

Patient Name _____

Date

ASM - Demographics

NBSTRN ID _____

Intake date

Consent

Consent obtained Yes No IRB ExemptAssent obtained Yes No Not applicableType of assent Written VerbalPermission to recontact Unknown Yes No

Demographics Information

Patient last name _____

Patient first name _____

Date of birth

Gestational Age (weeks) _____

Gestational Age (days) _____

Societal sex Unknown Male Female Ambiguous

Biological mother's maiden name _____

Condition

Patient condition category SACHDNC candidate disordersSpecify SACHDNC candidate disorder diagnosis for the patient Niemann Pick disease Types A and B

Patient disorder identification method

 Unknown Abnormal newborn screen Abnormal labs Clinical presentation Family member with this condition Missed by newborn screening

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 NoOther research studies are clinical trials Unknown Yes No

Research study-other, specify _____

Clinicaltrials.gov identifier _____

Education

Maternal education

 Unknown

Patient Name _____

Date

- 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 Philippines Russian Federation
 Syria Thailand Vietnam Other

Ancestral Origin-Asia-Other, specify _____

Ancestral Origin-Europe

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

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

Ancestral Origin-Europe-Italy Unknown 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 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 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 White

Race-White, specify Ashkenazi Jewish Non-Ashkenazi JewishPatient 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

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"/>

- 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

ASM - Enzymemutation Analysis

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 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

ASM on first newborn screen _____

ASM (% daily mean) on first newborn screen _____

Date second newborn screen collected

Reason for second newborn screen

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

Reason for second newborn screen-other, specify _____

ASM on second newborn screen _____

ASM (% daily mean) on second newborn screen _____

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 _____

ASM on third newborn screen _____

ASM (% daily mean) on third newborn screen _____

Enzymatic Confirmatory Testing

Date of enzymatic confirmatory testing

Confirmatory testing ASM _____

Confirmatory testing ASM normal reference range _____

Lab where enzymatic confirmation was done

 Icahn School of Medicine Thomas Jefferson University Mayo Clinic Greenwood Emory Duke Other

Patient Name _____

Date | |

Lab where enzymatic confirmation was done-other, specify _____

Newborn Hearing ScreenNewborn 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 OtherPatient 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

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- | | | |
|---|---|---|
| <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 |
| <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"/> Trichorrhexis nodosa | <input type="checkbox"/> Vision loss |
| <input type="checkbox"/> Vomiting | <input type="checkbox"/> Other | |

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

Mutation Testing

Type of genetic/genomic testing

- | | | |
|---|--|---|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Not done | <input type="checkbox"/> Done, not available |
| <input type="checkbox"/> Single gene | <input type="checkbox"/> Mutation panel | <input type="checkbox"/> Exome sequencing |
| <input type="checkbox"/> Full genome sequencing | <input type="checkbox"/> Copy number variant | <input type="checkbox"/> Deletion/duplication testing |
| <input type="checkbox"/> Other | | |

Chitotriosidase gene mutation analysis date Chitotriosidase gene mutation analysis Null, Null Mutation, Null Mutation, Mutation

Patient Name _____

Date Number of mutations in the SMPD1 gene 0 1 2

SMPD1 mutation zygosity

 Homozygous Compound heterozygous Homozygous or heterozygous + additional variantsType of first SMPD1 mutation Missense Nonsense Splicing error Deletion Insertion

First SMPD1 mutation-missense (HGVS (nucleotide) / HGVS (protein))

- 2T>G / M1R 152A>T / D51V 282C>G / C94W 314T>C / L105P
 340G>A / V114M 395T>C / V132A 416T>C / L139P 475T>C / C159R
 502G>A / G168R 533T>A / I178N 557C>T / P186L 592G>C / A198P
 604C>T / R202C 631T>C / W211R 679C>A / L227M 680T>C / L227P
 688C>T / R230C 689G>A / R230H 701G>A / G234D 728C>T / A243V
 730G>A / G244R 738G>C / W246C 739G>A / G247S 740G>A / G247D
 742G>A / E248K 748A>C / S250R 750C>A / S250R 757G>C / D253H
 759C>A / D253E 839A>C / D280A 847G>A / A283T 872G>A / R291H
 880C>A / Q294K 887G>A / R296Q 911T>C / L304P 940G>A / V314M
 943T>C / Y315H 961C>T / H321Y 973C>G / P325A 995C>G / P332R
 1028T>C / L343P 1076C>A / A359D 1076C>T / A359V 1106A>G / Y369C
 1117C>T / P373S 1133G>A / R378H 1133G>T / R378L 1141T>C / S381P
 1142C>T / S381F 1152G>A / M384I 1154A>G / N385S 1172A>C / N391T
 1177T>G / W393G 1244C>T / A415V 1268A>G / H423R 1267C>T / H423Y
 1280A>G / H427R 1297T>C / C433R 1301T>C / L434P 1311G>C / W437C
 1314C>A / S438R 1343A>G / Y448C 1355T>C / L452P 1358C>A / A453D
 1361C>T / A454V 1373G>A / G458D 1394T>C / F465S 1406A>C / Y469S
 1426C>T / R476W 1430C>T / P477L 1446C>A / F482L 1451C>A / A484E
 1460C>T / A487V 1462A>G / T488A 1468T>A / Y490N 1486G>T / G496C
 1486G>A / G496S 1492C>T / R498C 1493G>A / R498H 1493G>T / R498L
 1548T>G / H516Q 1550A>T / E517V 1556A>G / Y519C 1565A>G / N522S
 1575G>C / Q525H 1603T>C / W535R 1615T>C / Y539H 1652T>C / L551P
 1666C>T / H556Y 1693G>T / D565Y 1716C>G / F572L 1730A>G / H577R
 1730A>T / H577L 1734G>C / K578N 1735G>A / G579S 1805G>A / R602H
 1805G>C / R602P Other

First SMPD1 mutation-missense-other, specify

Patient Name _____

Date | |

First SMPD1 mutation-nonsense (HGVS (nucleotide) / HGVS (protein))

- 61C>T / Q21X 96G>A / W32X 509G>A / W170X 528G>A / W176X
 778G>T / E260X 788T>A / L263X 945C>A / Y315X 1252C>T / R418X
 1327C>T / R443X 1624C>T / R542X 1718G>A / W573X Other

First SMPD1 mutation-nonsense-other, specify

First SMPD1 mutation-splicing error (HGVS (nucleotide) / HGVS (protein)) 1092-1G>C Other

First SMPD1 mutation-splicing error-other, specify

First SMPD1 mutation-deletion (HGVS (nucleotide) / HGVS (protein))

- 84delC 101delG 511delG
 538_539delTT 573delT 581delC
 968delC 996delC 1111_1112delCT
 1146delC 1165delC 1175_1177delTCT
 1263+4_1263+7delAGGG 1380delT 1596delA
 1630delA 1657_1663delACCGCCT 1675_1676delGT
 1780_1782delACT 1785_1786delTT 1829_1831delGCC
 Other

First SMPD1 mutation-deletion-other, specify

First SMPD1 mutation-insertion (HGVS (nucleotide) / HGVS (protein))

- 518dupT 572dupC 581dupC 1103dupT 1864_1877dup14
 Other

First SMPD1 mutation-insertion-other, specify

Inheritance of first SMPD1 mutation Unknown Maternal PaternalType of second SMPD1 mutation Missense Nonsense Splicing error Deletion Insertion

Patient Name _____

Date

Second SMPD1 mutation-missense (HGVS (nucleotide) / HGVS (protein))

- 2T>G / M1R 152A>T / D51V 282C>G / C94W 314T>C / L105P
 340G>A / V114M 395T>C / V132A 416T>C / L139P 475T>C / C159R
 502G>A / G168R 533T>A / I178N 557C>T / P186L 592G>C / A198P
 604C>T / R202C 631T>C / W211R 679C>A / L227M 680T>C / L227P
 688C>T / R230C 689G>A / R230H 701G>A / G234D 728C>T / A243V
 730G>A / G244R 738G>C / W246C 739G>A / G247S 740G>A / G247D
 742G>A / E248K 748A>C / S250R 750C>A / S250R 757G>C / D253H
 759C>A / D253E 839A>C / D280A 847G>A / A283T 872G>A / R291H
 880C>A / Q294K 887G>A / R296Q 911T>C / L304P 940G>A / V314M
 943T>C / Y315H 961C>T / H321Y 973C>G / P325A 995C>G / P332R
 1028T>C / L343P 1076C>A / A359D 1076C>T / A359V 1106A>G / Y369C
 1117C>T / P373S 1133G>A / R378H 1133G>T / R378L 1141T>C / S381P
 1142C>T / S381F 1152G>A / M384I 1154A>G / N385S 1172A>C / N391T
 1177T>G / W393G 1244C>T / A415V 1268A>G / H423R 1267C>T / H423Y
 1280A>G / H427R 1297T>C / C433R 1301T>C / L434P 1311G>C / W437C
 1314C>A / S438R 1343A>G / Y448C 1355T>C / L452P 1358C>A / A453D
 1361C>T / A454V 1373G>A / G458D 1394T>C / F465S 1406A>C / Y469S
 1426C>T / R476W 1430C>T / P477L 1446C>A / F482L 1451C>A / A484E
 1460C>T / A487V 1462A>G / T488A 1468T>A / Y490N 1486G>T / G496C
 1486G>A / G496S 1492C>T / R498C 1493G>A / R498H 1493G>T / R498L
 1548T>G / H516Q 1550A>T / E517V 1556A>G / Y519C 1565A>G / N522S
 1575G>C / Q525H 1603T>C / W535R 1615T>C / Y539H 1652T>C / L551P
 1666C>T / H556Y 1693G>T / D565Y 1716C>G / F572L 1730A>G / H577R
 1730A>T / H577L 1734G>C / K578N 1735G>A / G579S 1805G>A / R602H
 1805G>C / R602P Other

Second SMPD1 mutation-missense-other, specify

Second SMPD1 mutation-nonsense (HGVS (nucleotide) / HGVS (protein))

- 61C>T / Q21X 96G>A / W32X 509G>A / W170X 528G>A / W176X
 778G>T / E260X 788T>A / L263X 945C>A / Y315X 1252C>T / R418X
 1327C>T / R443X 1624C>T / R542X 1718G>A / W573X Other

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Second SMPD1 mutation-nonsense-other, specify

Second SMPD1 mutation-splicing error (HGVS (nucleotide) / HGVS (protein)) 1092-1G>C Other

Second SMPD1 mutation-splicing error-other, specify

Second SMPD1 mutation-deletion (HGVS (nucleotide) / HGVS (protein))

- 84delC 101delG 511delG
 538_539delTT 573delT 581delC
 968delC 996delC 1111_1112delCT
 1146delC 1165delC 1175_1177delTCT
 1263+4_1263+7delAGGG 1380delT 1596delA
 1630delA 1657_1663delACCGCCT 1675_1676delGT
 1780_1782delACT 1785_1786delTT 1829_1831delGCC
 Other

Second SMPD1 mutation-deletion-other, specify

Second SMPD1 mutation-insertion (HGVS (nucleotide) / HGVS (protein))

- 518dupT 572dupC 581dupC 1103dupT 1864_1877dup14
 Other

Second SMPD1 mutation-insertion-other, specify

Inheritance of second mutation Unknown Maternal Paternal

Diagnosis based on biochemical and genetic testing

- Niemann-Pick A Niemann-Pick B Niemann-Pick A/B Undetermined Unaffected

Patient Name _____

Date

Undetermined diagnosis-explain

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

ASM - Initial Medical History

Date medical history performed

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 _____

Patient Name _____

Date

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

Patient Name _____

Date 5 minute APGAR score Unknown Not done 0 1 2 3 4 5 6 7 8 9 1010 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 HistoryConsanguinity Unknown Yes NoType of consanguinity First cousins or closer OtherPedigree obtained Yes NoFamily members with confirmed Niemann-Pick Unknown Yes NoType of Niemann-Pick Unknown A B A/B

Relationship of family members with confirmed unknown Neimann-Pick to proband

- Mother Father Sister Brother Grandmother Grandfather Aunt
 Uncle First cousin Half-sister Half-brother Other

Relationship of family members with confirmed unknown Neimann-Pick to proband-Other, specify _____

Relationship of family members with confirmed Neimann-Pick Type A to proband

- Mother Father Sister Brother Grandmother Grandfather Aunt
 Uncle First cousin Half-sister Half-brother Other

Relationship of family members with confirmed Neimann-Pick Type A to proband-Other, specify _____

Relationship of family members with confirmed Neimann-Pick Type B to proband

- Mother Father Sister Brother Grandmother Grandfather Aunt
 Uncle First cousin Half-sister Half-brother Other

Relationship of family members with confirmed Neimann-Pick Type B to proband-Other, specify _____

Relationship of family members with confirmed Neimann-Pick Type A/B to proband

- Mother Father Sister Brother Grandmother Grandfather Aunt
 Uncle First cousin Half-sister Half-brother Other

Relationship of family members with confirmed Neimann-Pick Type A/B to proband-Other, specify _____

Family members with symptoms suggestive of Niemann-Pick Unknown Yes No

Patient Name _____

Date Type of Niemann-Pick suggested by symptoms Unknown A B A/B

Relationship of family members with suggested unknown Niemann-Pick to proband

Mother Father Sister Brother Grandmother Grandfather Aunt
 Uncle First cousin Half-sister Half-brother Other

Relationship of family members with suggested unknown Niemann-Pick to proband-Other, specify _____

Relationship of family members with suggested Niemann-Pick Type A to proband

Mother Father Sister Brother Grandmother Grandfather Aunt
 Uncle First cousin Half-sister Half-brother Other

Relationship of family members with suggested Niemann-Pick Type A to proband-Other, specify _____

Relationship of family members with suggested Niemann-Pick Type B to proband

Mother Father Sister Brother Grandmother Grandfather Aunt
 Uncle First cousin Half-sister Half-brother Other

Relationship of family members with suggested Niemann-Pick Type B to proband-Other, specify _____

Relationship of family members with suggested Niemann-Pick Type A/B to proband

Mother Father Sister Brother Grandmother Grandfather Aunt
 Uncle First cousin Half-sister Half-brother Other

Relationship of family members with suggested Niemann-Pick Type A/B to proband-Other, specify _____

Family member(s) with history of high cholesterol

Unknown None Mother Father Sister Brother Grandmother
 Grandfather Aunt Uncle First cousin Half-sister Half-brother Other

Family member(s) with history of high cholesterol-Other, specify _____

Family member(s) with history of heart disease

Unknown None Mother Father Sister Brother Grandmother
 Grandfather Aunt Uncle First cousin Half-sister Half-brother Other

Family member(s) with history of heart disease-Other, specify _____

Family member(s) with history of stroke

Unknown None Mother Father Sister Brother Grandmother
 Grandfather Aunt Uncle First cousin Half-sister Half-brother Other

Family member(s) with history of stroke-Other, specify _____

Patient Name _____

Date

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 JewishMother is Hispanic or Latino Not reported Yes No

Paternal race

- 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 JewishFather is Hispanic or Latino Not reported Yes NoHospitalizations 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/primary caregiver was given a written emergency letter Unknown Yes NoPatient/primary caregiver was given a sick day plan specific to this condition Unknown Yes No

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

 Unknown Yes No

Comments

Initial medical history comments

Patient Name _____

Date **ASM - Followup Medical History**Visit Date 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 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 NoOther research studies are clinical trials Unknown Yes No

Identify the research study _____

Clinicaltrials.gov identifier _____

Care CoordinationMissed 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

- | | |
|---|---|
| <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 |

Patient Name _____

Date

- 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

Patient Name _____

Date

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 moreDate of transfusion 1 Date of transfusion 2 Date of transfusion 3

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

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

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

Skeletal Changes

Skeletal changes since last visit

- Unknown None Broken or fractured bone
- Joint pain Patient uses orthotics or braces Back pain

Bone broken or fractured _____

Event preceding the bone break or fracture

Approximate age of onset for joint 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 patient began using orthotics or braces

- 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 back pain

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

Patient has had a bone densitometry test Unknown Yes No

Patient Name _____

Date

Date of bone densitometry test

Neurological Changes

Neurological changes since last visit

- Unknown None Headaches more than once per week
- Seizures Stroke or TIA Abnormal heat tolerance
- Abnormal cold tolerance Acroparesthesias Acute pain crises
- Non-cardiac syncope Vertigo Hearing loss
- Tinnitus Other

Neurological changes since last visit-Other, specify _____

Approximate age of onset for headaches

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

Relevant seizure history

Relevant stroke and TIA history

Frequency with which the patient experiences acroparesthesia

- Unknown Less than once a month Monthly Weekly
- Daily

Approximate age of onset for acute pain

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

Number of acute pain crises in the past year _____

Cause of the non-cardiac syncope

- Unknown Cerebrovascular (e.g. stroke) Psychological
- Reflex/Vasovagal Other

Cause of non-cardiac syncope-other, specify _____

Patient Name _____

Date **Infectious Diseases Changes**

Infectious disease (ID) changes since last visit

- Unknown None Patient had bronchitis Patient had otitis media
 Patient had sinusitis Patient had pneumonia

Patient was hospitalized for otitis media Unknown Yes NoPatient has myringotomy tubes Unknown Yes No**Cardiovascular Changes**

Cardiovascular changes since last visit

- Unknown None Valvular heart disease Myocardial infarction
 Chest pain Cardiovascular surgery Arrhythmia Congestive heart failure
 Hypertension Cardiac syncope

Description of patient's valvular heart disease

Approximate age of onset for valvular heart disease

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

Approximate age at time of myocardial infarction

- 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 chest pain

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

Approximate age at time of cardiovascular surgery

- 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 arrhythmia

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

Patient Name _____

Date

Approximate age at time of congestive heart failure

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

Pulmonary Changes

Pulmonary changes since last visit

- Unknown None Shortness of breath
 Patient requires oxygen daily Symptoms of asthma Shortness of breath with exercise
 Pulmonary involvement Obstructive disease Restrictive disease

Describe patient's shortness of breath

Approximate age of onset for shortness of breath

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

Approximate age at which patient began using oxygen daily

- 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 asthma symptoms

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

Approximate age at which patient began experiencing shortness of breath with exercise

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

Psychiatric Changes

Psychiatric changes since last visit Unknown None Depression Anxiety Other

Describe the patient's depression

Patient Name _____

Date Patient is on medication for depression Unknown Yes NoPatient required hospitalization for depression Unknown Yes NoPatient 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

- 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 NoStandardized developmental screening tool(s) used Unknown Yes NoDevelopmental status Typical AtypicalSeverity of atypical development Unknown Mild delay Moderate delay Severe delay

Developmental milestones that were achieved in a typical order and timeframe

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

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

Referred for further developmental assessment

- Unknown Yes
 No Previously referred
 Family declined further assessments

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

- Unknown Developmental/behavioral pediatrician

Patient Name _____

Date [M][M] [D][D] [Y][Y]

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

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"/>

ASM - Physical Exam

Date of Physical Exam

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

Patient Name _____

Date

Physical Exam

Exam findings Unknown Not done Normal Abnormal

Describe all abnormal exam findings

Presence of hernias None Inguinal Umbilical Epigastric Incisional

HEENT Exam

HEENT Exam Findings Unknown Not done Normal Abnormal

Abnormal HEENT exam findings

Unknown Coarse facial features Retinal cherry red spot Enlarged tonsils

Corneal abnormalities Hearing loss Other

Abnormal HEENT exam findings-other, specify

Describe all abnormal HEENT exam findings

Neck Exam

Neck exam findings Unknown Not done Normal Abnormal

Describe all abnormal neck findings Back extremities 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

Patient Name _____

Date

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

Abnormal heart exam findings Unknown Other

Abnormal heart exam findings-other, specify

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

Abnormal abdomen exam findings Unknown Enlarged liver Enlarged spleen Other

How many cm below the costal margin is the liver?_____

How many cm below the costal margin is the spleen?_____

Abnormal abdomen exam findings-other, specify

Describe all abnormal abdomen exam findings

Extremity Exam

Extremity exam findings Unknown Not done Normal Abnormal

Abnormal extremity exam findings Unknown Thin extremities Other

Abnormal extremity exam findings-other,specify

Patient Name _____

Date

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
 Hyporeflexia
 Ataxia
 Other

Abnormal neurological exam findings-other, specify

Describe all abnormal neurological exam findings

Skin Exam

Skin exam findings Unknown Not done Normal Abnormal

Abnormal skin exam findings Unknown Petechiae Bruising Other

Abnormal skin exam findings-other, specify

Describe all abnormal skin exam findings

Genitourinary Exam

Genitourinary Exam Findings Unknown Not done Normal Abnormal

Describe all abnormal genitourinary exam findings

Patient Name _____

Date

Comments

Physical exam comments

Patient Name _____

Date

ASM - Consultations

Consultations

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

Six-minute Walk Test

Six-minute walk test date

Device used

- None Straight cane Wide-based cane 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 _____

Ophthalmologic Exam

Ophthalmologic exam date

Age at time of ophthalmologic exam _____

Abnormal ophthalmologic exam findings

- | | |
|---|---|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> None |
| <input type="checkbox"/> Cherry red spot on left retina | <input type="checkbox"/> Cherry red spot on right retina |
| <input type="checkbox"/> Macular halo on left eye | <input type="checkbox"/> Macular halo on right eye |
| <input type="checkbox"/> Corneal abnormalities | <input type="checkbox"/> Abnormalities in other eye structures (iris, lens, retina) |
| <input type="checkbox"/> Ptosis | <input type="checkbox"/> Astigmatous |
| <input type="checkbox"/> Opticatrophy | |

Abnormalities to the structures of the eye-specify

Vision in the patient's right eye

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"/>

- 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

Echocardiogram type 2D M-mode

Patient height during echocardiogram _____

Patient weight during echocardiogram _____

Valvular heart disease present Unknown Yes No

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

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

Patient Name _____

Date Severity of the pulmonary valve regurgitation Mild Moderate Severe UnknownSeverity of the pulmonary valve stenosis Mild Moderate Severe UnknownSeverity 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 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)

Patient Name _____

Date

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

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

Patient Name _____

Date **Pulmonary Function Test**Pulmonary function testing (PFT) completed at this visit Yes NoReason PFT not completed Not covered Current illness Unable to schedule Unable to cooperate OtherDate of Pulmonary function testing

Age at time of pulmonary function testing _____

Name of lab performing PFT _____

Providers were certified Unknown Yes NoPFT met criteria for reliability and quality testing Unknown Yes NoPatient conformation Upright SupineMeasurement 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 _____

Patient Name _____

Date

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)_____

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

Authors used for predicted values

PFT Comments

Comments

Consultations comments

Patient Name _____

Date

ASM - Treatment

Common LSD Medications

Common LSD Medications

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

ACE inhibitor dose _____

ACE inhibitor dose units grams IU micrograms mg mg/kg ml tab units/kgACE inhibitor frequency Unknown Once/day Twice/day Three times/day Four times/day Other

ACE inhibitor frequency-other, specify _____

ACE inhibitor start date ACE inhibitor end date

Anti-arrhythmic dose _____

Anti-arrhythmic dose units grams IU micrograms mg mg/kg ml tab units/kgAnti-arrhythmic frequency Unknown Once/day Twice/day Three times/day Four times/day Other

Anti-arrhythmic frequency-other, specify _____

Anti-arrhythmic start date Anti-arrhythmic end date

Anti-coagulant dose _____

Anti-coagulant dose units grams IU micrograms mg mg/kg ml tab units/kgAnti-coagulant frequency Unknown Once/day Twice/day Three times/day Four times/day Other

Anti-coagulant frequency-other, specify _____

Anti-coagulant start date Anti-coagulant end date

Anti-depressant dose _____

Anti-depressant dose units grams IU micrograms mg mg/kg ml tab units/kgAnti-depressant frequency Unknown Once/day Twice/day Three times/day Four times/day Other

Anti-depressant frequency-other, specify _____

Anti-depressant start date

Patient Name _____

Date Anti-depressant end date

Anti-diarrheal dose _____

Anti-diarrheal dose units grams IU micrograms mg mg/kg ml tab units/kgAnti-diarrheal frequency Unknown Once/day Twice/day Three times/day Four times/day Other

Anti-diarrheal frequency-other, specify _____

Anti-diarrheal start date Anti-diarrheal end date

Anti-migraine dose _____

Anti-migraine dose units grams IU micrograms mg mg/kg ml tab units/kgAnti-migraine frequency Unknown Once/day Twice/day Three times/day Four times/day Other

Anti-migraine frequency-other, specify _____

Anti-migraine start date Anti-migraine end date

Anti-platelet dose _____

Anti-platelet dose units grams IU micrograms mg mg/kg ml tab units/kgAnti-platelet frequency Unknown Once/day Twice/day Three times/day Four times/day Other

Anti-platelet frequency-other, specify _____

Anti-platelet start date Anti-platelet end date

ARB dose _____

ARB dose units grams IU micrograms mg mg/kg ml tab units/kgARB frequency Unknown Once/day Twice/day Three times/day Four times/day Other

ARB frequency-other, specify _____

ARB start date ARB end date

Beta blocker dose _____

Beta blocker dose units grams IU micrograms mg mg/kg ml tab units/kgBeta blocker frequency Unknown Once/day Twice/day Three times/day Four times/day Other

Beta blocker frequency-other, specify _____

Beta blocker start date Beta blocker end date

Bisphosphonates dose _____

Bisphosphonates dose units grams IU micrograms mg mg/kg ml tab units/kgBisphosphonates frequency Unknown Once/day Twice/day Three times/day Four times/day Other

Bisphosphonates frequency-other, specify _____

Bisphosphonates start date

Patient Name _____

Date

Bisphosphonates end date

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

Ca channel blocker end date

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

Cerezyme end date

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

Digestive enzymes end date

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

Digitalis end date

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

Diuretic end date

Fabrazyme dose _____

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"/>
--

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

Fabrazyme end date

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

Folic acid end date

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

Fosamax end date

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

Lasix (furosemide) end date

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

Lyrica (pregabalin) end date

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

Patient Name _____

Date

Methotrexate frequency-other, specify _____

Methotrexate start date Methotrexate end date

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 Myozyme/Lumizyme end date

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 Neurontin (gabapentin) end date

Rituximab dose _____

Rituximab dose units grams IU micrograms mg mg/kg ml tab units/kgRituximab frequency Unknown Once/day Twice/day Three times/day Four times/day Other

Rituximab frequency-other, specify _____

Rituximab start date Rituximab end date

Statins dose _____

Statins dose units grams IU micrograms mg mg/kg ml tab units/kgStatins frequency Unknown Once/day Twice/day Three times/day Four times/day Other

Statins frequency-other, specify _____

Statins start date Statins end date

Vitamin D dose _____

Vitamin D dose units grams IU micrograms mg mg/kg ml tab units/kgVitamin D frequency Unknown Once/day Twice/day Three times/day Four times/day Other

Vitamin D frequency-other, specify _____

Vitamin D start date

Patient Name _____

Date Vitamin D end date

VPRIV dose _____

VPRIV dose units grams IU micrograms mg mg/kg ml tab units/kgVPRIV frequency Unknown Once/day Twice/day Three times/day Four times/day Other

VPRIV frequency-other, specify _____

VPRIV start date VPRIV end date

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 LSD Medication-Other end date **Other Medications**

Other medications

- | | | | |
|--|--|--|---|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> None | <input type="checkbox"/> Actonel | <input type="checkbox"/> Analgesics |
| <input type="checkbox"/> Antacids | <input type="checkbox"/> Antianxiety | <input type="checkbox"/> Antibiotics | <input type="checkbox"/> Anticonvulsants |
| <input type="checkbox"/> Antiemetics | <input type="checkbox"/> Antihistamines | <input type="checkbox"/> Antihypertensives | <input type="checkbox"/> Antiinflammatories |
| <input type="checkbox"/> Antipsychotics | <input type="checkbox"/> Antipyretics | <input type="checkbox"/> Aspirin | <input type="checkbox"/> Bronchodilators |
| <input type="checkbox"/> Calcium | <input type="checkbox"/> Contraceptives-injections | <input type="checkbox"/> Contraceptives-oral | <input type="checkbox"/> Corticosteroids |
| <input type="checkbox"/> Growth hormone | <input type="checkbox"/> Immunosuppressives | <input type="checkbox"/> Iron | <input type="checkbox"/> Laxatives |
| <input type="checkbox"/> Lipid-lowering medication | <input type="checkbox"/> Sleeping medications | <input type="checkbox"/> Vitamins | <input type="checkbox"/> Other |

Experimental Treatment

Experimental Treatment

- | | | | |
|-------------------------------|--|---|----------------------------------|
| <input type="checkbox"/> None | <input type="checkbox"/> Transplants done abroad | <input type="checkbox"/> Other cell therapies | <input type="checkbox"/> DUOC-01 |
| <input type="checkbox"/> ERT | <input type="checkbox"/> Gene therapy | <input type="checkbox"/> Combination therapy | <input type="checkbox"/> Other |

Experimental Treatment-Other, specify _____

NutritionMode 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 |

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

- | | | | |
|-------------------------------------|--|--|--|
| <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 | | |

Comments

Treatment comments

Patient Name _____

Date

ASM - Additional Testing

Auditory Testing

Auditory testing completed No YesType 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 YesSwallow study results Unknown Within normal limits Abnormalities found

Describe abnormal swallow study results

Radiology/Imaging

Radiology/Imaging tests performed

 Unknown None HRCT of the chest at all 4 levels Hand x-ray to determine bone age Chest X-ray Ultrafast CT of the coronary arteries Abdominal MRI DEXA scan Abdominal ultrasound

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

Brain MRI

Brain CT

Other

HRCT

HRCT date

Abnormalities found in the HRCT

- | | | |
|--|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> None | <input type="checkbox"/> Increased interstitial markings |
| <input type="checkbox"/> Ground glass appearance | <input type="checkbox"/> Nodules or granulomas | <input type="checkbox"/> Consolidation |
| <input type="checkbox"/> Effusion | <input type="checkbox"/> Other | |

Abnormalities found in the HRCT-other, specify

Hand X-ray

Hand x-ray date

Chronologic age of patient at time of hand x-ray (year/month) _____

Bone age of the patient (year/month) based on hand x-ray results _____

Interpretation of hand x-ray

Standard deviation of hand x-ray results

- | | |
|--|--|
| <input type="radio"/> 2 or more standard deviations above the mean | <input type="radio"/> 1 to 2 standard deviations above the mean |
| <input type="radio"/> 0 to 1 standard deviations above the mean | <input type="radio"/> 0 to 1 standard deviations below the mean |
| <input type="radio"/> 1 to 2 standard deviations below the mean | <input type="radio"/> 2 or more standard deviations below the mean |

Chest X-ray

Chest x-ray date

ILD Score _____

Abnormalities found in chest x-ray

- | | | |
|--|--|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> None | <input type="checkbox"/> Increased interstitial markings |
| <input type="checkbox"/> Ground glass appearance | <input type="checkbox"/> Nodules or granulomas | <input type="checkbox"/> Consolidation |
| <input type="checkbox"/> Effusion | <input type="checkbox"/> Other | |

Abnormalities found in chest x-ray-other, specify

Patient Name _____

Date

Ultrafast CT

Ultrafast CT date

Ultrafast CT results Unknown Within normal limits Abnormalities found

Abnormal Ultrafast CT results-specify

Total Agatston score/Volume score_____

Left Main (LMA) Agatston score/Volume score_____

Left Anterior Descending (LAD) Agatston score/Volume score_____

Left Circumflex (LCx) Agatston score/Volume score_____

Right Coronary (RCA) Agatston score/Volume score_____

Abdominal MRI

Abdominal MRI date

Abdominal MRI results Unknown Within normal limits Abnormalities found

Please describe any abnormalities found in the abdominal MRI.

Patient's weight on the day of the scan (kg)_____

Liver volume (mL)_____

Liver Multiples of Normal CALCULATED

Spleen volume (mL)_____

Spleen Multiples of Normal CALCULATED

DEXA Scan

DEXA scan date

Age at time of DEXA scan_____

AP Spine T-score_____

AP Spine Z-score_____

Indications of AP Spine T and Z-scores

- Unknown
- Osteoporosis
- Osteopenia
- Normal bone density
- Other

Patient Name _____

Date

Indications of AP Spine T and Z-scores-other, specify

Femoral Neck (left) T-score _____

Femoral Neck (left) Z-score _____

Indications of the Femoral Neck T and Z-scores

- Unknown Osteoporosis Osteopenia Normal bone density
- Other

Indications of the Femoral Neck T and Z-scores-other, specify

Total Hip (left) T-score _____

Total Hip (left) Z-score _____

Indications of the Total Hip T and Z-scores

- Unknown Osteoporosis Osteopenia Normal bone density
- Other

Indications of the Total Hip T and Z-scores-other, specify

DEXA scan Interpretation

Abdominal Ultrasound

Abdominal ultrasound date

Patient's weight on the day of the ultrasound (kg) _____

Liver span (cm) _____

Liver volume (mL) _____

Liver Multiples of Normal CALCULATED

Spleen span (cm) _____

Spleen volume (mL) _____

Patient Name _____

Date

Spleen Multiples of Normal CALCULATED

Brain MRI

Brain MRI results Unknown Normal Abnormal

Describe the abnormal brain MRI results

Brain CT

Describe the results of the brain CT scan

Other Radiology/Imaging Tests

Describe the other radiology/imaging tests conducted and findings

Comments

Additional testing comments

Patient Name _____

Date

ASM - Labs

Lipid Panel

Fasting lipid panel results

- Unknown Not done Total cholesterol Triglycerides HDL
 LDL cholesterol LDL triglycerides VLDL cholesterol VLDL triglycerides

Date of fasting lipid panel 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 _____

Patient Name _____

Date **Chemistry**

Chemistry results

- | | |
|---|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Not done |
| <input type="checkbox"/> Albumin | <input type="checkbox"/> Alkaline phosphatase |
| <input type="checkbox"/> Alanine aminotransferase (ALT) | <input type="checkbox"/> Aspartate aminotransferase (AST) |
| <input type="checkbox"/> Total bilirubin | <input type="checkbox"/> Blood urea nitrogen (BUN) |
| <input type="checkbox"/> Calcium | <input type="checkbox"/> Chloride |
| <input type="checkbox"/> Creatinine | <input type="checkbox"/> Gamma-glutamyl transpeptidase (GGT) |
| <input type="checkbox"/> Globulin | <input type="checkbox"/> Glucose |
| <input type="checkbox"/> IGF-1 | <input type="checkbox"/> IGF-1 binding protein |
| <input type="checkbox"/> Lactate dehydrogenase (LDH) | <input type="checkbox"/> Phosphorous |
| <input type="checkbox"/> Potassium | <input type="checkbox"/> Total protein |
| <input type="checkbox"/> Sodium | <input type="checkbox"/> Thyroid stimulating hormone (TSH) |
| <input type="checkbox"/> Total thyroxine (T4) | <input type="checkbox"/> Total triiodothyronine (T3) |
| <input type="checkbox"/> Uric acid | <input type="checkbox"/> D-dimer |
| <input type="checkbox"/> PT/PTT | <input type="checkbox"/> Vitamin D (25 OH) |

Chemistry collection date Albumin Within normal limits Abnormal In progress Results unavailable

Albumin value _____

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

Albumin reference range _____

Alkaline phosphatase Within normal limits Abnormal In progress Results unavailable

Alkaline phosphatase value _____

Alkaline phosphatase units U/L ukat/L

Alkaline phosphatase reference range _____

Alanine aminotransferase Within normal limits Abnormal In progress Results unavailable

Alanine aminotransferase value _____

Alanine aminotransferase units U/L ukat/L

Alanine aminotransferase reference range _____

Aspartate aminotransferase Within normal limits Abnormal In progress Results unavailable

Aspartate aminotransferase value _____

Aspartate aminotransferase units U/L ukat/L

Aspartate aminotransferase reference range _____

Total bilirubin Within normal limits Abnormal In progress Results unavailable

Total bilirubin value _____

Patient Name _____

Date Total bilirubin units umol/L mg/dL

Total bilirubin reference range _____

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 _____

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 _____

Creatinine Within normal limits Abnormal In progress Results unavailable

Creatinine value _____

Creatinine units _____

Creatinine 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 _____

Globulin reference range _____

Glucose Within normal limits Abnormal In progress Results unavailable

Glucose value _____

Glucose units mmol/L mg/dL

Glucose reference range _____

IgF-1 Within normal limits Abnormal In progress Results unavailable

IgF-1 value _____

IgF-1 units ng/mL ug/L

IgF-1 reference range _____

IGF-1 binding protein Within normal limits Abnormal In progress Results unavailable

IGF-1 binding protein value _____

Patient Name _____

Date

IGF-1 binding protein units _____

IGF-1 binding protein reference range _____

Lactate dehydrogenase Within normal limits Abnormal In progress Results unavailable

Lactate dehydrogenase value _____

Lactate dehydrogenase units U/L

Lactate dehydrogenase reference range _____

Phosphorous Within normal limits Abnormal In progress Results unavailable

Phosphorous value _____

Phosphorous units mmol/L mg/dL

Phosphorous reference range _____

Potassium Within normal limits Abnormal In progress Results unavailable

Potassium value _____

Potassium units mEq/L mmol/L

Potassium 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 _____

Sodium Within normal limits Abnormal In progress Results unavailable

Sodium value _____

Sodium units mEq/L mmol/L

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

Thyroxine (total) value _____

Thyroxine (total) units pmol/L ng/mL

Thyroxine (total) reference range _____

Total thyroxine (T4) Within normal limits Abnormal In progress Results unavailable

Total thyroxine (T4) value _____

Total thyroxine (T4) units _____

Total thyroxine (T4) reference range _____

Total Iron Binding Capacity (TIBC) Within normal limits Abnormal In progress Results unavailable

Total Iron Binding Capacity (TIBC) value _____

Patient Name _____

Date Total Iron Binding Capacity (TIBC) units umol/L mg/dL

Total Iron Binding Capacity (TIBC) reference range _____

D-dimer Within normal limits Abnormal In progress Results unavailable

D-dimer value _____

D-dimer units _____

D-dimer reference range _____

PT/PTT Within normal limits Abnormal In progress Results unavailable

PT/PTT value _____

PT/PTT reference range _____

Vitamin D (25 OH) Within normal limits Abnormal In progress Results unavailable

Vitamin D (25 OH) value _____

Vitamin D (25 OH) units _____

Vitamin D (25 OH) reference range _____

Hematology

Complete blood count (CBC) results

- | | | |
|---|---------------------------------------|--|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Not done | <input type="checkbox"/> White blood cell count (WBCC) |
| <input type="checkbox"/> Hemoglobin | <input type="checkbox"/> Hematocrit | <input type="checkbox"/> Red blood cell count (RBCC) |
| <input type="checkbox"/> Platelet count | <input type="checkbox"/> Neutrophil # | <input type="checkbox"/> Lymphocyte # |
| <input type="checkbox"/> Monocyte # | <input type="checkbox"/> Eosinophil # | <input type="checkbox"/> Basophil # |

CBC collection date 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 $10^6/uL$ $10^{12}/uL$

Patient Name _____

Date

Red blood cell count reference range _____

Platelet count Within normal limits Abnormal In progress Results unavailable

Platelet count value _____

Platelet count units THOU/uL 10⁹/L 10³/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 _____

UrinalysisUrinalysis performed Unknown Yes NoUrine collection date Urine color Unknown Dark yellow Pale yellow Nearly colorless Brown PinkUrine clarity Clear Slightly cloudy Cloudy TurbidBlood in the urine Unknown Yes NoUrine RBCC Within normal limits Abnormal In progress Results unavailable

Urine RBCC value _____

Urine RBCC units _____

Urine RBCC reference range _____

Patient Name _____

Date

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
- Ketones Leukocyte esterase Nitrite Bilirubin Urobilinogen
- 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 _____

Patient Name _____

Date 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 _____

Urobilinogen Within normal limits Abnormal In progress Results unavailable

Urobilinogen value _____

Urobilinogen units _____

Urobilinogen 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

Niemann-Pick biomarkers

- Unknown Not done ASM enzyme activity Cathepsin B
 CCL18 Chitotriosidase IL-1beta MIP-1 alpha
 Sphingomyelin TNF-alpha

ASM enzyme activity Within normal limits Abnormal In progress Results unavailable

ASM enzyme activity value _____

ASM enzyme activity units _____

ASM enzyme activity reference range _____

Cathepsin B Within normal limits Abnormal In progress Results unavailable

Cathepsin B value _____

Patient Name _____

Date

Cathepsin B units _____

Cathepsin B reference range _____

CCL18 Within normal limits Abnormal In progress Results unavailable

CCL18 value _____

CCL18 units _____

CCL18 reference range _____

Chitotriosidase Within normal limits Abnormal In progress Results unavailable

Chitotriosidase value _____

Chitotriosidase units _____

Chitotriosidase reference range _____

IL-1beta Within normal limits Abnormal In progress Results unavailable

IL-1beta value _____

IL-1beta units _____

IL-1beta reference range _____

MIP-1 alpha Within normal limits Abnormal In progress Results unavailable

MIP-1 alpha value _____

MIP-1 alpha units _____

MIP-1 alpha reference range _____

Sphingomyelin Within normal limits Abnormal In progress Results unavailable

Sphingomyelin value _____

Sphingomyelin units _____

Sphingomyelin reference range _____

TNF-alpha Within normal limits Abnormal In progress Results unavailable

TNF-alpha value _____

TNF-alpha units _____

TNF-alpha reference range _____

Other LabsOther labs collected Unknown Yes NoNumber 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 _____

Patient Name _____						
Date <table border="1"><tr><td>M</td><td>M</td></tr><tr><td>D</td><td>D</td></tr><tr><td>Y</td><td>Y</td></tr></table>	M	M	D	D	Y	Y
M	M					
D	D					
Y	Y					

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

--

Patient Name _____

Date

ASM - Sick Visits And Hospitalizations

Sick Visits

Sick visits since last outpatient visit Unknown Yes No

Number of sick visits _____

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

Location for sick visit 1

 Unknown Emergency department Retail clinic Primary care Specialty center Urgent care Direct hospital admission OtherPatient 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 Reason for sick visit 2 Unknown Condition related Condition unrelatedSick visit 2 was a condition exacerbation Unknown Yes No

Location for sick visit 2

 Unknown Emergency department Retail clinic Primary care Specialty center Urgent care Direct hospital admission OtherPatient was admitted to the hospital as a result of sick visit 2 Unknown Yes No

Name of hospital for sick visit 2 _____

ICD-9 codes for sick visit 2 known Yes No

ICD-9 codes for sick visit 2 _____

Diagnosis for sick visit 2 _____

Number of inpatient days for sick visit 2 _____

Number of ICU days for sick visit 2 _____

Date of sick visit 3 Reason for sick visit 3 Unknown Condition related Condition unrelatedSick visit 3 was a condition exacerbation Unknown Yes No

Patient Name _____

Date | |

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 | |

Date of discharge for first hospitalization | |

Reason for first hospitalization

Location of second hospitalization _____

Date of admission for second hospitalization | |

Date of discharge for second hospitalization | |

Reason for second hospitalization

Location of third hospitalization _____

Date of admission for third hospitalization | |

Date of discharge for third hospitalization | |

Reason for third hospitalization

Patient Name _____

Date

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

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

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

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

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

Reason for third surgery-Other, specify _____

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

--