next page.)

○20/10 ○20/15 ○20/20 ○20/25 ○20/30 ○20/35
○20/40 ○20/45 ○20/50 or worse

Vision in the patient's left eye

○20/10 ○20/15 ○20/20 ○20/25 ○20/30 ○20/35
○20/40 ○20/45 ○20/50 or worse

Patient's intraocular pressure (IOP) in mmHg_____

Describe the patient's IOP ○Unknown ○Ocular hypertension (OHT) ○Ocular hypotony ○Normal eye pressure

Patient has a field of vision within the normal range ○Unknown ○Yes ○No

Explain in what ways the patient exhibits an incomplete visual field

Patient is able to see all colors ○Unknown ○Yes ○No

Colors patient is unable to see ☐Unknown ☐Red ☐Orange ☐Yellow ☐Green ☐Blue ☐Indigo ☐Violet

Eyes are roughly equal in size, shape, and appearance ○Unknown ○Yes ○No

Describe any differences in eye shape, size, or appearance between the left and right eyes

Eyes exhibit saccadic movement ○Unknown ○Yes ○No

Describe non-saccadic eye movements

Describe any other clinically relevant observations related to eye appearance, function, pressure, vision, or coordination

Echocardiogram

Echocardiogram date MM DD YY

Echocardiogram type ○2D ○M-mode

Patient height during echocardiogram_____

Patient weight during echocardiogram_____

Valvular heart disease present ○Unknown ○Yes ○No

Valves affected by valvular heart disease

☐Unknown ☐Mitral valve ☐Tricuspid valve ☐Aortic valve ☐Pulmonary valve

Mitral valve abnormalities

☐Unknown ☐Mitral valve regurgitation ☐Mitral valve stenosis ☐Mitral valve prolapse

☐Other

Severity of the mitral valve regurgitation ○Mild ○Moderate ○Severe ○Unknown

Severity of the mitral valve stenosis ○Mild ○Moderate ○Severe ○Unknown

Severity of the mitral valve prolapse ○Mild ○Moderate ○Severe ○Unknown

Describe any mitral valve abnormalities

Tricuspid valve abnormalities

☐Unknown ☐Tricuspid valve regurgitation ☐Tricuspid valve stenosis

☐Tricuspid valve prolapse ☐Other

Severity of the tricuspid valve regurgitation ○Mild ○Moderate ○Severe ○Unknown

Severity of the tricuspid valve stenosis ○Mild ○Moderate ○Severe ○Unknown

Severity of the tricuspid valve prolapse ○Mild ○Moderate ○Severe ○Unknown

Describe any tricuspid valve abnormalities

Aortic valve abnormalities

☐Unknown ☐Aortic valve regurgitation ☐Aortic valve stenosis

☐Aortic valve prolapse ☐Other

Severity of the aortic valve regurgitation ○Mild ○Moderate ○Severe ○Unknown

Severity of the aortic valve stenosis ○Mild ○Moderate ○Severe ○Unknown

Severity of the aortic valve prolapse ○Mild ○Moderate ○Severe ○Unknown

Describe any aortic valve abnormalities

Pulmonary valve abnormalities

☐Unknown ☐Pulmonary valve regurgitation ☐Pulmonary valve stenosis

☐Pulmonary valve prolapse ☐Other

Severity of the pulmonary valve regurgitation ○Mild ○Moderate ○Severe ○Unknown

Severity of the pulmonary valve stenosis ○Mild ○Moderate ○Severe ○Unknown

Severity of the pulmonary valve prolapse ○Mild ○Moderate ○Severe ○Unknown

Describe any pulmonary valve abnormalities

Describe any other relevant findings of the echocardiogram

Electrocardiogram

Electrocardiogram date M M D D Y Y

Type of electrocardiogram test ○Standard electrocardiogram ○Holter monitor

Heart rate (bpm)_____

PR Interval (msec)_____

QT Interval (msec)_____

QTc Interval (msec)_____

Conduction abnormalities

☐Unknown ☐None ☐Atrioventricular block

☐Right bundle branch block (RBBB) ☐Left bundle branch block (LBBB) ☐Wolff-Parkinson-White (WPW)

☐Junctional rhythm ☐Other

Conduction abnormalities-other, specify_____

Atrial rhythm abnormalities

☐Unknown ☐None ☐Sinus bradycardia (HR<60)

☐Sinus tachycardia (HR>100) ☐Atrial flutter ☐Premature atrial contraction (PAC)

☐Atrial fibrillation (A-Fib) ☐Supraventricular tachycardia ☐Ectopic atrial rhythm tachycardia

☐Other

Atrial rhythm abnormalities-other, specify_____

Ventricular rhythm abnormalities

☐Unknown

☐None

☐Premature ventricular contraction (PVC)

☐Ventricular tachycardia (V-Tach)

☐Ventricular fibrillation (V-Fib)

(Continued on next page.)

☐Non-sustained ventricular tachycardia (NSVT) (greater than or equal to 3 beats)

☐Other

Ventricular rhythm abnormalities-other, specify_____

Electrocardiogram findings of note

Stress Test

Date of stress test |M|M| |D|D| |Y|Y|

Type of stress test ○Unknown ○Exercise ○Adenosine/Persantine ○Dobutamine ○Other

Type of stress test-other, specify_____

Stress test protocol_____

Resting systolic blood pressure_____

Resting diastolic blood pressure_____

Peak exercise systolic blood pressure_____

Peak exercise diastolic blood pressure_____

Resting heart rate (bpm)_____

Peak heart rate (bpm)_____

Percent of maximum predicted heart rate (MPHR)_____

Symptoms during exercise ○Unknown ○Yes ○No

Evidence of ischemia ○Unknown ○Yes ○No

Describe all other relevant stress test findings

Describe all relevant 24 hour Holter results

Other Consultations

Describe all other relevant consultations

Pulmonary Function Test

Pulmonary function testing (PFT) completed at this visit ○Yes ○No

Reason PFT not completed ☐Not covered ☐Current illness ☐Unable to schedule ☐Unable to cooperate ☐Other

Date of Pulmonary function testing |M|M| |D|D| |Y|Y|

Age at time of pulmonary function testing_____

Name of lab performing PFT_____

Providers were certified ○Unknown ○Yes ○No

PFT met criteria for reliability and quality testing ○Unknown ○Yes ○No

Patient conformation ○Upright ○Supine

Measurement used for predicted values ☐Straight arm span ☐Height

Straight arm span_____

Height_____

FVC-Actual (L)_____

FVC-Predicted (L)_____

FVC-% Predicted_____

FEV1-Actual (L)_____

FEV1-Predicted (L)_____

FEV1-% Predicted_____

FEV1/FVC- Actual_____

FEV1/FVC- Predicted_____

FEF 25-75%-Actual (L/s)_____

FEF 25-75%-Predicted (L/s)_____

FEF 25-75%-% Predicted_____

FET100%-Actual (s)_____

PEF-Actual (L/s)_____

PEF-Predicted (L/s)_____

PEF-% Predicted_____

FIVC-Actual (L)_____

FIVC-Predicted (L)_____

FIVC-% Predicted_____

FIF50% - Actual (L/s)_____

FEF50% - Actual (L/s)_____

FEF50% - Predicted (L/s)_____

FEF50% - % Predicted_____

FEF/FIF50-Actual_____

MVV-Predicted (L/min)_____

TLC-Actual (L)_____

TLC-Predicted (L)_____

TLC-% Predicted_____

VC-Actual (L)_____

VC-Predicted (L)_____

VC-% Predicted_____

FRC PL-Actual (L)_____

FRC PL-Predicted (L)_____

FRC PL-% Predicted_____

FRC N2-Predicted (L)_____

RV-Actual (L)_____

RV-Predicted (L)_____

RV-% Predicted_____

RV/TLC-Actual_____

RV/TLC-Predicted_____

ERV-Actual (L)_____

ERV-Predicted (L)_____

ERV-% Predicted_____

Vtg-Actual (L)_____

IC-Actual (L)_____

IC-Predicted (L)_____

IC-% Predicted_____

DLCO-Actual (mL/mmHg/min)_____

DLCO-Predicted (mL/mmHg/min)_____

DLCO-% Predicted_____

DL Adj-Actual (mL/mmHg/min)_____

DL Adj-Predicted (mL/mmHg/min)_____

DL Adj-% Predicted_____

VA-Actual (L)_____

DLCO/VA-Actual (mL/mHg/min/L)_____

DLCO/VA-Predicted (mL/mHg/min/L)_____

DLCO/VA-% Predicted_____

PI Max-Predicted (cmH2O)_____

PE Max-Predicted (cmH2O)_____

Authors used for predicted values

PFT Comments

Comments

Consultations comments

GALC - Treatment

Hematopoeietic stem cell transplant ○Not done ○Done

Hematopoeietic stem cell transplant-Donor type ○Cord blood ○Bone marrow ○PBSC

Hematopoeietic stem cell transplant-Regimen ○Ablative ○Reduced intensity

Hematopoeietic stem cell transplant-Chimerism ○Full donor ○Mixed donor

Hematopoeietic stem cell transplant-Enzyme levels ○Normal ○Low

Common LSD Medications

Common LSD Medications

□Unknown	□None	□ACE inhibitor	□Anti-arrhythmic
□Anti-coagulant	□Anti-depressant	□Anti-diarrheal	□Anti-migraine
□Anti-platelet	□ARB	□Beta blocker	□Bisphosphonates
□Ca channel blocker	□Cerezyme	□Digestive enzymes	□Digitalis
□Diuretic	□Fabrazyme	□Folic acid	□Fosamax
□Lasix (furosemide)	□Lyrica (pregabalin)	□Methotrexate	□Myozyme/Lumizyme
□Neurontin (gabapentin)	□Rituximab	□Statins	□Vitamin D
□VPRIV	□Other		

ACE inhibitor dose_____

ACE inhibitor dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

ACE inhibitor frequency ○Unknown ○Once/day ○Twice/day ○Three times/day ○Four times/day ○Other

ACE inhibitor frequency-other, specify_____

ACE inhibitor start date |M|M| |D|D| |Y|Y|

ACE inhibitor end date |M|M| |D|D| |Y|Y|

Anti-arrhythmic dose_____

Anti-arrhythmic dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

Anti-arrhythmic frequency ○Unknown ○Once/day ○Twice/day ○Three times/day ○Four times/day ○Other

Anti-arrhythmic frequency-other, specify_____

Anti-arrhythmic start date |M|M| |D|D| |Y|Y|

Anti-arrhythmic end date |M|M| |D|D| |Y|Y|

Anti-coagulant dose_____

Anti-coagulant dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

Anti-coagulant frequency ○Unknown ○Once/day ○Twice/day ○Three times/day ○Four times/day ○Other

Anti-coagulant frequency-other, specify_____

Anti-coagulant start date |M|M| |D|D| |Y|Y|

Anti-coagulant end date |M|M| |D|D| |Y|Y|

Anti-depressant dose_____

Anti-depressant dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

Anti-depressant frequency ○Unknown ○Once/day ○Twice/day ○Three times/day ○Four times/day ○Other

Anti-depressant frequency-other, specify_____

Anti-depressant start date |M|M| |D|D| |Y|Y|

Anti-depressant end date |M|M| |D|D| |Y|Y|

Anti-diarrheal dose_____

Anti-diarrheal dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

Anti-diarrheal frequency ○Unknown ○Once/day ○Twice/day ○Three times/day ○Four times/day ○Other

Anti-diarrheal frequency-other, specify_____

Anti-diarrheal start date |M|M| |D|D| |Y|Y|

Anti-diarrheal end date |M|M| |D|D| |Y|Y|

Anti-migraine dose_____

Anti-migraine dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

Anti-migraine frequency ○Unknown ○Once/day ○Twice/day ○Three times/day ○Four times/day ○Other

Anti-migraine frequency-other, specify_____

Anti-migraine start date |M|M| |D|D| |Y|Y|

Anti-migraine end date |M|M| |D|D| |Y|Y|

Anti-platelet dose_____

Anti-platelet dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

Anti-platelet frequency ○Unknown ○Once/day ○Twice/day ○Three times/day ○Four times/day ○Other

Anti-platelet frequency-other, specify_____

Anti-platelet start date |M|M| |D|D| |Y|Y|

Anti-platelet end date |M|M| |D|D| |Y|Y|

ARB dose_____

ARB dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

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

ARB frequency-other, specify_____

ARB start date |M|M| |D|D| |Y|Y|

ARB end date |M|M| |D|D| |Y|Y|

Beta blocker dose_____

Beta blocker dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

Beta blocker frequency ○Unknown ○Once/day ○Twice/day ○Three times/day ○Four times/day ○Other

Beta blocker frequency-other, specify_____

Beta blocker start date |M|M| |D|D| |Y|Y|

Beta blocker end date |M|M| |D|D| |Y|Y|

Bisphosphonates dose_____

Bisphosphonates dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

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

Bisphosphonates frequency-other, specify_____

Bisphosphonates start date |M|M| |D|D| |Y|Y|

Bisphosphonates end date |M|M| |D|D| |Y|Y|

Ca channel blocker dose_____

Ca channel blocker dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

Ca channel blocker frequency

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

○Other

Ca channel blocker frequency-other, specify_____

Ca channel blocker start date |M|M| |D|D| |Y|Y|

Ca channel blocker end date |M|M| |D|D| |Y|Y|

Cerezyme dose_____

Cerezyme dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

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

Cerezyme frequency-other, specify_____

Cerezyme start date |M|M| |D|D| |Y|Y|

Cerezyme end date |M|M| |D|D| |Y|Y|

Digestive enzymes dose_____

Digestive enzymes dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

Digestive enzymes frequency

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

○Other

Digestive enzymes frequency-other, specify_____

Digestive enzymes start date |M|M| |D|D| |Y|Y|

Digestive enzymes end date |M|M| |D|D| |Y|Y|

Digitalis dose_____

Digitalis dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

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

Digitalis frequency-other, specify_____

Digitalis start date |M|M| |D|D| |Y|Y|

Digitalis end date |M|M| |D|D| |Y|Y|

Diuretic dose_____

Diuretic dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

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

Diuretic frequency-other, specify_____

Diuretic start date |M|M| |D|D| |Y|Y|

Diuretic end date |M|M| |D|D| |Y|Y|

Fabrazyme dose_____

Fabrazyme dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

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

Fabrazyme frequency-other, specify_____

Fabrazyme start date |M|M| |D|D| |Y|Y|

Fabrazyme end date |M|M| |D|D| |Y|Y|

Folic acid dose_____

Folic acid dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

Folic acid frequency ○Unknown ○Once/day ○Twice/day ○Three times/day ○Four times/day ○Other

Folic acid frequency-other, specify_____

Folic acid start date |M|M| |D|D| |Y|Y|

Folic acid end date |M|M| |D|D| |Y|Y|

Fosamax dose_____

Fosamax dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

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

Fosamax frequency-other, specify_____

Fosamax start date |M|M| |D|D| |Y|Y|

Fosamax end date |M|M| |D|D| |Y|Y|

Lasix (furosemide) dose_____

Lasix (furosemide) dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

Lasix (furosemide) frequency

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

○Other

Lasix (furosemide) frequency-other, specify_____

Lasix (furosemide) start date |M|M| |D|D| |Y|Y|

Lasix (furosemide) end date |M|M| |D|D| |Y|Y|

Lyrica (pregabalin) dose_____

Lyrica (pregabalin) dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

Lyrica (pregabalin) frequency

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

○Other

Lyrica (pregabalin) frequency-other, specify_____

Lyrica (pregabalin) start date |M|M| |D|D| |Y|Y|

Lyrica (pregabalin) end date |M|M| |D|D| |Y|Y|

Methotrexate dose_____

Methotrexate dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

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

Methotrexate frequency-other, specify_____

Methotrexate start date |M|M| |D|D| |Y|Y|

Methotrexate end date |M|M| |D|D| |Y|Y|

Myozyme/Lumizyme dose_____

Myozyme/Lumizyme dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

Myozyme/Lumizyme frequency

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

○Other

Myozyme/Lumizyme frequency-other, specify_____

Myozyme/Lumizyme start date |M|M| |D|D| |Y|Y|

Myozyme/Lumizyme end date |M|M| |D|D| |Y|Y|

Neurontin (gabapentin) dose_____

Neurontin (gabapentin) dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

Neurontin (gabapentin) frequency

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

○Other

Neurontin (gabapentin) frequency-other, specify_____

Neurontin (gabapentin) start date |M|M| |D|D| |Y|Y|

Neurontin (gabapentin) end date |M|M| |D|D| |Y|Y|

Rituximab dose_____

Rituximab dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

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

Rituximab frequency-other, specify_____

Rituximab start date |M|M| |D|D| |Y|Y|

Rituximab end date |M|M| |D|D| |Y|Y|

Statins dose_____

Statins dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

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

Statins frequency-other, specify_____

Statins start date |M|M| |D|D| |Y|Y|

Statins end date |M|M| |D|D| |Y|Y|

Vitamin D dose_____

Vitamin D dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

Vitamin D frequency ○Unknown ○Once/day ○Twice/day ○Three times/day ○Four times/day ○Other

Vitamin D frequency-other, specify_____

Vitamin D start date |M|M| |D|D| |Y|Y|

Vitamin D end date |M|M| |D|D| |Y|Y|

VPRIV dose_____

VPRIV dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

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

VPRIV frequency-other, specify_____

VPRIV start date |M|M| |D|D| |Y|Y|

VPRIV end date |M|M| |D|D| |Y|Y|

LSD Medication-Other name_____

LSD Medication-Other dose_____

LSD Medication-Other dose units ○grams ○IU ○micrograms ○mg ○mg/kg ○ml ○tab ○units/kg

LSD Medication-Other frequency

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

○Other

LSD Medication-Other frequency-other, specify_____

LSD Medication-Other start date |M|M| |D|D| |Y|Y|

LSD Medication-Other end date |M|M| |D|D| |Y|Y|

Other Medications

Other medications

☐Unknown	☐None	☐Actonel	☐Analgesics
☐Antacids	☐Antianxiety	☐Antibiotics	☐Anticonvulsants
☐Antiemetics	☐Antihistamines	☐Antihypertensives	☐Antiinflammatories
☐Antipsychotics	☐Antipyretics	☐Aspirin	☐Bronchodilators
☐Calcium	☐Contraceptives-injections	☐Contraceptives-oral	☐Corticosteroids
☐Growth hormone	☐Immunosuppressives	☐Iron	☐Laxatives
☐Lipid-lowering medication	☐Sleeping medications	☐Vitamins	☐Other

Experimental Treatment

Experimental Treatment

☐None	☐Transplants done abroad	☐Other cell therapies	☐DUOC-01
☐ERT	☐Gene therapy	☐Combination therapy	☐Other

Experimental Treatment-Other, specify_____

Palliative care ○Unknown ○Yes ○No

Nutrition

Mode of nutrition delivery ☐Unknown ☐Oral ☐NG tube ☐NJ tube ☐G-tube ☐GJ tube ☐TPN

Types of milk/formula taken

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

Comments

Treatment comments

GALC - Additional Testing

Auditory Testing

Auditory testing completed ○No ○Yes

Type of auditory testing completed □ABR □BAER □Other

Type of auditory testing completed-other, specify_____

ABR results ○Unknown ○Within normal limits ○Abnormalities found

Describe abnormal ABR results

BAER results ○Unknown ○Within normal limits ○Abnormalities found

Describe abnormal BAER results

Other auditory testing results ○Unknown ○Within normal limits ○Abnormalities found

Describe abnormal other auditory testing results

Swallow Study

Swallow study completed ○No ○Yes

Swallow study results ○Unknown ○Within normal limits ○Abnormalities found

Describe abnormal swallow study results

Radiology/Imaging

Radiology/Imaging tests performed □Unknown □None □Brain MRI □EEG □NCV □VER □Other

NCV

NCV date_____

NCV results ○Unknown ○Within normal limits ○Abnormalities found

Please describe any abnormalities found in the NCV

VER

VER date_____

VER results ○Unknown ○Within normal limits ○Abnormalities found

Please describe any abnormalities found in the VER

EEG

Evidence of seizures ○Yes ○No

Describe abnormal EEG results

Brain MRI

Brain MRI results ○Unknown ○Normal ○Abnormal

Describe the abnormal brain MRI results

Brain Imaging

Describe any white matter changes

Evidence of hypomyelination ○Yes ○No

Evidence of calcification ○Yes ○No

Other Radiology/Imaging Tests

Describe the other radiology/imaging tests conducted and findings

Comments

Additional testing comments

GALC - Labs

Lipid Panel

Fasting lipid panel results

☐Unknown ☐Not done ☐Total cholesterol ☐Triglycerides ☐HDL
☐LDL cholesterol ☐LDL triglyercides ☐VLDL cholesterol ☐VLDL triglycerides

Date of fasting lipid panel |M|M| |D|D| |Y|Y|

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_____

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

Triglycerides value_____

Triglycerides units_____

Triglycerides reference range_____

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

HDL value_____

HDL units_____

HDL reference range_____

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

LDL cholesterol value_____

LDL cholesterol units_____

LDL cholesterol reference range_____

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

LDL triglycerides value_____

LDL triglycerides units_____

LDL triglycerides reference range_____

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

VLDL cholesterol value_____

VLDL cholesterol units_____

VLDL cholesterol reference range_____

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

VLDL triglycerides value_____

VLDL triglycerides units_____

VLDL triglycerides reference range_____

Hematology

Complete blood count (CBC) results

☐Unknown ☐Not done ☐White blood cell count (WBCC)
☐Hemoglobin ☐Hematocrit ☐Red blood cell count (RBCC)
☐Platelet count ☐Neutrophil # ☐Lymphocyte #
☐Monocyte # ☐Eosinophil # ☐Basophil #

CBC collection date |M|M| |D|D| |Y|Y|

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

White blood cell count value_____

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

White blood cell count reference range_____

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

Hemoglobin value_____

Hemoglobin units ○g/dL ○g/L

Hemoglobin reference range_____

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

Hematocrit value_____

Hematocrit units ○% ○Proportion of total hemoglobin

Hematocrit reference range_____

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

Red blood cell count value_____

Red blood cell count units ○10^6/uL ○10^12/uL

Red blood cell count reference range_____

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_____

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

Neutrophil # value_____

Neutrophil # units_____

Neutrophil # reference range_____

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

Lymphocyte # value_____

Lymphocyte # units_____

Lymphocyte # reference range_____

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

Monocyte # value_____

Monocyte # units_____

Monocyte # reference range_____

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

Eosinophil # value_____

Eosinophil # units_____

Eosinophil # reference range_____

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

Basophil # value_____

Basophil # units_____

Basophil # reference range_____

Urinalysis

Urinalysis performed ○Unknown ○Yes ○No

Urine collection date M M D D Y Y

Urine color ○Unknown ○Dark yellow ○Pale yellow ○Nearly colorless ○Brown ○Pink

Urine clarity ○Clear ○Slightly cloudy ○Cloudy ○Turbid

Blood in the urine ○Unknown ○Yes ○No

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

Urine RBCC value_____

Urine RBCC units_____

Urine RBCC reference range_____

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

Urine WBCC value_____

Urine WBCC units_____

Urine WBCC reference range_____

Microorganisms in the urine ○Unknown ○None ○Few ○Moderate ○Many

Hyaline casts per low per field_____

Crystals in the urine ○Unknown ○Yes ○No

Crystals in the urine-specify

Urinalysis results

☐Unknown ☐Specific gravity ☐pH ☐Protein ☐Glucose

(Continued on next page.)

Patient Name_____

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

☐Ketones ☐Leukocyte esterase ☐Nitrite ☐Bilirubin ☐Ubrolinogen

☐Creatinine Kinase ☐Hex4

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

Specific gravity value_____

Specific gravity units_____

Specific gravity reference range_____

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

pH value_____

pH units_____

pH reference range_____

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

Protein value_____

Protein units_____

Protein reference range_____

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

Glucose value_____

Glucose units_____

Glucose reference range_____

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

Ketones value_____

Ketones units_____

Ketones reference range_____

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

Leukocyte esterase value_____

Leukocyte esterase units_____

Leukocyte esterase reference range_____

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

Nitrite value_____

Nitrite units_____

Nitrite reference range_____

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

Bilirubin value_____

Bilirubin units_____

Bilirubin reference range_____

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

Ubrolinogen value_____

Ubrolinogen units_____

Ubrolinogen reference range_____

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

Creatine Kinase value_____

Creatine Kinase units_____

Creatine Kinase reference range_____

Hex4 ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Hex4 value_____

Hex4 units_____

Hex4 reference range_____

Biomarkers

Krabbe biomarkers □Unknown □Not done □Chimerism □CSF Protein □GALC enzyme activity

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

Chimerism value_____

Chimerism units_____

Chimerism reference range_____

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

CSF Protein value_____

CSF Protein units_____

CSG Protein reference range_____

GALC enzyme activity ○Within normal limits ○Abnormal ○In progress ○Results unavailable

GALC enzyme activity value_____

GALC enzyme activity units_____

GALC enzyme activity reference range_____

Other Labs

Other labs collected ○Unknown ○Yes ○No

Number of other labs collected ○1 ○2 ○3 ○4 or more

Other lab 1 name_____

Other lab 1 significance ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Other lab 1 value_____

Other lab 1 units_____

Other lab 1 reference range_____

Other lab 2 name_____

Other lab 2 significance ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Other lab 2 value_____

Other lab 2 units_____

Other lab 2 reference range_____

Other lab 3 name_____

Other lab 3 significance ○Within normal limits ○Abnormal ○In progress ○Results unavailable

Other lab 3 value_____

Other lab 3 units_____

Other lab 3 reference range_____

Other lab name(s), value(s), unit(s), and reference range(s)

Home Monitoring

Home monitoring recommended ○Unknown ○Yes ○No

Home monitoring done since the last outpatient visit ○Unknown ○Yes ○No

Type of home monitoring_____

Comments

Labs comments

GALC - Sick Visits And Hospitalizations

Sick Visits

Sick visits since last outpatient visit ○Unknown ○Yes ○No

Number of sick visits_____

Date of sick visit 1 |M|M| |D|D| |Y|Y|

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

Sick visit 1 was a condition exacerbation ○Unknown ○Yes ○No

Location for sick visit 1

○Unknown ○Emergency department ○Retail clinic

○Primary care ○Specialty center ○Urgent care

○Direct hospital admission ○Other

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

Name of hospital for sick visit 1_____

ICD-9 codes for sick visit 1 known ○Yes ○No

ICD-9 codes for sick visit 1_____

Diagnosis for sick visit 1_____

Number of inpatient days for sick visit 1_____

Number of ICU days for sick visit 1_____

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

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

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

Location for sick visit 2

○Unknown ○Emergency department ○Retail clinic ○Primary care

○Specialty center ○Urgent care ○Direct hospital admission ○Other

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

Name of hospital for sick visit 2_____

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

ICD-9 codes for sick visit 2_____

Diagnosis for sick visit 2_____

Number of inpatient days for sick visit 2_____

Number of ICU days for sick visit 2_____

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

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

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

Location for sick visit 3

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

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

Name of hospital for sick visit 3_____

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

ICD-9 codes for sick visit 3_____

Diagnosis for sick visit 3_____

Number of inpatient days for sick visit 3_____

Number of ICU days for sick visit 3_____

Hospitalizations and Surgeries

Hospitalizations since last visit ○ Unknown ○ Yes ○ No

Number of hospitalizations since last visit ○ 1 ○ 2 ○ 3 ○ More than 3

Location of first hospitalization_____

Date of admission for first hospitalization M M D D Y Y

Date of discharge for first hospitalization M M D D Y Y

Reason for first hospitalization

Location of second hospitalization_____

Date of admission for second hospitalization M M D D Y Y

Date of discharge for second hospitalization M M D D Y Y

Reason for second hospitalization

Location of third hospitalization_____

Date of admission for third hospitalization M M D D Y Y

Date of discharge for third hospitalization M M D D Y Y

Reason for third hospitalization

Date(s), location(s), and reason(s) for additional hospitalizations

Surgeries since last visit ○Unknown ○Yes ○No

Number of surgeries since last visit ○1 ○2 ○3 ○More than 3

Location of first surgery_____

Date of first surgery |M|M| |D|D| |Y|Y|

Reason for first surgery

○Infusaport	○G-tube continuous	○G-tube bolus
○G/J-tube	○Tracheostomy	○Hip surgery
○Tendon release	○Scoliosis	○Hernia
○Joint surgery (carpal tunnel, hips)	○Appendectomy	○Cardiac valve transplant
○Tonsillectomy and adenoidectomy	○Cervical spine stability	○Hearing tubes
○Other		

Reason for first surgery-Other, specify_____

Location of second surgery_____

Date of second surgery |M|M| |D|D| |Y|Y|

Reason for second surgery

○Infusaport	○G-tube continuous	○G-tube bolus
○G/J-tube	○Tracheostomy	○Hip surgery
○Tendon release	○Scoliosis	○Hernia
○Joint surgery (carpal tunnel, hips)	○Appendectomy	○Cardiac valve transplant
○Tonsillectomy and adenoidectomy	○Cervical spine stability	○Hearing tubes
○Other		

Reason for second surgery-Other, specify_____

Location of third surgery_____

Date of third surgery |M|M| |D|D| |Y|Y|

Reason for third surgery

○Infusaport	○G-tube continuous	○G-tube bolus
○G/J-tube	○Tracheostomy	○Hip surgery
○Tendon release	○Scoliosis	○Hernia
○Joint surgery (carpal tunnel, hips)	○Appendectomy	○Cardiac valve transplant
○Tonsillectomy and adenoidectomy	○Cervical spine stability	○Hearing tubes
○Other		

Reason for third surgery-Other, specify_____

Date(s), location(s), and reason(s) for additional surgeries