

Patient Name \_\_\_\_\_

Date      

## GAA - Demographics

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NBSTRN ID \_\_\_\_\_

Intake date      

### Consent

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

### Demographics Information

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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

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Patient condition category  SACHDNC candidate disordersSpecify SACHDNC candidate disorder diagnosis for the patient  Pompe 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

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Maternal education

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

- 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 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

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- |                                    |                                      |                                      |   |                                 |
|------------------------------------|--------------------------------------|--------------------------------------|---|---------------------------------|
| <input type="checkbox"/> Greece    | <input type="checkbox"/> Hungary     | <input type="checkbox"/> Iceland     | <input type="checkbox"/> Ireland        | <input type="checkbox"/> Italy  |
| <input type="checkbox"/> Lithuania | <input type="checkbox"/> Malta       | <input type="checkbox"/> Netherlands | <input type="checkbox"/> Norway         | <input type="checkbox"/> Poland |
| <input type="checkbox"/> Romania   | <input type="checkbox"/> Serbia      | <input type="checkbox"/> Slovakia    | <input type="checkbox"/> Slovenia       | <input type="checkbox"/> Spain  |
| <input type="checkbox"/> Sweden    | <input type="checkbox"/> Switzerland | <input type="checkbox"/> Ukraine     | <input type="checkbox"/> United Kingdom | <input type="checkbox"/> Other  |

Ancestral Origin-Europe-Italy  Unknown  Sicily

Ancestral Origin-Europe-Romania  Unknown  Transylvania

Ancestral Origin-Europe-United Kingdom  Unknown  England  Northern Ireland  Scotland  Wales

Ancestral Origin-Europe-Other, specify \_\_\_\_\_

Ancestral Origin-North America

- |                                  |   |  |   |                                   |
|----------------------------------|---|--|---|-----------------------------------|
| <input type="checkbox"/> Unknown | <input type="checkbox"/> Aleutian Islands | <input type="checkbox"/> Canada        | <input type="checkbox"/> Dominican Republic | <input type="checkbox"/> Honduras |
| <input type="checkbox"/> Mexico  | <input type="checkbox"/> Puerto Rico      | <input type="checkbox"/> United States | <input type="checkbox"/> Other              |                                   |

Ancestral Origin-North America-Canada  Unknown  French Canadian

Ancestral Origin-North America-Other, specify \_\_\_\_\_

Ancestral Origin-South America  Unknown  Colombia  Venezuela  Other

Ancestral Origin-South America-Other, specify \_\_\_\_\_

Ancestral Origin-Oceania  Unknown  Australia  Other

Ancestral Origin-Oceania-Other, specify \_\_\_\_\_

Ancestral Origin-Other  Unknown  Amish  Arabic  Hutterite  Mennonite  Jewish  Other

Ancestral Origin-Other-Jewish  Unknown  Ashkenazic  Sephardic

Ancestral Origin-Other, specify \_\_\_\_\_

Race

- |  |   |
|--|---|
| <input type="checkbox"/> Not reported                              | <input type="checkbox"/> American Indian/Alaskan Native |
| <input type="checkbox"/> Asian                                     | <input type="checkbox"/> Black or African American      |
| <input type="checkbox"/> Native Hawaiian or Other Pacific Islander | <input type="checkbox"/> White                          |

Race-White, specify  Ashkenazi Jewish  Non-Ashkenazi Jewish

Patient is Hispanic or Latino  Not reported  Yes  No

**Socioeconomics**

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Maternal age (in years) at patient's birth \_\_\_\_\_

Mother's marital status at patient's birth

- |                                     |   |                               |                                |                                 |
|-------------------------------------|---|-------------------------------|--------------------------------|---------------------------------|
| <input type="radio"/> Unknown       | <input type="radio"/> Married             | <input type="radio"/> Widowed | <input type="radio"/> Divorced | <input type="radio"/> Separated |
| <input type="radio"/> Never married | <input type="radio"/> Living with partner |                               |                                |                                 |

**Medical Coverage**

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Maternal medical coverage at time of delivery

- Unknown
- None

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- 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

Family constellation

## Comments

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Intake demographics comments

Patient Name \_\_\_\_\_

Date      

## GAA - 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

State NBS serial number on first newborn screen \_\_\_\_\_

GAA activity result on first newborn screen (umol/L/hr) \_\_\_\_\_

GAA activity result normal reference range on first newborn screen \_\_\_\_\_

GAA activity tissue type on first newborn screen

 Dried blood spot  Lymphocyte  Leukocyte  Skin  Muscle 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 \_\_\_\_\_

State NBS serial number on second newborn screen \_\_\_\_\_

GAA activity result on second newborn screen (umol/L/hr) \_\_\_\_\_

GAA activity result normal reference range on second newborn screen \_\_\_\_\_

GAA activity tissue type on second newborn screen

 Dried blood spot  Lymphocyte  Leukocyte  Skin  Muscle 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 \_\_\_\_\_

State NBS serial number on third newborn screen \_\_\_\_\_

GAA activity result on third newborn screen (umol/L/hr) \_\_\_\_\_

GAA activity result normal reference range on third newborn screen \_\_\_\_\_

Patient Name \_\_\_\_\_

Date      

GAA activity tissue type on third newborn screen

- Dried blood spot  Lymphocyte  Leukocyte  Skin  Muscle  
 Other

## Enzymatic Confirmatory Testing

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Date of enzymatic confirmatory testing      CRIM status  Unknown  Not done  Positive  NegativeCRIM status determination  Blood  Fibroblasts  GenotypeCRIM status from blood  Unknown  Not done  Positive  Negative

Confirmatory testing GAA enzyme activity result \_\_\_\_\_

GAA enzyme activity level units  Unknown  nmol/hr/mg protein  nmol/min/mg protein  Other

GAA enzyme activity level units-other, specify \_\_\_\_\_

GAA enzyme activity normal reference range \_\_\_\_\_

GAA enzyme activity substrate  Unknown  Maltose  Other

GAA enzyme activity substrate-other, specify \_\_\_\_\_

GAA enzyme assay method

- Unknown  Fibroblast  Muscle  Leukocyte  Lymphocyte  Dry blood spot  
 Other

GAA enzyme assay method-other, specify \_\_\_\_\_

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

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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



Patient Name \_\_\_\_\_

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- |  |   |  |
|--|---|--|
| <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 \_\_\_\_\_

Feeding status at time of initial contact    Breast milk    Formula    Parenteral**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 \_\_\_\_\_

Mutation 2 \_\_\_\_\_

Polymorphisms \_\_\_\_\_

Lab where diagnostic testing performed \_\_\_\_\_

Genotype data concordant with Pompe newborn screening results    Yes    NoMutation is de novo    Unknown    Yes    No

Pseudodeficiency alleles \_\_\_\_\_

Maternal 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



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# GAA - 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 \_\_\_\_\_

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

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

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

Medical issues related to prematurity

- |   |   |   |
|---|---|---|
| <input type="checkbox"/> None                 | <input type="checkbox"/> Intracranial hemorrhage    | <input type="checkbox"/> Necrotizing enterocolitis        |
| <input type="checkbox"/> Respiratory distress | <input type="checkbox"/> Retinopathy of prematurity | <input type="checkbox"/> Total parenteral nutrition (TPN) |
| <input type="checkbox"/> Sepsis               | <input type="checkbox"/> Blood transfusions         | <input type="checkbox"/> 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

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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

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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 History**Consanguinity    Unknown    Yes    NoType of consanguinity    First cousins or closer    OtherPedigree obtained    Yes    NoFamily members with confirmed Pompe    Unknown    Yes    No

Relationship of family members with confirmed Pompe to proband

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

Relationship of family members with confirmed Pompe to proband-Other, specify \_\_\_\_\_

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

Relationship of family members with suggested Pompe to proband

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

Relationship of family members with suggested Pompe 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

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

Paternal race-White     Ashkenazi Jewish     Non-Ashkenazi Jewish

Father is Hispanic or Latino     Not reported     Yes     No

Hospitalizations prior to intake     Unknown     Yes     No

Number of hospitalizations prior to intake related to this condition\_\_\_\_\_

Number of hospitalizations prior to intake not related to this condition\_\_\_\_\_

Genetic counseling provided     Unknown     Yes     No

Provider of genetic counseling

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

Provider of 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      ***GAA - 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            |

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

Family constellation at visit

**Emergency Management**

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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 \_\_\_\_\_

Patient Name \_\_\_\_\_

Date      **Education**Special education assessment recommended  Unknown  Yes  No  Special education services already received

Reason special education services received

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

Reason special education services received-other, specify \_\_\_\_\_

Special education category

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

Special education, other- specify \_\_\_\_\_

**Procedures**Anesthesia 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

Patient Name \_\_\_\_\_

Date      **Hematologic Changes**

Hematologic changes since last visit

- Unknown                       None                       Patient bruises easily                       Patient bleeds easily  
 Nosebleeds                       Bleeding from gums                       Heavy menstrual bleeding                       Transfusions required

Approximate age of onset for bruising easily

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



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- Patient uses gastrostomy tube     GERD     Liver disease  
 Frequent diarrhea

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

Approximate age of onset for frequent abdominal pain

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

Approximate age at which gastrostomy tube was inserted

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

Approximate age of onset for GERD

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

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

Approximate age of onset for liver disease

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

### Cardiovascular Changes

---

Cardiovascular changes since last visit     Unknown     None     Congestive heart failure

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     Shortness of breath with exercise     Orthopnea  
 Respiratory distress

Describe patient's shortness of breath

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"/>

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 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 is on medication for depression     Unknown     Yes     No

Patient required hospitalization for depression     Unknown     Yes     No

Patient is suicidal     Unknown     Yes     No

Approximate age of onset for depression

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

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

Approximate age of onset for anxiety

- 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  No

Standardized developmental screening tool(s) used  Unknown  Yes  No

Developmental status  Typical  Atypical

Severity of atypical development  Unknown  Mild delay  Moderate delay  Severe delay

Developmental milestones that were achieved in a typical order and timeframe

Unknown  None  Cognitive  Fine motor  Gross motor

Social-emotional  Speech-language  Other

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

\_\_\_\_\_

Referred for further developmental assessment

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

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

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

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

\_\_\_\_\_

Progressive loss of developmental milestones since last visit  Unknown  Yes  No

### Transplants Since Last Visit

---

Organ transplant since last visit  Unknown  Yes  No

Transplant organ  Unknown  Bone marrow  Stem Cell  Liver  Kidney  Heart  Other

Transplant organ-other, specify \_\_\_\_\_

Date of organ transplant

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

# GAA - 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             Macroglossia             Ptosis                     Strabismus             Facial myleopathy  
 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 <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>
--

### 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                       Murmur                       Gallop                       Rhythm abnormalities  
 Displaced PMI                       Other

Describe murmur

Describe gallop

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    Other

How many cm below the costal margin is the liver?\_\_\_\_\_

Abnormal abdomen exam findings-other, specify

Patient Name \_\_\_\_\_

Date

Describe all abnormal abdomen exam findings

### Extremity Exam

---

Extremity exam findings    Unknown    Not done    Normal    Abnormal

Abnormal extremity exam findings    Unknown    Thin extremities    Edema    Other

Edema location \_\_\_\_\_

Abnormal extremity exam findings-other,specify

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    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



Patient Name \_\_\_\_\_

Date

## Genitourinary Exam

---

Genitourinary Exam Findings    Unknown    Not done    Normal    Abnormal

Describe all abnormal genitourinary exam findings

## Comments

---

Physical exam comments

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

## GAA - 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  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 <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"/>

- 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      

## GAA - Treatment

---

Pharmacotherapy specific to Pompe

 Unknown       None       Chaperone enzymes     IV enzymes       Substrate reduction

Chaperone enzymes dose \_\_\_\_\_

Chaperone enzymes dose units     grams     IU     micrograms     mg     mg/kg     ml     tab     units/kgChaperone enzymes start date      Chaperone enzymes end date      

IV enzymes dose \_\_\_\_\_

IV enzymes dose units     grams     IU     micrograms     mg     mg/kg     ml     tab     units/kgIV enzymes start date      IV enzymes end date      

## 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 \_\_\_\_\_

Patient Name \_\_\_\_\_

Date      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      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 \_\_\_\_\_

Patient Name \_\_\_\_\_

Date      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      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

Patient Name \_\_\_\_\_

Date        

Digitalis frequency-other, specify \_\_\_\_\_

Digitalis start date      Digitalis end 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 \_\_\_\_\_

Patient Name \_\_\_\_\_

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

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

Lyrica (pregabalin) frequency

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

Lyrica (pregabalin) frequency-other, specify \_\_\_\_\_

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

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

Methotrexate dose \_\_\_\_\_

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

Methotrexate frequency-other, specify \_\_\_\_\_

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

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

Myozyme/Lumizyme dose \_\_\_\_\_

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

Myozyme/Lumizyme frequency

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

Myozyme/Lumizyme frequency-other, specify \_\_\_\_\_

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

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

Neurontin (gabapentin) dose \_\_\_\_\_

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

Neurontin (gabapentin) frequency

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

Neurontin (gabapentin) frequency-other, specify \_\_\_\_\_

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

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

Rituximab dose \_\_\_\_\_

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

Rituximab frequency-other, specify \_\_\_\_\_

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

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

Statins dose \_\_\_\_\_

Patient Name \_\_\_\_\_

Date      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      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              |

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

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Experimental Treatment

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

Experimental Treatment-Other, specify \_\_\_\_\_

### Nutrition

---

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

Types of milk/formula taken

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

### Comments

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Treatment 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"/>

# GAA - Additional Testing

## Auditory Testing

Auditory testing completed  No  Yes

Type of auditory testing completed  ABR  BAER  Other

Type of auditory testing completed-other, specify \_\_\_\_\_

ABR results  Unknown  Within normal limits  Abnormalities found

Describe abnormal ABR results

BAER results  Unknown  Within normal limits  Abnormalities found

Describe abnormal BAER results

Other auditory testing results  Unknown  Within normal limits  Abnormalities found

Describe abnormal other auditory testing results

## Swallow Study

Swallow study completed  No  Yes

Swallow study results  Unknown  Within normal limits  Abnormalities found

Describe abnormal swallow study results

## Radiology/Imaging

Radiology/Imaging tests performed  Unknown  None  Chest x-ray  Other

### Chest X-ray

Chest x-ray date

ILD Score \_\_\_\_\_

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"/>

Abnormalities found in chest x-ray

- |  |  |   |
|--|--|---|
| <input type="checkbox"/> Unknown                 | <input type="checkbox"/> None                  | <input type="checkbox"/> Increased intersitial 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

**Other Radiology/Imaging Tests**

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Describe the other radiology/imaging tests conducted and findings

**Comments**

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Additional testing comments

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

# GAA - 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

- Unknown                                       Not done                                       Albumin  
 Alanine aminotransferase (ALT)    Aspartate aminotransferase (AST)    Total bilirubin  
 Creatinine     Lactate dehydrogenase (LDH)                       PT/PTT

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 \_\_\_\_\_

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 \_\_\_\_\_

Total bilirubin units    umol/L    mg/dL

Total bilirubin reference range \_\_\_\_\_

Creatinine    Within normal limits    Abnormal    In progress    Results unavailable

Creatinine value \_\_\_\_\_

Creatinine units \_\_\_\_\_

Creatinine reference range \_\_\_\_\_

Lactate dehydrogenase    Within normal limits    Abnormal    In progress    Results unavailable

Lactate dehydrogenase value \_\_\_\_\_

Lactate dehydrogenase units    U/L

Lactate dehydrogenase reference range \_\_\_\_\_

PT/PTT    Within normal limits    Abnormal    In progress    Results unavailable

PT/PTT value \_\_\_\_\_

PT/PTT 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 <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"/>

Monocyte #  Within normal limits  Abnormal  In progress  Results unavailable

Monocyte # value \_\_\_\_\_

Monocyte # units \_\_\_\_\_

Monocyte # reference range \_\_\_\_\_

Eosinophil #  Within normal limits  Abnormal  In progress  Results unavailable

Eosinophil # value \_\_\_\_\_

Eosinophil # units \_\_\_\_\_

Eosinophil # reference range \_\_\_\_\_

Basophil #  Within normal limits  Abnormal  In progress  Results unavailable

Basophil # value \_\_\_\_\_

Basophil # units \_\_\_\_\_

Basophil # reference range \_\_\_\_\_

### Urinalysis

---

Urinalysis performed  Unknown  Yes  No

Urine collection date

Urine color  Unknown  Dark yellow  Pale yellow  Nearly colorless  Brown  Pink

Urine clarity  Clear  Slightly cloudy  Cloudy  Turbid

Blood in the urine  Unknown  Yes  No

Urine RBCC  Within normal limits  Abnormal  In progress  Results unavailable

Urine RBCC value \_\_\_\_\_

Urine RBCC units \_\_\_\_\_

Urine RBCC reference range \_\_\_\_\_

Urine WBCC  Within normal limits  Abnormal  In progress  Results unavailable

Urine WBCC value \_\_\_\_\_

Urine WBCC units \_\_\_\_\_

Urine WBCC reference range \_\_\_\_\_

Microorganisms in the urine  Unknown  None  Few  Moderate  Many

Hyaline casts per low per field \_\_\_\_\_

Crystals in the urine  Unknown  Yes  No

Crystals in the urine-specify

Urinalysis results

Unknown

Specific gravity

pH

Protein

Glucose

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"/>

- 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 \_\_\_\_\_

**Glomerular Filtration Rate**

Glomerular filtration rate (GFR) results

 Unknown  Not done  Creatinine clearance  Inulin clearance Iothalamate clearance  51Cr EDTA  99Tc DTPAGlomerular filtration rate (GFR) tests date      Creatinine clearance  Within normal limits  Abnormal  In progress  Results unavailable

Creatinine clearance value \_\_\_\_\_

Creatinine clearance units  mL/min

Creatinine clearance reference range \_\_\_\_\_

Inulin clearance  Within normal limits  Abnormal  In progress  Results unavailable

Inulin clearance value \_\_\_\_\_

Inulin clearance units  mL/min

Inulin clearance reference range \_\_\_\_\_

Iothalamate clearance  Within normal limits  Abnormal  In progress  Results unavailable

Iothalamate clearance value \_\_\_\_\_

Iothalamate clearance units  mL/min

Iothalamate clearance reference range \_\_\_\_\_

51Cr EDTA clearance  Within normal limits  Abnormal  In progress  Results unavailable

51Cr EDTA clearance value \_\_\_\_\_

51Cr EDTA clearance units  mL/min

51Cr EDTA clearance reference range \_\_\_\_\_

99Tc DTPA clearance  Within normal limits  Abnormal  In progress  Results unavailable

99Tc DTPA clearance value \_\_\_\_\_

99Tc DTPA clearance units  mL/min

99Tc DTPA clearance reference range \_\_\_\_\_



Patient Name \_\_\_\_\_

Date      **Biomarkers**

---

## Pompe biomarkers

- Unknown  Not done
- BNP  Creatine Kinase
- Creatine Kinase-MB  Factor VII (proconvertin stable factor)
- Hex4

BNP  Within normal limits  Abnormal  In progress  Results unavailable

BNP value \_\_\_\_\_

BNP units \_\_\_\_\_

BNP reference range \_\_\_\_\_

Creatine Kinase  Within normal limits  Abnormal  In progress  Results unavailable

Creatine Kinase value \_\_\_\_\_

Creatine Kinase units \_\_\_\_\_

Creatine Kinase reference range \_\_\_\_\_

Creatine Kinase-MB  Within normal limits  Abnormal  In progress  Results unavailable

Creatine Kinase-MB value \_\_\_\_\_

Creatine Kinase-MB units \_\_\_\_\_

Creatine Kinase-MB reference range \_\_\_\_\_

Factor VII (proconvertin stable factor)  Within normal limits  Abnormal  In progress  Results unavailable

Factor VII value \_\_\_\_\_

Factor VII units \_\_\_\_\_

Factor VII reference range \_\_\_\_\_

Hex4  Within normal limits  Abnormal  In progress  Results unavailable

Hex4 value \_\_\_\_\_

Hex4 units \_\_\_\_\_

Hex4 reference range \_\_\_\_\_

Anti-drug antibodies  Within normal limits  Abnormal  In progress  Results unavailable

Anti-drug antibodies value \_\_\_\_\_

Anti-drug antibodies units \_\_\_\_\_

Anti-drug antibodies reference range \_\_\_\_\_

CRIM status monitoring  Unknown  Not done  Positive  Negative**Other Labs**

---

Other labs collected  Unknown  Yes  NoNumber of other labs collected  1  2  3  4 or more

Other lab 1 name \_\_\_\_\_

Patient Name \_\_\_\_\_

Date

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

Other lab 1 value \_\_\_\_\_

Other lab 1 units \_\_\_\_\_

Other lab 1 reference range \_\_\_\_\_

Other lab 2 name \_\_\_\_\_

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

Other lab 2 value \_\_\_\_\_

Other lab 2 units \_\_\_\_\_

Other lab 2 reference range \_\_\_\_\_

Other lab 3 name \_\_\_\_\_

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

Other lab 3 value \_\_\_\_\_

Other lab 3 units \_\_\_\_\_

Other lab 3 reference range \_\_\_\_\_

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

### Home Monitoring

---

Home monitoring recommended  Unknown  Yes  No

Home monitoring done since the last outpatient visit  Unknown  Yes  No

Type of home monitoring \_\_\_\_\_

### Comments

---

Labs comments

Patient Name \_\_\_\_\_

Date      

## ***GAA - 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" 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"/>

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

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

Reason for third surgery-Other, specify \_\_\_\_\_

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

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