

Patient Name _____

Date

MPS - 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 MPS I disease

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 _____

Patient is participating in a Genzyme disease registry Unknown Yes No

Education

Maternal education

Patient Name _____

Date

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

Paternal education

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

Ancestral Origin, Race and EthnicityAncestral Origin Unknown Africa Asia Europe North America South America Oceania Other

Ancestral Origin-Africa

- Unknown Egypt Eritrea Ethiopia Liberia Somalia South Africa
 Other

Ancestral Origin-Africa-Other, specify _____

Ancestral Origin-Asia

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

Ancestral Origin-Asia-Other, specify _____

Ancestral Origin-Europe

- Unknown Austria Belgium Bulgaria Croatia
 Czech Republic Denmark Finland France Germany

Patient Name _____

Date

- Greece Hungary Iceland Ireland Italy
 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

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"/>

- 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

Comments

Intake demographics comments

Patient Name _____

Date

MPS - 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

IDUA enzyme activity on first newborn screen _____

IDUA enzyme activity units on first newborn screen

 nmol/hr/mg protein nmol/min/mg protein nmol/hr/mL nmol/min/mL micromol/L/hr pmol/punch/hr kat/kg protein microkat/kg protein micromol/g/hr

IDUA enzyme sample type on first newborn screen

 Dried blood spot Fibroblast Leukocyte Plasma OtherDate 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 _____

IDUA enzyme activity on second newborn screen _____

IDUA enzyme activity units on second newborn screen

 nmol/hr/mg protein nmol/min/mg protein nmol/hr/mL nmol/min/mL micromol/L/hr pmol/punch/hr kat/kg protein microkat/kg protein micromol/g/hr

IDUA enzyme sample type on second newborn screen

 Dried blood spot Fibroblast Leukocyte Plasma OtherDate 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 _____

IDUA enzyme activity on third newborn screen _____

Patient Name _____

Date

IDUA enzyme activity units on third newborn screen

- nmol/hr/mg protein nmol/min/mg protein nmol/hr/mL nmol/min/mL
 micromol/L/hr pmol/punch/hr kat/kg protein microkat/kg protein
 micromol/g/hr

IDUA enzyme sample type on third newborn screen

- Dried blood spot Fibroblast Leukocyte Plasma Other

Enzymatic Confirmatory Testing

Date of enzymatic confirmatory testing

Confirmatory testing IDUA result _____

Confirmatory testing IDUA normal reference range _____

Lab assay used for IDUA enzymatic confirmation

- Dried blood spot Fibroblast Leukocyte Plasma Other

Lab assay used for IDUA enzymatic confirmation-other, specify _____

Method for IDUA enzymatic confirmation Unknown MS/MS Fluorometric

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 _____

Newborn Hearing Screen

Newborn hearing screen performed Unknown Yes NoR Ear: Equipment Type Unknown DPOAE TEOAE ABR AABR ALGO OtherRight ear: Screening test results Pass Refer Not TestedL Ear: Equipment Type Unknown DPOAE TEOAE ABR AABR ALGO OtherLeft ear: Screening test results Pass Refer Not Tested

Recommendation

- Repeat hearing screen Referral for diagnostic testing Risk factor monitoring only
 Refused further action No further action required

Date of Audiological Diagnostic Evaluation Right ear: Diagnosis: Hearing loss? Yes No

Right ear: Diagnosis: Degree of hearing loss

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

Right ear: Diagnosis: Type of hearing loss

- Sensorineural Conductive
 Mixed Auditory neuropathy/Auditory dys-synchrony

Patient Name _____

Date 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

- | | | |
|---|---|---|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> None | <input type="checkbox"/> Alopecia |
| <input type="checkbox"/> Apnea | <input type="checkbox"/> Arrhythmia | <input type="checkbox"/> Ataxia |
| <input type="checkbox"/> Athetosis | <input type="checkbox"/> Autistic-like features | <input type="checkbox"/> Body odor |
| <input type="checkbox"/> Brain abnormalities | <input type="checkbox"/> Brain malformations | <input type="checkbox"/> Candidiasis |
| <input type="checkbox"/> Cardiomyopathy | <input type="checkbox"/> Cataract(s) | <input type="checkbox"/> Cerebral edema |
| <input type="checkbox"/> Chorea | <input type="checkbox"/> Cirrhosis | <input type="checkbox"/> Clonus |
| <input type="checkbox"/> Cognitive impairment | <input type="checkbox"/> Coma | <input type="checkbox"/> Confusion |
| <input type="checkbox"/> Conjunctivitis | <input type="checkbox"/> Contracture(s)-musculoskeletal | <input type="checkbox"/> Corneal erosion |
| <input type="checkbox"/> Dehydration | <input type="checkbox"/> Dermatitis | <input type="checkbox"/> Developmental delay(s) |
| <input type="checkbox"/> Disorientation | <input type="checkbox"/> Drooling/hypersalivation | <input type="checkbox"/> Dysarthria |
| <input type="checkbox"/> Dysmetria | <input type="checkbox"/> Dismorphism | <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 |

Patient Name _____

Date

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

Mutation 1

- A327P L218P L490P P533R Q380R Q70X R628X S633L T388R
 W402X Other

Mutation 1- rare, specify _____

Mutation 2

- A327P L218P L490P P533R Q380R Q70X R628X S633L T388R
 W402X Other

Mutation 2-rare, specify _____

Variants of unknown significance _____

Most appropriate MPS I diagnosis Severe Attenuated UndeterminedMutation is de novo Unknown Yes NoPseudodeficiency alleles for MPS I A79T T99I D223N V322E G409R H82QMaternal genetic testing done Unknown Yes No Genotyping in progressPaternal genetic testing done Unknown Yes No Genotyping in progressPhase of mutation(s) Unknown Maternal Paternal**Comments**

Enzyme mutation analysis comments

Patient Name _____

Date

MPS - 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)
- Intrauterine insemination (IUI)
- Donor sperm
- Donor embryo
- Preimplantation genetic diagnosis (PGD)
- Surrogate
- Donor egg
- Other

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

Medical issues related to prematurity

- None
- Respiratory distress
- Sepsis
- Intracranial hemorrhage
- Retinopathy of prematurity
- Blood transfusions
- Necrotizing enterocolitis
- Total parenteral nutrition (TPN)
- 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 MPS I Unknown Yes No

Relationship of family members with confirmed MPS I to proband

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

Relationship of family members with confirmed MPS I to proband-Other, specify _____

Family member(s) with symptoms suggestive of MPS I Unknown Yes No

Relationship of family members with suggested MPS I to proband

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

Relationship of family members with suggested MPS I 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 JewishMother is Hispanic or Latino Not reported Yes No

Paternal race

Patient Name_____
Date <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>

- Not reported
- Asian
- Native Hawaiian or Other Pacific Islander
- Paternal race-White Ashkenazi Jewish Non-Ashkenazi Jewish
- American Indian/Alaskan Native
- Black or African American
- White

Father is Hispanic or Latino Not reported Yes No

Patient History

HEENT history

- None
- Enlarged tonsils
- Respiratory infections
- Coarse facial features
- Sleep disturbances/snoring
- Pneumonia
- Macroglossia
- Ear infections
- Reactive airway disease/asthma

Musculoskeletal history None Stiffness/joint contracture Kyphosis/gibbus

Ophthalmological history None Corneal clouding

Cardiovascular history None Cardiac valve abnormalities Murmur

Specify abnormal cardiac valve(s) Aortic Tricuspid Mitral Pulmonary

Genitourinary history None Hepatomegaly Splenomegaly Umbilical hernia Inguinal hernia

Hospitalizations prior to intake Unknown Yes No

Number of hospitalizations prior to intake related to this condition_____

Number of hospitalizations prior to intake not related to this condition_____

Genetic counseling provided Unknown Yes No

Provider of genetic counseling

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

Provider of genetic counseling, other- specify_____

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

Patient Name _____

Date ***MPS - 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 _____

Patient is participating in a Genzyme disease registry since the last visit Unknown Yes No**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

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

Patient Name _____

Date

- | | |
|--|--|
| <input type="checkbox"/> Orthopedics | <input type="checkbox"/> Otolaryngology |
| <input type="checkbox"/> Physical medicine and rehabilitation (PM&R) | <input type="checkbox"/> Physical therapy |
| <input type="checkbox"/> Primary care provider | <input type="checkbox"/> Psychiatry |
| <input type="checkbox"/> Psychology | <input type="checkbox"/> Public health nursing |
| <input type="checkbox"/> Pulmonology | <input type="checkbox"/> Respiratory therapy |
| <input type="checkbox"/> Speech-Language therapy | <input type="checkbox"/> Surgery |
| <input type="checkbox"/> Transplant | <input type="checkbox"/> Other |

Specify other current health services-other, specify _____

Preventive care status

- | | |
|--|--|
| <input type="radio"/> Unknown | <input type="radio"/> None |
| <input type="radio"/> On schedule for preventative care services | <input type="radio"/> 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

Patient Name _____

Date

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

Reason special education services received-other, specify _____

Special education category

- Unknown Autism spectrum disorders
 Blind-visually impaired Deaf and hard of hearing
 Deaf-Blind Developmental cognitive disabilities: mild-moderate
 Developmental cognitive disabilities: severe- profound Developmental delay
 Emotional/Behavioral disorders Physically impaired
 Severely multiply impaired Specific learning disabilities
 Speech or language impairments Traumatic brain injury
 Other health disabilities

Special education, other- specify _____

ProceduresAnesthesia since last visit Unknown Yes NoType 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

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"/>

- 7 8 9 10 11 12 13 14
 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

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

Patient Name _____

Date

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

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 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

Patient Name _____

Date

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

 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 NoTransplant organ Unknown Bone marrow Stem Cell Liver Kidney Heart Other

Transplant organ-other, specify_____

Date of organ transplant

Reason for transplant

 Unknown Liver failure Condition-related treatment

 Condition-related organ failure Other

Reason for transplant-other, specify_____

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

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

MPS - 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 inHow 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 AbnormalConstitutional 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

Musculoskeletal Exam

Musculoskeletal exam findings Unknown Not done Normal Abnormal

Abnormal musculoskeletal exam findings

- | | | | |
|--|---|--|------------------------------------|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Stiffness/joint contacture | <input type="checkbox"/> Kyphosis/gibbus | <input type="checkbox"/> Scoliosis |
| <input type="checkbox"/> Genu valgum | <input type="checkbox"/> Pes cavus | <input type="checkbox"/> Toe-walking | <input type="checkbox"/> Lordosis |
| <input type="checkbox"/> Claw hand deformity | <input type="checkbox"/> Other | | |

Musculoskeletal exam findings-Other, specify _____

Describe all abnormal musculoskeletal exam findings

HEENT Exam

HEENT Exam Findings Unknown Not done Normal Abnormal

Abnormal HEENT exam findings

- | | | |
|---|---|---|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Coarse facial features | <input type="checkbox"/> Macroglossia |
| <input type="checkbox"/> Enlarged tonsils | <input type="checkbox"/> Sleep disturbances/snoring | <input type="checkbox"/> Ear infections |
| <input type="checkbox"/> Respiratory infections | <input type="checkbox"/> Pneumonia | <input type="checkbox"/> Reactive airway disease/asthma |
| <input type="checkbox"/> Other | | |

Abnormal HEENT exam findings-other, specify

Describe all abnormal HEENT exam findings

Patient Name _____

Date

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

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 Cardiac valve abnormalities Murmur

Rhythm abnormalities Other

Specify abnormal cardiac valve(s) Aortic Tricuspid Mitral Pulmonary

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

Patient Name _____

Date

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 Cognitive impairment Clinically significant myelopathy
- Carpal tunnel syndrome Other

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

Patient Name _____

Date

Describe all abnormal genitourinary exam findings

Comments

Physical exam comments

Patient Name _____

Date

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

- 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 No

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

Date of Pulmonary function testing

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 _____

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" 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"/>

Authors used for predicted values

PFT Comments

Comments

Consultations comments

Patient Name _____

Date

MPS - Treatment

Enzyme replacement therapy Not done DoneType of ERT Unknown Laronidase (Aldurazyme) Investigational drugHematopoietic stem cell transplant Not done DoneHematopoietic stem cell transplant-Donor type Cord blood Bone marrow PBSCHematopoietic stem cell transplant-Regimen Ablative Reduced intensityHematopoietic stem cell transplant-Chimerism Full donor Mixed donorHematopoietic stem cell transplant-Enzyme levels Normal Low

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 _____

Patient Name _____

Date

Anti-coagulant start date

Anti-coagulant end date

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

Anti-depressant end date

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

Anti-diarrheal end date

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

Anti-migraine end date

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

Anti-platelet end date

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

ARB end date

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 _____

Patient Name _____

Date 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 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/kgCerezyme 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/kgDigitalis frequency Unknown Once/day Twice/day Three times/day Four times/day Other

Digitalis frequency-other, specify _____

Digitalis start date Digitalis end date

Patient Name _____

Date

Diuretic dose _____

Diuretic dose units grams IU micrograms mg mg/kg ml tab units/kgDiuretic 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 _____

Fabrazyme dose units grams IU micrograms mg mg/kg ml tab units/kgFabrazyme 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/kgFolic 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/kgFosamax 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

Patient Name _____

Date 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/kgMethotrexate frequency Unknown Once/day Twice/day Three times/day Four times/day Other

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 _____

Patient Name _____

Date 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 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

-
- None
-
- Transplants done abroad
-
- Other cell therapies
-
- DUOC-01

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

- 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

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

Comments

Treatment comments

Patient Name _____

Date

MPS - 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

- | | | | |
|---|-------------------------------|---|------------------------------------|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> None | <input type="checkbox"/> Abdominal ultrasound | <input type="checkbox"/> Brain MRI |
| <input type="checkbox"/> Brain Ultrasound | <input type="checkbox"/> NCV | <input type="checkbox"/> Skeletal Xray | <input type="checkbox"/> Spine MRI |
| <input type="checkbox"/> Other | | | |

Patient Name _____

Date **NCV**

NCV date _____

NCV results Unknown Within normal limits Abnormalities found

Please describe any abnormalities found in the NCV

Abdominal UltrasoundAbdominal 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) _____

Spleen Multiples of Normal CALCULATED**Skeletal X-ray**Skeletal x-ray date Skeletal x-ray results Unknown Within normal limits Abnormalities found**Spine MRI**Spine MRI results Unknown Within normal limits Abnormalities found

Describe abnormal spine MRI results

 Marrow infiltration
 Lytic lesions
 Avascular necrosis
 Infarction
 Other

Describe abnormal spine MRI results-other, specify

Brain MRIBrain MRI results Unknown Normal Abnormal

Describe the abnormal brain MRI results

Patient Name _____

Date

Evidence of hydrocephaly Yes No

Brain Ultrasound

Brain Ultrasound results Unknown Within normal limits Abnormalities found

Evidence of hydrocephaly Yes No

Other Radiology/Imaging Tests

Describe the other radiology/imaging tests conducted and findings

Comments

Additional testing comments

Patient Name _____

Date

MPS - 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 **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 $10^6/uL$ $10^{12}/uL$

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 _____

Patient Name _____

Date 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 _____

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

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

- 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 _____

Patient Name _____

Date

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 _____

BiomarkersMPS I Biomarkers Unknown Not done Antibody levels (if on ERT) IDUA Enzyme assay Urine GAGAntibody level Within normal limits Abnormal In progress Results unavailable

Antibody level value _____

Antibody level units _____

Antibody level reference range _____

IDUA enzyme activity Within normal limits Abnormal In progress Results unavailable

IDUA enzyme activity value _____

IDUA enzyme activity units _____

IDUA enzyme activity reference range _____

Urine GAG Within normal limits Abnormal In progress Results unavailable

Urine GAG value _____

Urine GAG units _____

Urine GAG 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 _____

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

Other lab 2 value _____

Patient Name _____

Date

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

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

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

[Empty box 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" 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"/>

Reason for third surgery-Other, specify _____

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

--