# Appendix 11a Clinical Record Form for Gaucher disease patients

Gaucher Patient:

Date of visit ___/ ___ / _____

# Gaucher CRFs

## Inclusion Criteria

Has Clinician given consent for the patient or their carer to be approached?

No ☐ Do not continue
Yes ☐

Has the patient given informed signed consent?

No ☐ Do not continue
Yes ☐

Has the parent/carer given informed signed consent?

No ☐ Do not continue
Yes ☐

Reason for not giving consent (if given):

_____

The patient is consented for:

Records Only ☐
Records and Questionnaires ☐

Hospital ID _____

Patient Identification number _____

Date of consent __ __ / __ __ / __ __ __ __

Version of consent form: _____

# Patient Information

**General Patient Information for Gauchers Adults and Children**

To be completed following consent

Date of birth __ __ / __ __ / __ __ __ __

Patient age, in years, at date of consent _____

Gender: Male ☐
Female ☐

Type of Gaucher
☐ Type 1 (Adult onset, no abnormal eye movement)
☐ Type 3 (Abnormal eye movement)

Does the patient have a carer? No ☐
Yes ☐

**Onset of first symptom**

The date of onset of first symptom is:
Roughly / exactly / can't remember: Month and Year ___ / ____ if month unknown just enter year

Other family members affected?
No ☐
Yes ☐
Unknown ☐

If Yes how many --
Relationship/s to patient Mother ☐
Father ☐
Sibling ☐
Other ☐ Please specify_____

Month and Year of Diagnosis Month and Year _____/_____

**Method of diagnosis** (tick any appropriate)
Recorded ☐
Not recorded ☐

**Glucocerebrosidase Enzyme** Normal ☐
Deficient ☐
Not recorded ☐

**DNA mutation** Yes ☐ No ☐ Not known ☐

If yes amino acid effect:
Pick one

N370S/L444P ☐
N370S/N370S ☐
N370S/84GG ☐
N370S/D409H ☐
N370S/IVS2+1 ☐
N370S/unknown☐
L444P/unknown ☐
L444P/D409H ☐
L444P/L444P ☐
Other please specify: ----------

**Bone marrow Biopsy** negative ☐ positive ☐ Not recorded ☐

**Current Employment Status**

Paid Employed ☐
Unemployed ☐
Unpaid Employment ☐

Full time ☐
Part time ☐

# Type of Gauchers:

| Definitions for classification: | |
|---|---|
| Type 1 | Adult onset, no abnormal eye movement (if not type 2 or 3 then type 1) |
| Type 2 | Infantile onset, acute neuronopathic |
| Type 3 | Abnormal eye movement |

**Type 1**

Does the patient have Neurological involvement? No ☐
Yes ☐ If Yes:

Peripheral neuropathy: Yes ☐ No ☐
Parkinsonian features: Yes ☐ No ☐ If yes:

Age of presentation _____

Is the patient on treatment for Parkinsonian features? No ☐
Yes ☐

Please specify _____

Does the patient have Dementia: No ☐
Yes ☐

Does the patient have another type of neurological involvement?
No ☐
Yes ☐

Please Specify _____

**Type 3** ☐
**Abnormal eye movement** No ☐ Yes ☐

Has diagnosis changed before entry on to study? No ☐ Yes ☐

Please give date of change of diagnosis from Type 1 To: Type 3 __ __ / __ __ / __ __ __ __

## Reason for diagnosis:

**Was the patient detected by family screening** No ☐
Yes ☐

**Chitotriosidase genotype** +/+ normal
+/-
-/- no chito, null

Measure date: __ __ / __ __ / __ __ __ __ (moved)

**Did the patient present with symptoms** No ☐
Yes ☐

What symptoms did the patient present with?

**Type 1:** (pick one or more)
Organomegaly ☐
Bone pain ☐
Unexpected finding in biopsy ☐
Other (please specify) ☐_____
During pregnancy ☐
*Full blood count*: *Thrombocytopenia* ☐
*Complications during delivery* ☐
*Antenatal test- Family screening* ☐
*(Previous child with LSD)*

**Type 3:**
Is presentation: Visceral ☐
Neurological ☐

# Withdrawal from the study

**Is the patient still in the full study?** No ☐
Yes ☐

Withdrawal date: __ __ / __ __ / __ __ __ __

Reason for Withdrawal

☐ Patient has life threatening disease
☐ Failure to comply (due to medication)
☐ Evidence of disease progression
☐ Patient is pregnant
☐ Death
☐ Patient is on a clinical trial
☐ Questionnaire burden
☐ Patient turned 16 and didn't re-consent
☐ Other Please specify_____

Continue in the study notes only? No ☐
Yes ☐

**Record of death**

Date of death: __ __ / __ __ / __ __ __ __

Did clinician certify this death as condition related? No ☐
Yes ☐

Please record the wording from the dearth certificate:

Part 1a ---------- Free text (Disease or condition directly leading to death)

Part 1b ---------- Free text (Other disease or condition, if any, leading to 1a)

Part 1c ---------- Free text (Other disease or condition, if any, leading to 1b)

Part 2 ---------- Free text (Other significant conditions CONTRIBUTING TO THE DEATH but not related to the disease or condition causing it)

# Haematological

**Spleen**

**Enlarged or normal from scan?** **No** ☐
**Yes** ☐

**Measure date: __ __ / __ __ / __ __ __ __**

**Size or volume available from scan?** **No** ☐
**Yes** ☐

Volume: ____ ml

**And/Or**

**Size (cm) _____**

**Measure date: __ __ / __ __ / __ __ __ __**

**Enlarged on palpation** **No** ☐
**Yes** ☐

**Measure date: __ __ / __ __ / __ __ __ __**

**Has the patient been Splenectomised?** No ☐
Yes ☐

Reason for Splenectomy: _____

Date of splenectomy __ __ / __ __ / __ __ __ __

Splenectomy: full ☐
partial ☐

**Platelet count**
Done ☐
Not done ☐

**Platelet** count _____$10^9$/l Measure date: __ __ / __ __ / __ __ __ __

**Haemoglobin**
Done ☐
Not done ☐

**Haemoglobin**___ g/dl (on Full Blood Count**)** Measure date: __ __ / __ __ / __ __ __ __

Bleeding Episodes **-** has the patient had unexplained bleeding or bruising in the previous 12 months?

Yes ☐
No ☐

Chitotriosidase level _____ **Pick units _____**

**Liver Organ volume**

**Enlarged or normal from scan?** **No** ☐
**Yes** ☐

**Measure date: __ __ / __ __ / __ __ __ __**

**Size or volume available from scan?** **No** ☐
**Yes** ☐

**Volume**: ____ ml

**And/Or**

**size (cm) _____**

**Measure date: __ __ / __ __ / __ __ __ __**

**Enlarged on palpation** **No** ☐
**Yes** ☐

**Measure date: __ __ / __ __ / __ __ __ __**

**Liver damage –** does the patient have liver fibrosis or cirrhosis? Yes ☐
No ☐
Test not carried out ☐

**Does the patient have - Portal hypertension- (on ultrasound)?** Yes ☐
No ☐
Test not carried out ☐

**Has the patient had a Liver Transplant in the last 12 months?** Yes ☐
No ☐

If Yes: date of transplant: __ __ / __ __ / __ __ __ __

**Liver function test**
GGT level (Gamma-glutamyl transpeptidase)
Done ☐
Not done ☐
Measure date: __ __ / __ __ / __ __ __

GGT level results _____U/l

ALT Level (Alanine transaminase)
Done ☐
Not done ☐
Measure date: __ __ / __ __ / __ __ __

ALT level results _____U/l

**Variceal Haemorrhage** – how many episodes in the previous six months? _____ (number)

# Bone, bone marrow

**Bone pain –** has the patient had bone pain in the last 12 months? Yes ☐
No ☐
Not recorded ☐

If yes is it constant ☐
sporadic ☐

**Analgesia required –** has the patient had to use analgesia in the last 6 months? Yes ☐ No ☐
If yes have they had to use opiate analgesic? Yes ☐ No ☐

**MRI**
**Avascular necrosis** - are there any new avascular necrosis on MRI in the last 12 months?
Yes ☐
No ☐
Not done ☐

Date of MRI scan __ __ / __ __ / __ __ __

**Bone Marrow Burden on MRI**
Done ☐
Not done ☐

BMB Score: _____ Measure date: __ __ / __ __ / __ __ __

**Bone crises -** has the patient had doctor-diagnosed bone crisis in the last 12 months?
Yes ☐
No ☐
Not recorded ☐
How many? _____

**Has the patient had Joint replacement surgery in the last 12 months?** Yes ☐ No ☐

If Yes: date of replacement __ __ / __ __ / __ __ __ (must be before the date of the visit to the clinic)

**Osteoporosis**
Has the patient had Fragility fractures in the last 12 months? Yes ☐
No ☐
If Yes what is the number of fractures the patient has had _____ (number)

**DEXA – Bone Marrow Density**

Done ☐
Not done ☐
Measure date: __ __ / __ __ / __ __ __

**Bone Mineral Density T score (Adult)**

Forearm total ____
Hip Total ____
Lumbar Spine total ____

**Bone Mineral Density Z score (Children under 16)**

Forearm total ____
Hip Total ____
Lumbar Spine total ____

**Is the patient on Bisphosphonates** ? Yes ☐ No ☐

# Neurological

**Any abnormal eye movement**? Yes ☐ No ☐

**Cognitive measures –**
**IQ Test**
done ☐ not done ☐ IQ score (whole number)___

Measure date: __ __ / __ __ / __ __ __ __

**Developmental Quotient Test**
done ☐ not done ☐ DQ score (whole number) ___

Measure date: __ __ / __ __ / __ __ __ __

**Has the patient had fits in the last 12 months?** Yes ☐ No ☐

**Does the patient have extrapyramidal involvement?** Yes ☐ No ☐

# Other Measures

**Lung**
Does the patient have pulmonary hypertension - from Echo?
Yes ☐
No ☐
Not done ☐

Measure date: __ __ / __ __ / __ __ __ __

**Does the patient have malignancies?** No ☐ Yes ☐ If Yes, please specify: _____

**Has a severity score been recorded?** Yes ☐ No ☐ Measure date: __ __ / __ __ / __ __ __ __

If yes, with: Zimran Severity Score ☐
Other severity Score ☐

Severity score: ____ (Whole number)

# Neurological - Child

**Any abnormal eye movement**? Yes ☐ No ☐

**IQ Test**
done ☐ not done ☐ IQ score (whole number)___

Measure date: __ __ / __ __ / __ __ __ __

**Developmental Quotient Test**
done ☐ not done ☐ DQ score (whole number) ___

Measure date: __ __ / __ __ / __ __ __ __

**Has the patient had fits in the last 12 months?** Yes ☐ No ☐

**Does the patient have extrapyramidal involvement?** Yes ☐ No ☐

**Using any parent reported Motor milestones scale, are the patients able to do the following? If they are able, what age in months was the milestone met?**

Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Patient currently able to sit unsupported: Able ☐ Months ____
Unable ☐

Patient currently able to stand independently: Able ☐ Months ____
Unable ☐

Patient currently able to walk: Able ☐ Months ____
Unable ☐

Patient currently able to walk upstairs one step at a time: Able ☐ Months ____
Unable ☐

**Does the patient have a squint?** Yes ☐ No ☐

**Are Brainstem evoked responses normal?** Yes ☐ No ☐

Measure date: __ __ / __ __ / __ __ __ __

# Audiometry

Hearing Test:
Done ☐
Not done ☐
Measure date: __ __ / __ __ / __ __ __ __

Hearing Normal: Yes ☐
No ☐ If No….

| **Type of hearing loss (pick one)** | | **Degree of hearing loss (average decibels over tested frequencies) (pick one)** | |
|---|---|---|---|
| Conductive | ☐ | Mild (0-40 decibels) | ☐ |
| Sensorineural | ☐ | Moderate (41-70 decibels) | ☐ |
| Mixed (combination of conductive & sensorineural | ☐ | Severe (71-95 decibels) | ☐ |
| | | Profound (over 95 decibels) | ☐ |

Note: to calculate the degree of hearing loss, add all the decibels and divide by the number of data points.

# **Severity scoring tool** - for Neuropathic Gaucher Disorder

Done ☐
Not done ☐

Measure date: __ __ / __ __ / __ __ __ __

**Horizontal gaze palsy**

- ☐ Yes, on a regular basis
- ☐ Yes, sometimes
- ☐ No

**Epilepsy**

- ☐ Yes, on a regular basis
- ☐ Yes, sometimes
- ☐ No

**Development/ cognitive ability**

- ☐ Normal
- ☐ Mildly impaired (IQ less than 85 or equivalent)
- ☐ Moderate (IQ between 50–57 or equivalent)
- ☐ Severe (more than half their chronological age)

**Neurology pattern Ataxia/ gait**

- ☐ Normal, apparent only on tandem walking
- ☐ Ataxia on straight gait, able to walk without assistance
- ☐ Able to walk only with assistance
- ☐ Unable to walk

**Cerebellar signs/ataxia**

- ☐ No intention tremor
- ☐ Intention tremor not affecting function
- ☐ Intention tremor with marked impact on function

**Pyramidal**

- ☐ Normal tone with increased reflexes
- ☐ Mildly to moderately increased tone and reflexes
- ☐ Increased tone reflexes with sustained/unsustained clonus
- ☐ Severe spasticity with inability to walk

**Extrapyramidal**

- ☐ Normal
- ☐ Variable tone and posturing not impairing function, with or without therapy
- ☐ Variable tone and posturing impairing function, despite therapy
- ☐ Significant rigidity with no/minimal benefit from therapy

**Swallowing difficulties/oral bulbar function**

- ☐ Normal
- ☐ Mild dysphagia (excess drooling)
- ☐ Moderate dysphagia (risk of aspiration, modification to diet required)
- ☐ Severe dysphagia (requiring non-oral feeding)

**Speech**

- ☐ Normal (and those too young yet to speak)
- ☐ Mild to moderate dysarthria impairing intelligibility to unfamiliar listener
- ☐ Severe dysarthria with most speech unintelligible to familiar and unfamiliar listener
- ☐ Anarthria

**Ophthamology**

- ☐ Normal
- ☐ Cranial nerve palsy (previously corrected or not)
- ☐ Cranial nerve palsy (reappearing despite surgical correction)

**Spinal alignment (kyphosis)**

- ☐ Normal
- ☐ Mild kyphosis – but flexible
- ☐ Moderate kyphosis – partially corrected
- ☐ Severe kyphosis – fixed

Total Calculated (maximum 33) ☐

# Growth

**Body measurements**

Measure date: __ __ / __ __ / __ __ __ __

Height _____cm
Weight _____kg
Head circumference _____cm

**Delayed puberty** Yes ☐ No ☐ inappropriate ☐

# Other Measures

**Lung**

Does the patient have pulmonary hypertension from Echocardiogram?

Yes ☐
No ☐
Not done ☐

Measure date: __ __ / __ __ / __ __ __ __

# Significant Co-morbidity

**Does the patient have any co-morbidities? Yes / No**

**How many? ____**

Please enter significant co-morbidities in the spaces below, and then answer yes or no to the organs affected

1

2

3

4

5

6

| | **1** | **2** | **3** | **4** | **5** | **6** |
|---|---|---|---|---|---|---|
| Haematological | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | |
| Bone, bone marrow | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | |
| Neurological | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | |
| Lungs | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | |

**Children Only:**

| | | | | | | |
|---|---|---|---|---|---|---|
| Audiometry: | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No |
| Growth | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No |

# Con-meds

**Is the patient on any con meds?**

| Medication | Prescription? | Dose | Frequency | Date started | Date finished | Ongoing? |
|---|---|---|---|---|---|---|

# Record of Therapy (Enzyme Replacement Therapy, substrate reduction therapy etc):

Is patient on treatment No ☐
Yes ☐

**Initial Treatment**
Initially treatment ERT ☐
clinical trial? ☐

Date of first treatment: __ __ / __ __ / __ __ __ __

Age when first treatment received: _____

Initial type of treatment (ERT): _____

Initial weight ____(kg)

Initial dose: ___(units)

Initial frequency: 3 times per week ☐
2 times per week ☐
weekly ☐
every 2 weeks ☐
every 3 weeks ☐
monthly ☐

Initial type of treatment (substrate reduction therapy)

Initial dose: ___mg

Initial frequency: 3 times per week ☐
2 times per week ☐
1 time per week ☐
2 weekly ☐
Other ☐ Please Specify_____

**Current treatment**

Current treatment

Date started current treatment ___/ ___ / _____ (dd/mm/yyyy)

Current type of treatment (ERT)

Current weight (kg)

Current dose: ___units

Current frequency: 3 times per week ☐
2 times per week ☐
weekly ☐
every 2 weeks ☐
every 3 weeks ☐
monthly ☐

Hospital/ home infusion: Infusion in Hospital ☐
Infusion at Home ☐

Home infusion Nurse infuses ☐
Nurse cannulates and leaves ☐
Patient cannulates and infuses ☐

Is the patient currently experiencing:

Febrile reactions? No ☐
Yes ☐

Anaphylactoid reactions? No ☐
Yes ☐

Does the patient require any pre-medication:
No ☐
Yes ☐

Current type of treatment (substrate reduction therapy) _____

Current Dose _____(mg)

Current frequency: 3 times per week ☐
2 times per week ☐
1 time per week ☐
2 weekly ☐
Other ☐
Please Specify_____

**Antibody status**

Measure date: __ __ / __ __ / __ __ __ __

Antibody status to infused product, Tested No ☐
Yes ☐

Antibody Status, Test Result Positive No ☐
Yes ☐
Don't know ☐

# Record of Therapy (Enzyme Replacement Therapy, Substrate Reduction Therapy etc):

Is patient on treatment No ☐
Yes ☐

Has patient stopped treatment since their last visit? No ☐
Yes ☐

Has patient started treatment since their last visit? No ☐
Yes ☐

**Initial Treatment**
Initial treatment _____

Date of first treatment: __ __ / __ __ / __ __ __ __

Initial type of treatment (ERT):

Initial weight ____ kg

Initial dose: ___units

Initial frequency: Current frequency:
3 times per week ☐
2 times per week ☐
weekly ☐
every 2 weeks ☐
every 3 weeks ☐
monthly ☐

Initial type of treatment (substrate reduction therapy) _____

Initial dose: ___mg

Initial frequency: Current frequency:
3 times per week ☐
2 times per week ☐
1 time per week ☐
2 weekly ☐
Other ☐ Please Specify_____

**Current treatment**

Current treatment _____

Date started current treatment _____ (dd/mm/yyyy)

Current type of treatment (ERT) _____-

Current weight ___kg

Current Dose ____ units

Current frequency: 3 times per week ☐
2 times per week ☐
weekly ☐
every 2 weeks ☐
every 3 weeks ☐
monthly ☐

Hospital/ home infusion: Infusion in Hospital ☐
Infusion at Home ☐

Home infusion Nurse infuses ☐
Nurse cannulates and leaves ☐
Patient cannulates and infuses ☐

Is the patient currently experiencing:
Febrile reactions? No ☐
Yes ☐

Anaphylactoid reactions? No ☐
Yes ☐

Does the patient require any pre-medication: No ☐
Yes ☐

Current type of treatment (substrate reduction therapy) _____-

Current dose: ___mg

Current frequency: 3 times per week ☐
2 times per week ☐
1 time per week ☐
2 weekly ☐

**Antibody status**

Measure date: __ __ / __ __ / __ __ __ __

Antibody status to infused product, Tested No ☐
Yes ☐

Antibody Status Test Positive No ☐
Yes ☐

**Stopped Treatment**
Final treatment ERT ☐
clinical trial ☐

Date of last treatment: __ __ / __ __ / __ __ __ __

Final type of treatment (ERT)_____

Final weight ____kg

Final dose: ___units

Final frequency: 3 times per week ☐
2 times per week ☐
weekly ☐

every 2 weeks ☐
every 3 weeks ☐
monthly ☐

Hospital/ home infusion: Infusion in Hospital ☐
Infusion at Home ☐

Home infusion Nurse infuses ☐
Nurse cannulates and leaves ☐
Patient cannulates and infuses ☐

Final type of treatment (substrate reduction therapy)_____

Final dose: ___mg

Final frequency: 3 times per week ☐
2 times per week ☐
1 time per week ☐
2 weekly ☐
Other ☐ Please Specify_____

Why was treatment stopped? (Drop down menu)

Please specify _____

Visit Date: _____

# Appendix 11b Clinical Record Form for Fabry disease patients

Fabry Patient ID:

Date of visit ___/ ___ / _____

# Fabry CRFs

## Inclusion Criteria

Has Clinician given consent for the patient or their carer to be approached?

No ☐ Do not continue
Yes ☐

Has the patient given informed signed consent?

No ☐ Do not continue
Yes ☐

Has the consultee given informed signed assent?

No ☐ Do not continue
Yes ☐

Reason for not giving consent:

---

Patient has consented for:

Records Only ☐
Records and Questionnaires ☐

Patient Identification number _____

Date of consent __ __ / __ __ / __ __ __ __

Version of consent form: _____

# Patient Information

To be completed following consent

Date of birth __ __ / __ __ / __ __ __ __

Gender: Male ☐
Female ☐

Does the patient have a carer? No ☐
Yes ☐

Onset of first symptom – month and year
Roughly / exactly / can't remember: ___ / ____

Other family members affected?
No ☐
Yes ☐ If Yes how many --
Relationship/s to patient Mother ☐
Father ☐
Sibling ☐
Other ☐ Please specify_____
(enter unique ID no.s of relations)

Month & Year of diagnosis
Month ------- Unknown ☐ If Month unknown: Year -------

**Method of diagnosis** (tick any appropriate)

**Alpha-galactosidase A activity**
**- Leucocyte** Normal ☐ Also known as GLA, a-GLA, GALA
Deficient ☐

**Alpha-galactosidase A activity**
**- plasma** Normal ☐
Deficient ☐

**Alpha-L-iduronidase cDNA** Done ☐
Not done ☐

**cDNA results as amino acid change** _____
Please use the following format
e.g. Asn272Ser/Asn272Ser

**Prenatal Diagnosis** No ☐
**(chorion villus biopsy)** Yes ☐

**Enzyme assay results** _____

**Urine GAG test** No ☐
Yes ☐ Result _____

**Reason for diagnosis**

**Was patient detected by family screening?** No ☐
Yes ☐

**Did patient present with symptoms?** No ☐
Yes ☐

**Which symptoms?**
- Angiokeratoma ☐
- Cornea verticilata ☐
- Sweating abnormality ☐
- GI symptoms ☐
- Stroke, cardiomyopathy ☐
- Renal failure ☐
- Other – please specify ☐ _____

**Physician of initial presentation**
- cardiologist ☐
- neurologist ☐
- dermatologist ☐
- ophthalmologist ☐
- geneticist ☐
- paediatrician ☐
- gastroenterologist ☐
- Other – please specify ☐ _____

**Employment Status**
- Paid Employed ☐
- Unemployed ☐
- Unpaid Employment ☐

If employed
- Full time ☐
- Part time ☐

# Withdrawal from the study

**Is the patient still in the full study?** No ☐
Yes ☐

Withdrawal date: __ __ / __ __ / __ __ __ __

Reason for Withdrawal

☐ Patient has life threatening disease
☐ Failure to comply (due to medication)
☐ Evidence of disease progression
☐ Patient is pregnant
☐ Death
☐ Patient is on a clinical trial
☐ Questionnaire burden
☐ Patient turned 16 and didn't reconsent
☐ Other Please specify_____

Continue in the study notes only? No ☐
Yes ☐

**Record of death**

Date of death: __ __ / __ __ / __ __ __ __

Did clinician certify this death as condition related? No ☐
Yes ☐

Please record the wording from the dearth certificate:

Part 1a ---------- Free text (Disease or condition directly leading to death)

Part 1b ---------- Free text (Other disease or condition, if any, leading to 1a)

Part 1c ---------- Free text (Other disease or condition, if any, leading to 1b)

Part 2 ---------- Free text (Other significant conditions CONTRIBUTING TO THE DEATH but not related to the disease or condition causing it)

# Body Measurements

**Height**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Height (cm) _____

**Weight**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Weight (Kg) _____

# Heart Measures

**Echocardiogram** Done ☐
Not done ☐ Measure date: __ __ / __ __ / __ __ __ __

**LVEDD** _____mm LV End Diastole
Left Ventricular Diastolic diameter (LVDd)

**IVSd** _____ mm Interventricular septal diastolic diameter

**PWTd** _____ mm Posterior Wall thickness /
posterior LV Wall diastole diameter

**Result (Calculated by database)**
**LVmass**_____ g

**Body Surface Area** _____ m squared
(**BSA**)

**Result**
**LVMI** _____ g /m squared
Measure of Hypertrophy

Does the patient have clinically significant Arrhythmia? No ☐
Yes ☐

Does the patient have a pacemaker? No ☐
Yes ☐

Measure date: __ __ / __ __ / __ __ __ __

Date of fitting pacemaker: __ __ / __ __ / __ __ __ __

Does the pacemaker have a defibrillator? No ☐
Yes ☐

Does the patient have heart failure? No ☐
Yes ☐

Measure date: __ __ / __ __ / __ __ __ __

NYHA Category _____-

**PR Interval – Children Only**

Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

PR Interval – results _____ milliseconds

# Neurological

**Has the patient ever had a TIA / stroke?**

No ☐
Yes ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Age at 1st stroke ___

Does the patient have any persistent neurological impairment from the stroke?
No ☐
Yes ☐

Has the patient had a TIA/stroke since their last assessment?
No ☐
Yes ☐

Is there evidence of cognitive impairment?
No ☐
Yes ☐

# Kidney

**Spot Protein / Creatinine Ratio**

Done ☐
Not done ☐ Measure date: __ __ / __ __ / __ __ __ __

Results _____ mg/mmol

**Albumin / Creatinine Ratio**

Done ☐
Not done ☐ Measure date: __ __ / __ __ / __ __ __ __

Results _____ mg/mmol

**Serum Creatinine**

Done ☐
Not done ☐ Measure date: __ __ / __ __ / __ __ __ __

Results _____ micromoles / l

**Estimated Glomerular Filtration Rate (eGFR)**

Done ☐
Not done ☐ Measure date: __ __ / __ __ / __ __ __ __

Results _____

**Dialysis**

No ☐
Yes ☐

Start date: __ __ / __ __ / __ __ __ __

Is the patient still on dialysis?
No ☐
Yes ☐

End date: __ __ / __ __ / __ __ __ __

Type of dialysis Peritoneal ☐
Haemo ☐

**Has the patient had a kidney transplant?**

No ☐
Yes ☐

Date of transplant: __ __ / __ __ / __ __ __ __

# Audiometry

**Evidence of hearing loss?** No ☐
Yes ☐

**Hearing Test:** Done ☐
Not done ☐

**Measure date:** __ __ / __ __ / __ __ __ __

**Type of hearing loss (pick one)**

Conductive ☐
Sensorineural ☐
Mixed (combination of conductive & sensorineural ☐

**Degree of hearing loss (average decibels over tested frequencies) (pick one)**

Mild (0-40 decibels) ☐
Moderate (41-70 decibels) ☐
Severe (71-95 decibels) ☐
Profound (over 95 decibels) ☐

Note: to calculate the degree of hearing loss, add all the decibels and divide by the number of data points

**Does the patient have a hearing aid?** No ☐
Yes ☐
Not recorded ☐

**Type of hearing aid:** _____

**Does the patient have tinnitus?** No ☐
Yes ☐
Not recorded ☐

**Measure date:** __ __ / __ __ / __ __ __ __

**Date of onset of tinnitus:** __ __ / __ __ / __ __ __ __

# Other Measures

**Brief Pain Inventory**

Done ☐
Not done ☐

**Measure date:** __ __ / __ __ / __ __ __ __

**Pain Severity** _____ mean of 4 pain items: questions 3-6

**Pain interference** _____ mean of 7 interference items: questions 9 a-g
(should only be calculated if more than 50%, or 4 out of 7 questions complete)

**Sweating**
Recorded ☐
Not recorded ☐

**Measure date:** __ __ / __ __ / __ __ __

Normal ☐
Increased ☐
Reduced ☐
Absent ☐

**Gastrointestinal**
In the last tree months has the patient experienced quality of life limiting GI symptoms?

No ☐
Yes ☐
Not recorded ☐

**Mood**
Are the clinical team aware of any suicide attempts in the last year?

No ☐
Yes ☐

Do the clinical team believe that the patient has clinical depression?

No ☐
Yes ☐

This was ascertained from:

A formal measure ☐
A clinical judgement ☐

# Significant Co-morbidity

Please enter up to three significant co-morbidities in the spaces below, and then answer yes or no to the organs affected

1

2

3

| | **1** | **2** | **3** |
|---|---|---|---|
| Heart | Yes / No | Yes / No | Yes / No |
| Neurological | Yes / No | Yes / No | Yes / No |
| Kidneys | Yes / No | Yes / No | Yes / No |
| Audiometry | Yes / No | Yes / No | Yes / No |
| Pain | Yes / No | Yes / No | Yes / No |
| Sweating | Yes / No | Yes / No | Yes / No |
| GI | Yes / No | Yes / No | Yes / No |

**Children Only:**

| | | | |
|---|---|---|---|
| Growth | Yes / No | Yes / No | Yes / No |

# Con-meds

| Medication | Prescription? | Dose | Frequency | Date started | Date finished | Ongoing? |
|---|---|---|---|---|---|---|

# Record of Therapy

**Record of Therapy (Enzyme Replacement Therapy, substrate reduction therapy etc):**

Is patient on ERT
No ☐
Yes ☐

**Initial Treatment**
Initially on
ERT ☐
clinical trial? ☐

Date of first Infusion: __ __ / __ __ / __ __ __ __

Age when first infusion received: _____

Initial type of treatment:
Fabrazyme ☐
Replagal ☐

Initial dose: ___ (mg)

Initial frequency: _____

**Current treatment**
Currently on
ERT ☐
Clinical trial? ☐

Current dose: ___ mg

Current frequency: Please Specify_____

Hospital/ home infusion:
Infusion in Hospital ☐
Infusion at Home ☐
If Home: pick list:
Nurse infuses ☐
Nurse cannulates and leaves ☐
Patient cannulates and infuses ☐
Relative cannulates and infuses ☐

Is the patient currently experiencing:

Febrile reactions?
No ☐
Yes ☐

Anaphylactoid reactions?
No ☐
Yes ☐

Other pre-medication:
No ☐
Yes ☐

**Antibody status**

Measure date: __ __ / __ __ / __ __ __ __

| | | | | | | |
|---|---|---|---|---|---|---|
| Antibody status to infused product, Tested | No | ☐ | | | | |
| | Yes | ☐ | If Yes, | Positive | No | ☐ |
| | | | | | Yes | ☐ |
| | | | | | Don't know | ☐ |

# Record of Therapy

**Record of Therapy (Enzyme Replacement Therapy, substrate reduction therapy etc):**

Is patient on ERT
No ☐
Yes ☐

Has patient stopped treatment since their last visit?
No ☐
Yes ☐

Has patient started treatment since their last visit?
No ☐
Yes ☐

**Initial Treatment**

Initially on
ERT ☐
clinical trial? ☐

Date of first Infusion: __ __ / __ __ / __ __ __ __

Age when first infusion received: _____

Initial type of treatment:
Fabrazyme ☐
Replagal ☐

Initial dose: ___ (mg)

Initial frequency: _____

**Current treatment**

Currently on
ERT ☐
Clinical trial? ☐

Current dose: ___ mg

Current frequency: Please Specify_____

Hospital/ home infusion:
Infusion in Hospital ☐
Infusion at Home ☐

If Home: pick list:
Nurse infuses ☐
Nurse cannulates and leaves ☐
Patient cannulates and infuses ☐
Relative cannulates and infuses ☐

Is the patient currently experiencing:

Febrile reactions?
No ☐
Yes ☐

Anaphylactoid reactions?
No ☐
Yes ☐

Other pre-medication: No ☐
Yes ☐

**Antibody status**

Measure date: __ __ / __ __ / __ __ __ __

Antibody status to infused product, Tested No ☐
Yes ☐ If Yes, Positive No ☐
Yes ☐
Don't know ☐

**Stopped Treatment**

Final treatment ERT ☐
clinical trial ☐

Date of last treatment: __ __ / __ __ / __ __ __ __

Final type of treatment: Fabrazyme ☐
Replagal ☐

Final dose: ___ (mg)

Final frequency: Please Specify_____

Hospital/ home infusion: Infusion in Hospital ☐
Infusion at Home ☐ If Home: pick list: Nurse infuses ☐
Nurse cannulates and leaves ☐
Patient cannulates and infuses ☐

Why was treatment stopped?

- ☐ Patient has life-threatening disease
- ☐ Failure to comply
- ☐ Evidence of disease pregression
- ☐ Patient is pregnant
- ☐ Death
- ☐ Patient is on a clinical trial
- ☐ Questionnaire burden
- ☐ Patient turned 16 and did not reconsent
- ☐ Other

# Appendix 11c Clinical Record Form for MPS I patients

MPS I Patient ID:

Date of visit ___/ ___ / _____

# MPS I CRFs

## Inclusion Criteria

Has Clinician given consent for the patient or their carer to be approached?

No ☐ Do not continue
Yes ☐

Has the patient given informed signed consent?

No ☐ Do not continue
Yes ☐

Has the parent/carer given informed signed consent?

No ☐ Do not continue
Yes ☐

Reason for not giving consent (if given):

_____

The patient is consented for:

Records Only ☐
Records and Questionnaires ☐

Hospital ID _____

Patient Identification number _____

Date of consent __ __ / __ __ / __ __ __ __

Version of consent form: _____

# Patient Information

**Date of birth** __ __ / __ __ / __ __ __ __

Patient age, in years, at date of consent _____

**Gender**

Male ☐
Female ☐

**Type of MPS I**

Hurler ☐
Hurler-Scheie ☐
Scheie ☐
Not yet known ☐

**Does the patient have a carer**

No ☐
Yes ☐

**Onset of first symptoms (i.e. When did the main presenting symptoms occur)**

The date of onset of first symptoms is:

Roughly ☐
Exactly ☐
Can't remember ☐

Month and Year: _____ / __ If month unknown just enter year

**Any other family members affected?**

No ☐
Yes ☐
Unknown ☐ If so, how many? __

Relationship(s) to patient:

Mother ☐
Father ☐
Sibling ☐
Other ☐

Please Specify _____
(enter Unique ID no of relations)

**Month and Year of Diagnosis**

Month and Year: _____ / __ If month unknown just enter year

**Method of diagnosis**

Recorded ☐
Not recorded ☐

Alpha – L-iduronidase activity

☐ Normal
☐ Deficient

Alpha –L-iduronidase cDNA

☐ Done
☐ Not Done

Allele 1 _____ (drop down menu)
Please specify _____
Allele 2_____ (drop down menu)
Please specify _____

Skin Biopsy – Enzyme Assay ☐ Normal
☐ Abnormal
If abnormal, Result: _____ (put in own units)

Prenatal Diagnosis ☐ No
(chorion villus biopsy) ☐ Yes

Enzyme Assay Results _____(put in own units)

Urine GAG test ☐ No
☐ Yes

Urine GAG test Results _____ (mg/mmol of creatine)

**Reason for diagnosis**
**Was patient detected by family screening?** ☐ No
☐ Yes

**Did the patient present with symptoms?** ☐ No
☐ Yes
If Yes, what symptoms did they present with?

_____

**Current Employment Status** Select as appropriate

| | | | |
|---|---|---|---|
| Employed | ☐ | | |
| Unemployed | ☐ | Full time | ☐ |
| Unpaid Employment | ☐ | Part time | ☐ |

# Withdrawal from the study

**Is the patient still in the full study?** No ☐
Yes ☐

Withdrawal date: __ __ / __ __ / __ __ __ __

Reason for Withdrawal

☐ Patient has life threatening disease
☐ Failure to comply (due to medication)
☐ Evidence of disease progression
☐ Patient is pregnant
☐ Death
☐ Patient is on a clinical trial
☐ Questionnaire burden
☐ Patient turned 16 and didn't re-consent
☐ Other

Please specify_____

Continue in the study notes only? No ☐
Yes ☐

**Record of death**

Date of death: __ __ / __ __ / __ __ __ __

Did clinician certify this death as condition related? No ☐
Yes ☐

Please record the wording from the dearth certificate:

Part 1a ---------- Free text (Disease or condition directly leading to death)

Part 1b ---------- Free text (Other disease or condition, if any, leading to 1a)

Part 1c ---------- Free text (Other disease or condition, if any, leading to 1b)

Part 2 ---------- Free text (Other significant conditions CONTRIBUTING TO THE DEATH but not related to the disease or condition causing it)

# Body Measurements

Measure date: __ __ / __ __ / __ __ __ __

Height (cm) _____

Weight (cm) _____

**Children only:**

Head Circumference (cm) _____ current age: _____

# Heart Measures - Child

Echocardiogram: Done ☐
Not done ☐ Measure date: __ __ / __ __ / __ __ __ __

Left Ventricular Ejection fraction <60%
No ☐
Yes ☐

Valve Disease as reported by cardiologist (on echo)
None ☐
Mild ☐
Moderate ☐
Severe ☐

Valve Disease severe enough to require medication
No ☐
Yes ☐

Valve replacements
No ☐
Yes ☐

Date of valve replacement: __ __ / __ __ / __ __ __ __

# Airway / Lung / ENT - Child

**Sleep Study** Done ☐
Not done ☐

Measure date: __ __ / __ __ / __ __ __ __

Time spent below saturation of 90% ____
(Given as a percentage of time of duration of the study)

Lowest saturation recorded (%) ____
(the value in percent of lowest recording that was not due to an artifact)

**Pulmonary Function Test (best of 3 if there is more than one)**

Patient upright Test done ☐
Test not done ☐
Can't do test ☐

Measure date: __ __ / __ __ / __ __ __ __

FVC (%) _____ percentage of predicted

FEV1 (%) _____ percentage of predicted

**Audiometry**

Using any age-appropriate audiometry test, hearing is:

Normal ☐
Abnormal ☐
Not done ☐

Measure date: __ __ / __ __ / __ __ __ __

| **Type of hearing loss (pick one)** | | **Degree of hearing loss (average decibels over tested frequencies) (pick one)** | |
|---|---|---|---|
| Conductive | ☐ | Mild (0-40 decibels) | ☐ |
| Sensorineural | ☐ | Moderate (41-70 decibels) | ☐ |
| Mixed (combination of conductive & sensorineural | ☐ | Severe (71-95 decibels) | ☐ |
| | | Profound (over 95 decibels) | ☐ |

Note: to calculate the degree of hearing loss, add all the decibels and divide by the number of data points.

**Difficulty with intubation for anaesthesia?** No ☐
Yes ☐
No operation carried out ☐

# Neurological – Child

**DQ Test:**

Done ☐
Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

DQ Score _____

**Hydrocephalus**

Present ☐
Absent ☐
Not Recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

**Carpal Tunnel Syndrome**

Present ☐
Absent ☐
Not Recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

# Bone & Joint – Child

**Patient Mobility**

Test: Applicable ☐
Not Applicable due to age ☐

Measure date: __ __ / __ __ / __ __ __ __

☐ can walk / stand unaided (i.e. can stand for 6 mins or walk for 5 meters)
☐ can walk aided one stick (i.e. can't stand for 6 mins without a stick)
☐ can walk aided more than one stick
☐ is wheelchair bound
☐ is bed-bound (i.e. can't get into wheelchair)

**6 Minute Walk Test**

Done ☐
Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

Distance walked (m) _____ Aided ☐
Unaided ☐

# Liver & Spleen – Child

## Liver

**Enlarged or normal from scan?**

- Enlarged ☐
- Normal ☐
- Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

**Size or volume available from scan?**

- No ☐
- Yes ☐
- Not recorded ☐

Liver organ size (cm) _____

**And/ Or**

Liver volume: ____ ml

Measure date: __ __ / __ __ / __ __ __ __

**Enlarged on palpation**

- No ☐
- Yes ☐
- Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

## Spleen

**Enlarged or normal from scan?**

- Enlarged ☐
- Normal ☐
- Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

**Size or volume available from scan?**

- No ☐
- Yes ☐
- Not recorded ☐

Spleen organ size (cm) _____

**And/Or**

Spleen volume: ____ ml

Measure date: __ __ / __ __ / __ __ __ __

**Enlarged on palpation**

- No ☐
- Yes ☐
- Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

# Ophthalmology – Child

**Visual Acuity**

Using any age-appropriate test, eyesight is: Normal ☐ Abnormal ☐

Visual Acuity Test (Snellen eye chart)

Done ☐ Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Eye ____ / ____ Right Eye ____ / ____

The first number given is the distance in metres from the chart. Usually this is a 6 (for 6 meters).
The second number could be 60, 36, 24, 18, 12, 9, 6 or 5

**Corneal Clouding**

Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Eye No ☐ Yes ☐ Right Eye No ☐ Yes ☐

**Intra Ocular Pressure**

Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Eye _____ mmHg Right Eye _____ mmHg

# Other Measures

**Urine GAG _____** mg/mmol of creatine

Measure date: __ __ / __ __ / __ __ __ __

# Bone Marrow Transplant

Done ☐
Not done ☐

Date of first transplant: __ __ / __ __ / __ __ __ __

Type of transplant
☐ sib
☐ MUD
☐ UCB
☐ Other
Please Specify_____

First transplant rejection
No ☐
Yes ☐
Rejection date: __ __ / __ __ / __ __ __ __

ERT used before transplant? No ☐
Yes ☐

Duration: ____ mths ____ yrs

---

Date of second transplant: __ __ / __ __ / __ __ __ __

Type of transplant
☐ sib
☐ MUD
☐ UCB
☐ Other
Please Specify_____

Second transplant rejection
No ☐
Yes ☐
Rejection date: __ __ / __ __ / __ __ __ __

---

Date of third transplant: __ __ / __ __ / __ __ __ __

Type of transplant
☐ sib
☐ MUD
☐ UCB
☐ Other
Please Specify_____

Third transplant rejection
No ☐
Yes ☐
Rejection date: __ __ / __ __ / __ __ __ __

Transplant complications:
☐ acute GvHD
☐ Chronic GvHD

☐ Viral reactivation
☐ VOD
☐ Pulmonary haemorrhage
☐ Other
Please Specify_____

Enzyme activity at 12 months post BMT _____
Unit: ☐ milli units / unit of hexosaminidase at 37$^{o}$C
☐ µmol/min/mg of protein

% donor chimerism at 12 months post BMT _____ %

# Heart Measures - Adult

Echocardiogram: Done Yes ☐
No ☐ Measure date: __ __ / __ __ / __ __ __ __

Valve Disease as reported by cardiologist (on echo)
None ☐
Mild ☐
Moderate ☐
Severe ☐

Valve replacements
No ☐
Yes ☐

Date of valve replacement: __ __ / __ __ / __ __ __ __

# Airway / Lung / ENT – Adult

**Ventilation**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

The patient:
☐ is free of ventilation
☐ is nocturnally ventilated only
☐ has intermittent daytime ventilation
☐ is ventilator-dependent (continuous) (drop down menu)

**Average total number of hours on ventilator per 24 hours _____**

**Sleep Study**
Done ☐
Not done ☐

Measure date: __ __ / __ __ / __ __ __ __

Time spent below saturation of 90% ____
(Given as a percentage of time of duration of the study)

Lowest saturation recorded (%) ____
(the value in percent of lowest recording that was not due to an artifact)

**Pulmonary Function Test (best of 3 if there is more than one)**

Patient upright
Test done ☐
Test not done ☐
Can't do test ☐

Measure date: __ __ / __ __ / __ __ __ __

FVC (%) _____ percentage of predicted

FEV1 (%) _____ percentage of predicted

**Has patient had surgical ENT intervention?**
No ☐
Yes ☐
please specify _____

Date of surgical intervention: __ __ / __ __ / __ __ __ __

**Audiometry**
**Hearing Test:**
Done ☐
Not done ☐

Hearing Normal
No ☐
Yes ☐

Measure date: __ __ / __ __ / __ __ __ __

| **Type of hearing loss (pick one)** | | **Degree of hearing loss (average decibels over tested frequencies) (pick one)** | |
|---|---|---|---|
| Conductive | ☐ | Mild (0-40 decibels) | ☐ |
| Sensorineural | ☐ | Moderate (41-70 decibels) | ☐ |
| Mixed (combination of conductive & sensorineural | ☐ | Severe (71-95 decibels) | ☐ |
| | | Profound (over 95 decibels) | ☐ |

Note: to calculate the degree of hearing loss, add all the decibels and divide by the number of data points.

| | | | |
|---|---|---|---|
| Hearing Aid Used | No | ☐ | |
| | Yes | ☐ | Type of hearing aid_____ |
| | Not Recorded | ☐ | |

# Neurological – Adult

**Neurological involvement**

No ☐
Yes ☐
Not Recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

**Carpal Tunnel Syndrome**

Present ☐
Absent ☐
Not Recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Has patient had surgery for carpal tunnel syndrome?

No ☐
Yes ☐ How many times? ____

Date of last Carpal Surgery: __ __ / __ __ / __ __ __ __

**Cervical Cord Compression**

Present ☐
Absent ☐
Not Recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Has patient had surgery for cervical cord compression?

No ☐
Yes ☐
Date of last Cervical Cord Compression Surgery: __ __ / __ __ / __ __ __ __

# Bone & Joint – Adult

**Patient Mobility**

Measure date: __ __ / __ __ / __ __ __ __

☐ can walk / stand unaided (i.e. can stand for 6 mins or walk for 5 meters)
☐ can walk aided one stick (i.e. can't stand for 6 mins without a stick)
☐ can walk aided more than one stick
☐ is wheelchair bound
☐ is bed-bound (i.e. can't get into wheelchair)

**6 Minute Walk Test**

Done ☐
Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

Distance walked (m) _____ Aided ☐
Unaided ☐

**Muscle Testing – limited MRC scale – 0-5, whole numbers only**

Done ☐
Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

| **Muscle Test** | **Left** | **Right** |
|---|---|---|
| Upper limb: | | |
| Shoulder abduction | | |
| Shoulder adduction | | |
| Elbow flexion | | |
| Elbow extension | | |
| Wrist flexion | | |
| Wrist extension | | |
| Lower limb: | | |
| Hip flexion | | |
| Hip extension | | |
| Knee flexion | | |
| Knee extension | | |
| Plantar flexion | | |
| Dorsi flexion | | |
| TOTAL | | |

Muscle Test Score _____ (maximum score 120)

The patient was: Passive ☐
Active ☐ during muscle testing

# Ophthalmology – Adult

**Visual Acuity** (Snellen eye chart)
Done ☐
Note Done ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Eye ____ / ____ Right Eye ____ / ____

The first number given is the distance in metres from the chart. Usually this is a 6 (for 6m).
The second number could be 60, 36, 24, 18, 12, 9, 6 or 5

**Corneal Clouding**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Eye No ☐ Yes ☐ Right Eye No ☐ Yes ☐

**Intra Ocular Pressure**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Eye _____ mmHg Right Eye _____ mmHg

**Retinal Disease – Adult**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Eye No ☐ Yes ☐ Right Eye No ☐ Yes ☐

# Significant Co-morbidity

**Does the patient have any co-morbidities? Yes / No**

**How many? ____**

Please enter significant co-morbidities in the spaces below, and then answer yes or no to the organs affected

1

2

3

4

5

6

| **1** | **2** | **3** | **4** | **5** | **6** |
|---|---|---|---|---|---|
| **Heart** | | | | | |
| Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No |
| Lungs/respiratory | | | | | |
| Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No |
| Neurological | | | | | |
| Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No |
| Skeletal/muscle | | | | | |
| Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No |
| Opthamology | | | | | |
| Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No |

**Children Only:**

| | | | | | |
|---|---|---|---|---|---|
| Liver and spleen | | | | | |
| Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No |
| Growth | | | | | |
| Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No |

# Con-meds

**Is the patient on any con meds?**

| Medication Prescription? | Dose | Frequency | Date started | Date finished | Ongoing? |
|---|---|---|---|---|---|

# Other Measures - Adult

**Urine GAG** _____ mg/mmol of creatine

Measure date: __ __ / __ __ / __ __ __ __

# Record of Therapy (Enzyme Replacement Therapy, etc.):

Is patient on treatment No ☐
Yes ☐

**Initial Treatment**
Initially on ERT ☐
clinical trial? ☐

Date of first Infusion: __ __ / __ __ / __ __ __ __

Age when first infusion received: _____

Initial type of treatment: _____

Initial dose (International Units): ___(mg)

Initial frequency: weekly ☐
every 2 weeks ☐
Other ☐ Please Specify_____

**Current treatment**

Currently on ERT or clinical trial? ☐

Date started current treatment: __ __ / __ __ / __ __ __ __

Current type of treatment: _____

Current dose (International Units): ___mg

Current frequency: weekly ☐
every 2 weeks ☐
Other ☐ Please Specify_____

Hospital/ home infusion: Infusion in Hospital ☐
Infusion at Home ☐ If Home: pick list:
Nurse infuses ☐
Nurse cannulates and leaves ☐
Patient cannulates and infuses ☐

Is the patient currently experiencing:

Febrile reactions? No ☐
Yes ☐

Anaphylactoid reactions? No ☐
Yes ☐

Other pre-medication: No ☐
Yes ☐

**Antibody status**

Measure date: __ __ / __ __ / __ __ __ __

Antibody status to infused product, Tested No ☐
Yes ☐

Antibody status Test Result Positive No ☐
Yes ☐
Don't know ☐

# Record of Therapy (Enzyme Replacement Therapy, etc):

Is patient on treatment No ☐
Yes ☐

Has patient stopped treatment since their last visit? No ☐
Yes ☐

Has patient started treatment since their last visit? No ☐
Yes ☐

**Initial Treatment**
Initially on ERT or clinical trial? _____

Date of first Infusion: __ __ / __ __ / __ __ __ __

Age when first infusion received: _____

Initial type of treatment: _____

Initial dose (International Units): ___mg

Initial frequency: weekly ☐
every 2 weeks ☐
Other ☐ Please Specify_____

**Current treatment**

Currently on ERT or clinical trial? _____

Date started current treatment: __ __ / __ __ / __ __ __ __

Current type of treatment _____

Current Dose (International Units): ____ mg

Current frequency: weekly ☐
every 2 weeks ☐
Other ☐ Please Specify_____

Hospital/ home infusion: Infusion in Hospital ☐
Infusion at Home ☐ If Home: pick list:
Nurse infuses ☐
Nurse cannulates and leaves ☐
Patient cannulates and infuses ☐

Is the patient currently experiencing:

Febrile reactions? No ☐
Yes ☐

Anaphylactoid reactions? No ☐
Yes ☐

Other pre-medication: No ☐
Yes ☐

**Antibody status**

Measure date: __ __ / __ __ / __ __ __ __

Antibody status to infused product, Tested No ☐
Yes ☐

Antibody Status Test Result Positive No ☐
Yes ☐

**Stopped Treatment**
Final treatment on ERT or clinical trial (drop down menu)

Date of last infusion: __ __ / __ __ / __ __ __ __

Final type of treatment _____

Final dose (International Units): ___mg

Final frequency: weekly ☐
every 2 weeks ☐
Other ☐ Please Specify_____

Hospital/ home infusion: Infusion in Hospital ☐
Infusion at Home ☐ If Home: pick list:
Nurse infuses ☐
Nurse cannulates and leaves ☐
Patient cannulates and infuses ☐

Why was treatment stopped?_____

Please specify: _____

Visit date: _____ current age: _____

# Appendix 11d Clinical Record Form for MPS II patients

MPS II Patient ID:

Date of visit ___/ ___ / _____

# MPS II CRFs

## Inclusion Criteria

Has Clinician given consent for the patient or their carer to be approached?

No ☐ Do not continue
Yes ☐

Has the patient given informed signed consent?

No ☐ Do not continue
Yes ☐

Has the parent/carer given informed signed consent?

No ☐ Do not continue
Yes ☐

Reason for not giving consent (if given):

_____

The patient is consented for:

Records Only ☐
Records and Questionnaires ☐

Hospital ID _____

Patient Identification number _____

Date of consent __ __ / __ __ / __ __ __ __

Version of consent form: _____

# Patient Information

**Date of birth** __ __ / __ __ / __ __ __ __

**Patient age, in years, at consent** _____

**Gender** Male ☐
Female ☐

**Type of MPS II**
Without neurological involvement ☐
With neurological involvement ☐

**Does the patient have a carer?** No ☐
Yes ☐

**Onset of first symptoms** Roughly / exactly / can't remember: ___ / ____

**Any other family members affected?**

No ☐
Yes ☐ If so, how many? __

Relationship to patient:

Mother ☐
Father ☐
Sibling ☐
Other ☐ Please Specify _____
(enter Unique ID no.s of relations)

**Date of diagnosis:** month and year: ___ / ____

**Method of diagnosis**:

☐ Recorded
☐ Not recorded

Iduuronate-2-sulfatase activity ☐ Normal
☐ Deficient

Iduuronate-2-sulfatase cDNA ☐ Done
☐ Not Done

cDNA result as amino acid change _____

Skin Biopsy – Enzyme Assay ☐ Normal
☐ Abnormal If abnormal, result: _____

Prenatal Diagnosis (chorion villus biopsy) ☐ No ☐ Yes

Enzyme Assay Results _____

Urine GAG test ☐ No ☐ Yes

Urine GAG test Results _____ (mg/mmol of creatine)

**Reason for diagnosis**

**Was patient detected by family screening?** ☐ No ☐ Yes

**Did the patient present with symptoms?** ☐ No ☐ Yes

If so, what? _____

**Current Employment Status**

| | | | |
|---|---|---|---|
| Paid Employed | ☐ | If employed | |
| Unemployed | ☐ | Full time | ☐ |
| Unpaid Employment | ☐ | Part time | ☐ |

# Body Measurements – Adults & Child

Measure date: __ __ / __ __ / __ __ __ __

Height (cm) _____

Weight (kg) _____

**Children only:**

Head Circumference (cm) _____

# Heart Measures - Adult

Echocardiogram: Done Yes ☐
No ☐ Measure date: __ __ / __ __ / __ __ __ __

Valve Disease as reported by cardiologist (on echo)
None ☐
Mild ☐
Moderate ☐
Severe ☐

Valve replacements
No ☐
Yes ☐

Date of replacement: __ __ / __ __ / __ __ __ __

# Airway / Lung / ENT – Adult

**Ventilation** Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

The patient:
☐ is free of ventilation
☐ is nocturnally ventilated only
☐ has intermittent daytime ventilation
☐ is ventilator-dependent (continuous)

Average number of hours on ventilator per 24 hours _____

**Sleep Study** Done ☐
Not done ☐

Measure date: __ __ / __ __ / __ __ __ __

Time spent below saturation of 90% ____
(Given as a percentage of time of duration of the study)

Lowest saturation recorded (%) ____
(the value in percent of time of duration of the study)

**Pulmonary Function Test (best of 3 if there is more than one)**

Patient upright
Test Done ☐
Test not done ☐
Can't do test ☐

Measure date: __ __ / __ __ / __ __ __ __

FVC (%) _____ percentage of predicted

FEV1 (%) _____ percentage of predicted

**Has patient had surgical ENT intervention?** No ☐
Yes ☐

Please specify _____

Date of surgical intervention: __ __ / __ __ / __ __ __ __

**Audiometry**

Hearing Test: Done ☐
Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

Hearing Normal: No ☐
Yes ☐

| **Type of hearing loss (pick one)** | | **Degree of hearing loss (average decibels over tested frequencies) (pick one)** | |
|---|---|---|---|
| Conductive | ☐ | Mild (0-40 decibels) | ☐ |
| Sensorineural | ☐ | Moderate (41-70 decibels) | ☐ |
| Mixed (combination of conductive & sensorineural | ☐ | Severe (71-95 decibels) | ☐ |
| | | Profound (over 95 decibels) | ☐ |

Note: to calculate the degree of hearing loss, add all the decibels and divide by the number of data points.

Hearing Aid Used No ☐
Yes ☐ Type of hearing aid_____
Not Recorded ☐

# Neurological – Adult

**Neurological involvement**

No ☐
Yes ☐
Not Recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

**Carpal Tunnel Syndrome**

Present ☐
Absent ☐
Not Recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Has patient had surgery for carpal tunnel syndrome?

No ☐
Yes ☐ How many times? ____

Date of last surgery: __ __ / __ __ / __ __

**Cervical Cord Compression**

Present ☐
Absent ☐
Not Recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Has patient had surgery for cervical cord compression?

No ☐
Yes ☐

Date of last surgery: __ __ / __ __ / __ __ __ __

# Bone & Joint – Adult

**Patient Mobility**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

☐ can walk / stand unaided (i.e. can stand for 6 mins or walk for 5 meters)
☐ can walk aided one stick (i.e. can't stand for 6 mins without a stick)
☐ can walk aided more than one stick
☐ is wheelchair bound
☐ is bed-bound (i.e. can't get into wheelchair)

**6 Minute Walk Test**

Done ☐
Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

Distance walked (m) _____ Aided ☐
Unaided ☐

# Ophthalmology – Adult

**Visual Acuity** (Snellen eye chart)
Done ☐
Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Eye ____ / ____ Right Eye ____ / ____

The first number given is the distance in metres from the chart. Usually this is 6m. The second number could be 60, 36, 24, 18, 12, 9, 6 or 5

**Corneal Clouding**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Eye No ☐ Right Eye No ☐
Yes ☐ Yes ☐

**Intra Ocular Pressure**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Eye _____ mmHg Right Eye _____ mmHg

**Retinal Disease**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Eye No ☐ Right Eye No ☐
Yes ☐ Yes ☐

# Heart Measures - Child

Echocardiogram: Done ☐
Not done ☐ Measure date: __ __ / __ __ / __ __ __ __

Left Ventricular Ejection fraction <60%
No ☐
Yes ☐

Valve Disease as reported by cardiologist (on echo) None ☐
Mild ☐
Moderate ☐
Severe ☐

Valve Disease severe enough to require medication
No ☐
Yes ☐

Valve replacements
No ☐
Yes ☐

Date of replacements: __ __ / __ __ / __ __ __ __

# Airway / Lung / ENT - Child

**Sleep Study** Done ☐
Not done ☐

Measure date: __ __ / __ __ / __ __ __ __

Time spent below saturation of 90% ____
(Given as a percentage of time of duration of the study)

Lowest saturation recorded (%) ____
(the value in percent of time of duration of the study)

**Pulmonary Function Test (best of 3 if there is more than one)**

Patient upright Test Done ☐
Test not done ☐
Can't do test ☐

Measure date: __ __ / __ __ / __ __ __ __

FVC (%) _____ percentage of predicted

FEV1 (%) _____ percentage of predicted

**Audiometry**
Using any age-appropriate audiometry test, hearing is: Normal ☐
Abnormal ☐
Not done ☐

Measure date: __ __ / __ __ / __ __ __ __

| **Type of hearing loss (pick one)** | | **Degree of hearing loss (average decibels over tested frequencies) (pick one)** | |
|---|---|---|---|
| Conductive | ☐ | Mild (0-40 decibels) | ☐ |
| Sensorineural | ☐ | Moderate (41-70 decibels) | ☐ |
| Mixed (combination of conductive & sensorineural | ☐ | Severe (71-95 decibels) | ☐ |
| | | Profound (over 95 decibels) | ☐ |

Note: to calculate the degree of hearing loss, add all the decibels and divide by the number of data points.

**Difficulty with intubation for anaesthesia?** ☐ No
☐ Yes
☐ No operation carried out

Date of operation: __ __ / __ __ / __ __ __ __

# Neurological – Child

**DQ Test:**

Done ☐
Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

DQ Score _____

**IQ Test:**

Done ☐
Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

IQ Score _____

**Hydrocephalus**

Present ☐
Absent ☐
Not Recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

**Carpal Tunnel Syndrome**

Present ☐
Absent ☐
Not Recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

# Bone & Joint – Child

**Patient Mobility**

Test: Applicable ☐
Not Applicable due to age ☐

Measure date: __ __ / __ __ / __ __ __ __

☐ can walk / stand unaided (i.e. can stand for 6 mins or walk for 5 meters)
☐ can walk aided one stick (i.e. can't stand for 6 mins without a stick)
☐ can walk aided more than one stick
☐ is wheelchair bound
☐ is bed-bound (i.e. can't get into wheelchair)

**6 Minute Walk Test**

Done ☐
Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

Distance walked (m) _____ Aided ☐
Unaided ☐

# Liver & Spleen – Child

## Liver

**Enlarged or normal from scan?**

- Enlarged ☐
- Normal ☐
- Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

**Size or volume available from scan?**

- No ☐
- Yes ☐
- Not recorded ☐

Liver organ size (cm) _____

**And/Or**

Liver volume: ____ ml

Measure date: __ __ / __ __ / __ __ __ __

**Enlarged on palpation**

- No ☐
- Yes ☐
- Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

## Spleen

**Enlarged or normal from scan?**

- Enlarged ☐
- Normal ☐
- Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

**Size or volume available from scan?**

- No ☐
- Yes ☐
- Not recorded ☐

Spleen organ size (cm) _____

**And/ Or**

Spleen volume: ____ ml

Measure date: __ __ / __ __ / __ __ __ __

**Enlarged on palpation**

- No ☐
- Yes ☐
- Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

# Ophthalmology – Child

**Visual Acuity**

Using any age-appropriate test, eyesight is: Normal ☐ Abnormal ☐

Visual Acuity Test (as tested by an appropriate test for age and DQ)

Done ☐

Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Eye ____ / ____ Right Eye ____ / ____

The first number given is the distance in metres from the chart. Usually this is 6m. The second number could be 60, 36, 24, 18, 12, 9, 6 or 5

**Corneal Clouding**

Recorded ☐

Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Eye No ☐ Yes ☐ Right Eye No ☐ Yes ☐

**Intra Ocular Pressure**

Recorded ☐

Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Eye _____ mmHg Right Eye _____ mmHg

**Optic disc swelling**

Recorded ☐

Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Eye No ☐ Yes ☐ Right Eye No ☐ Yes ☐

**Retinopathy**

**Optic disc swelling**

Recorded ☐

Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Eye No ☐ Yes ☐ Right Eye No ☐ Yes ☐

# Other Measures – Adult & Child

**Urine GAG** _____ mg/mmol of creatine

Measure date: __ __ / __ __ / __ __ __ __

# Bone Marrow Transplant - Child

Done ☐
Not done ☐

Date of first transplant: __ __ / __ __ / __ __ __ __

Type of first transplant
☐ sib
☐ MUD
☐ UCB
☐ Other Specify_____

First transplant rejection
No ☐
Yes ☐ Date of rejection: __ __ / __ __ / __ __ __ __

ERT used before transplant?
No ☐
Yes ☐

If Yes: Duration: ____ mths ____ yrs

---

Date of second transplant: __ __ / __ __ / __ __ __ __

Type of transplant
☐ sib
☐ MUD
☐ UCB
☐ Other Specify_____

Rejection
No ☐
Yes ☐ Date of rejection: __ __ / __ __ / __ __ __ __

---

Date of third transplant and type if needed: __ __ / __ __ / __ __ __ __

☐ sib
☐ MUD
☐ UCB
☐ Other Specify_____

Rejection
No ☐
Yes ☐ Date of rejection: __ __ / __ __ / __ __ __ __

---

Transplant complications:

☐ acute GvHD
☐ Chronic GvHD
☐ Viral reactivation
☐ VOD
☐ Pulmonary haemorrhage
☐ Other Please Specify_____

Enzyme activity at 12 months post BMT _____
Unit:
☐ milli units / unit of hexosaminidase at 37$^{o}$C
☐ µmol/min/mg of protein

% donor chimerism at 12 months post BMT _____ %

# Significant Co-morbidity

**Does the patient have any co-morbidities? Yes / No**

**How many? ____**

Please enter significant co-morbidities in the spaces below, and then answer yes or no to the organs affected

1

2

3

4

5

6

| **1** | **2** | **3** | **4** | **5** | **6** |
|---|---|---|---|---|---|
| **Heart** | | | | | |
| Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No |
| Airway | | | | | |
| Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No |
| Neurological | | | | | |
| Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No |
| Skeletal/muscle | | | | | |
| Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No |
| Opthamology | | | | | |
| Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No |

**Children Only:**

| | | | | | |
|---|---|---|---|---|---|
| Liver and spleen | | | | | |
| Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No |
| Growth | | | | | |
| Yes / No | Yes / No | Yes / No | Yes / No | Yes / No | Yes / No |

# Con-meds

| Medication Prescription? | Dose | Frequency | Date started | Date finished | Ongoing? |
|---|---|---|---|---|---|

# Record of Therapy
**(Enzyme Replacement Therapy, substrate reduction therapy etc):**

Is patient on treatment No ☐
Yes ☐

**Initial Treatment**
Initially on ERT ☐
clinical trial? ☐

Date of first treatment: __ __ / __ __ / __ __ __ __

Age when first infusion received: _____

Initial type of treatment: _____

Initial dose: ___(mg/kg)

Initial frequency:
3 x each week ☐
2 x each week ☐
Weekly ☐
Every 2 weeks ☐
Every 3 weeks ☐
Monthly ☐

**Current treatment**

Currently on ERT ☐
clinical trial? ☐

Date started on current treatment __ __ / __ __ / __ __ __ __

Current type of treatment

Current dose: ___mg/kg

Current frequency:
3 x each week ☐
2 x each week ☐
Weekly ☐
Every 2 weeks ☐
Every 3 weeks ☐
Monthly ☐

Hospital/ home infusion: Infusion in Hospital ☐
Infusion at Home ☐ If Home: pick list:
Nurse infuses ☐
Nurse cannulates and infuses ☐
Patient cannulates and infuses ☐
Relative cannulates and infuses ☐

Is the patient currently experiencing:

Febrile reactions? No ☐
Yes ☐

Analphylactoid reactions? No ☐
Yes ☐

Other pre-medication: No ☐
Yes ☐

**Antibody status**

Measure date: __ __ / __ __ / __ __ __ __

Antibody status to infused product, Tested No ☐
Yes ☐ If Yes, Positive No ☐
Yes ☐
Don't know ☐

# Withdrawal from the study

**Is the patient still in the full study?** No ☐
Yes ☐

Withdrawal date: __ __ / __ __ / __ __ __ __

Reason for Withdrawal

☐ Patient has life threatening disease
☐ Failure to comply (due to medication)
☐ Evidence of disease progression
☐ Patient is pregnant
☐ Death
☐ Patient is on a clinical trial
☐ Questionnaire burden
☐ Patient turned 16 and didn't reconsent
☐ Other Please specify_____

Continue in the study notes only? No ☐
Yes ☐

**Record of death**

Date of death: __ __ / __ __ / __ __ __ __

Did clinician certify this death as condition related? No ☐
Yes ☐

Please record the wording from the dearth certificate:

Part 1a ---------- Free text (Disease or condition directly leading to death)

Part 1b ---------- Free text (Other disease or condition, if any, leading to 1a)

Part 1c ---------- Free text (Other disease or condition, if any, leading to 1b)

Part 2 ---------- Free text (Other significant conditions CONTRIBUTING TO THE DEATH but not related to the disease or condition causing it)

# Appendix 11e Clinical Record From for Pompe patients

Pompe Patient ID:

Date of visit ___/ ___ / _____

## Pompe CRFs

## Inclusion Criteria

Has Clinician given consent for the patient or their carer to be approached?

No ☐ Do not continue
Yes ☐

Has the patient given informed signed consent?

No ☐ Do not continue
Yes ☐

Has the parent / carer given informed signed assent?

No ☐ Do not continue
Yes ☐

Reason for not giving consent:

_____

The patient consented for ☐ Records Only
☐ Records and Questionnaires

Patient Identification number _____

Date of consent __ __ / __ __ / __ __ __ __

Version of consent form: _____

# General Patient Information for non-cardiomyopathy and cardiomyopathy

Date of birth __ __ / __ __ / __ __ __ __

Gender:
Male ☐
Female ☐

Type of Pompe
Non-cardiomyopathy ☐
Cardiomyopathy ☐

Does the patient have a carer?
No ☐
Yes ☐

Onset of first symptoms is:
Roughly ☐
Exactly ☐
Can't remember ☐ Month ___ Year ____

Other family members affected?
No ☐
Yes ☐ If Yes how many --
Relationship/s to patient
Mother ☐
Father ☐
Sibling ☐
Other ☐ Please specify_____
(enter unique ID no of relations)

Month & Year of diagnosis
Month--
Unknown ☐ If Month unknown: Year _____

**Method of diagnosis (tick one of more below if done)**

☐ Recorded
☐ Not recorded

**GAA activity:** Normal ☐ (also known as: acid maltose, acid-alpha glucosidase)
Deficient ☐

**GAA cDNA** Not Done ☐
Done ☐ results:

Allele 1 _____

Allele 2 _____

**Muscle biopsy** ☐ result: negative ☐
positive ☐ (=glycogen in vacuoles or staining for enzyme acid phosphotase, depending on test)

**Employment Status**
Paid Employed ☐ If employed
Unemployed ☐ Full time ☐
Unpaid Employment ☐ Part time ☐

## Reason for diagnosis:

## Non cardiomyopathy – adult

Was the patient detected by family screening
No ☐
Yes ☐

Did the patient present with symptoms
No ☐
Yes ☐ If Yes did the patient have:

Proximal Myopathy
No ☐
Yes ☐ pick one:
Upper Limb ☐
Lower Limb ☐
Both ☐

Breathing Difficulties
No ☐
Yes ☐

Other symptoms
No ☐
Yes ☐

Please specify: _____

## Reason for diagnosis:
## Non cardiomyopathy – children (juvenile)

Was the patient detected by family screening? No ☐
Yes ☐

Did the patient present with symptoms? No ☐
Yes ☐ If Yes did the patient have:

Abnormal muscle weakness No ☐
Yes ☐

Poor Growth No ☐
Yes ☐

Recurrent chest infections No ☐
Yes ☐

Other symptoms ☐ Please specify:_____-___

## Reason for diagnosis:
## Cardiomyopathy (infantile)

With which of the following symptoms did the patient present: Failure to thrive No ☐
Yes ☐

Muscle weakness No ☐
Yes ☐

Cardiomyopathy No ☐
Yes ☐

# Withdrawal from the study

**Is the patient still in the full study?** No ☐
Yes ☐

Withdrawal date: __ __ / __ __ / __ __ __ __

Reason for Withdrawal

☐ Patient has life threatening disease
☐ Failure to comply (due to medication)
☐ Evidence of disease progression
☐ Patient is pregnant
☐ Death
☐ Patient is on a clinical trial
☐ Questionnaire burden
☐ Patient turned 16 and didn't reconsent
☐ Other Please specify_____

Continue in the study notes only? No ☐
Yes ☐

**Record of death**

Date of death: __ __ / __ __ / __ __ __ __

Did clinician certify this death as condition related? No ☐
Yes ☐

Please record the wording from the dearth certificate:

Part 1a ---------- Free text (Disease or condition directly leading to death)

Part 1b ---------- Free text (Other disease or condition, if any, leading to 1a)

Part 1c ---------- Free text (Other disease or condition, if any, leading to 1b)

Part 2 ---------- Free text (Other significant conditions CONTRIBUTING TO THE DEATH but not related to the disease or condition causing it)

# Bone & Joint

**Patient Mobility**

Test: Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

☐ can walk / stand unaided (i.e. can stand for 6 mins or walk for 5 meters)
☐ can walk aided one stick (i.e. can't stand for 6 mins without a stick)
☐ can walk aided more than one stick
☐ is wheelchair bound
☐ is bed-bound (i.e. can't get into wheelchair)

**6 Minute Walk Test**

Done ☐
Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

Distance walked (m) _____ Aided ☐
Unaided ☐

**Timed Gower's test for children**

Done ☐
Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

Positive ☐
Negative ☐
Gave up / can't do ☐

Seconds to complete _____

**Muscle Testing – limited MRC scale – 0-5, whole numbers only**

Done ☐
Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

| Muscle Test | Left | Right |
|---|---|---|
| Upper limb: | | |
| Shoulder abduction | | |
| Shoulder adduction | | |
| Elbow flexion | | |
| Elbow extension | | |
| Wrist flexion | | |
| Wrist extension | | |
| Lower limb: | | |
| Hip flexion | | |
| Hip extension | | |
| Knee flexion | | |
| Knee extension | | |
| Plantar flexion | | |
| Dorsi flexion | | |
| TOTAL | | |

Muscle Test Score _____ (maximum score 120)

The patient was: Passive ☐
Active ☐ during muscle testing

**Dynanometry Test**

Done ☐
Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

Grip Strength (kg) _____ (best of three)

# Respiratory

**Ventilation**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

The patient:
☐ is free of ventilation
☐ is nocturnally ventilated only
☐ has intermittent daytime ventilation
☐ is ventilator-dependent (continuous)

Average number of hours on ventilator per 24 hours _____

**Pulmonary Function Test (best of 3 if there is more than one)**

**Patient lying down**
Test Done ☐
Test not done ☐
Can't do test ☐

Measure date: __ __ / __ __ / __ __ __ __

FVC (%) _____ percentage of predicted

FEV1 (%) _____ percentage of predicted

**Patient upright**
Test Done ☐
Test not done ☐
Can't do test ☐

Measure date: __ __ / __ __ / __ __ __ __

FVC (%) _____ percentage of predicted

FEV1 (%) _____ percentage of predicted

**Swallowing difficulties**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Does the patient have swallowing difficulties – No ☐ Yes ☐

If Yes has patient had to modify diet in any way (from advice from Speech therapist) No ☐ Yes ☐

**Sleep study** (Children under 8)
Done ☐
Not Done ☐ If Done:

Measure date: __ __ / __ __ / __ __ __ __

Time spent below saturation of 90% _____%
(Given as a percentage of time of duration of the study)

Lowest saturation recorded _____%
(the value in percent of lowest recording that was not due to an artifact)

# Other measures

**Body Measurements – Height**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Height (m) _____

**Body Measurements – Weight**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Weight (Kg) _____

**Vitamin D**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Does the patient have clinically significant Vitamin D deficiency
Yes ☐
No ☐
Not tested ☐

# Heart

**Echocardiogram:**

Done ☐
Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

Left Ventricular Mass index _____$g/m^2$

Left Ventricular Ejection fraction _____%

Fraction Shortening _____%

Arrhythmias present ☐
absent ☐

# Bone & Joint – Children

Using any parent reported Motor milestones scale, is the patient able to do the following?
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Sit unsupported: Able ☐
Unable ☐ Months _____

Stand independently: Able ☐
Unable ☐ Months _____

Walk: Able ☐
Unable ☐ Months _____

Walk upstairs one step at a time:
Able ☐
Unable ☐ Months _____

**Muscle strength - Gross Motor Functional Measure (GMFM-66)**

Not done ☐
Done ☐

Measure date: __ __ / __ __ / __ __ __ __

Score: _____%

**Fractures**

Recorded ☐
Not Recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

No of fractures in the last 6 months: _____

# Airway / Lung / ENT

**Swallowing difficulties**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Does the patient feed orally? No ☐
Yes ☐

Can patient swallow liquid?

Can patient swallow pureed food?

Can patient swallow solid food?

**Ventilation**
Recorded ☐
Not recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

The patient:
☐ is free of ventilation
☐ is nocturnally ventilated only
☐ has intermittent daytime ventilation
☐ is ventilator-dependent (continuous)

Average number of hours on ventilator per 24 hours _____

**Sleep study**
Not Done ☐
Done ☐ If Done:

Measure date: __ __ / __ __ / __ __ __ __

Time spent below saturation of 90% _____%
(Given as a percentage of time of duration of the study)

Lowest saturation recorded _____%
(the value in percent of lowest recording that was not due to an artifact)

**Audiometry**

Hearing Test done: Yes ☐
No ☐

Measure date: __ __ / __ __ / __ __ __ __

Hearing Normal: Yes ☐
No ☐ If No….

| **Type of hearing loss (pick one)** | | **Degree of hearing loss (average decibels over tested frequencies) (pick one)** | |
|---|---|---|---|
| Conductive | ☐ | Mild (0-40 decibels) | ☐ |
| Sensorineural | ☐ | Moderate (41-70 decibels) | ☐ |
| Mixed (combination of conductive & sensorineural | ☐ | Severe (71-95 decibels) | ☐ |
| | | Profound (over 95 decibels) | ☐ |

Note: to calculate the degree of hearing loss, add all the decibels and divide by the number of data points.

Type of hearing aid: _____

**Pulmonary Function Test (best of 3 if there is more than one)**

**Patient lying down**

Test Done ☐
Test not done ☐
Can't do test ☐

Measure date: __ __ / __ __ / __ __ __ __

FVC (%) _____ percentage of predicted

FEV1 (%) _____ percentage of predicted

**Patient upright**

Test Done ☐
Test not done ☐
Can't do test ☐

FVC (%) _____ percentage of predicted

FEV1 (%) _____ percentage of predicted

# Growth & Diet

**Feeding**

Recorded ☐
Not Recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

High protein diet No ☐
Yes ☐

**Body measurements**

Recorded ☐
Not Recorded ☐

Measure date: __ __ / __ __ / __ __ __ __

Height cm _____

Weight Kg _____

Head circumference (cm) _____

# Significant Co-morbidity

Please enter up to three significant co-morbidities in the spaces below, and then answer yes or no to the organs affected

1

2

3

4

5

6

| **Non-cardiomyopathy** | **1** | **2** | **3** |
|---|---|---|---|
| Muscle | Yes / No | Yes / No | Yes / No |
| Lungs / respiratory | Yes / No | Yes / No | Yes / No |
| Growth | Yes / No | Yes / No | Yes / No |
| **Cardiomyopathy** | | | |
| Heart | Yes / No | Yes / No | Yes / No |
| Skeletal / muscle | Yes / No | Yes / No | Yes / No |
| Respiration | Yes / No | Yes / No | Yes / No |
| Growth | Yes / No | Yes / No | Yes / No |

# Con-meds

| Medication | Prescription? | Dose | Frequency | Date started | Date finished | Ongoing? |
|---|---|---|---|---|---|---|

# Record of Therapy (Enzyme Replacement Therapy, substrate reduction therapy etc):

Is patient on ERT No ☐
Yes ☐

**Initial Treatment**
Initially on ERT ☐
clinical trial? ☐

Date of first infusion:

Age when first infusion received:

Initial type of treatment: Myozyme ☐

Initial dose: ___mg

Initial frequency: weekly ☐
every 2 weeks ☐

**Current treatment**
Is patient currently on ERT ☐
clinical trial ☐

Date started current treatment? --/--/---

Current type of treatment

Current dose: ___mg

Current frequency: weekly ☐
every 2 weeks ☐

Hospital/ home infusion: Infusion in Hospital ☐
Infusion at Home ☐ If Home: pick list: Nurse infuses ☐
Nurse cannulates and leaves ☐
Patient cannulates and infuses ☐

Is the patient currently experiencing:

Febrile reactions? No ☐
Yes ☐

Analphylactoid reactions? No ☐
Yes ☐

Does the patient require any pre-medication: No ☐
Yes ☐

**Antibody status**

Measure date: __ __ / __ __ / __ __ __ __

Antibody status to infused product, Tested No ☐
Yes ☐ If Yes, Positive No ☐
Yes ☐
Don't know ☐

# Record of Therapy (Enzyme Replacement Therapy, substrate reduction therapy etc):

Is patient on ERT
No ☐
Yes ☐

Has patient stopped treatment since their last visit?
No ☐
Yes ☐

Has patient started treatment since their last visit?
No ☐
Yes ☐

**Initial Treatment**

Initially on
ERT ☐
clinical trial? ☐

Date of first infusion: --/--/---

Age when first infusion received:

Initial type of treatment:

Initial dose: ___mg

Initial frequency:
weekly ☐
every 2 weeks ☐

**Current treatment**

Is patient currently on
ERT ☐
clinical trial ☐

Date started current treatment?

Current type of treatment

Current dose: ___mg

Current frequency:
weekly ☐
every 2 weeks ☐

Hospital/ home infusion:
Infusion in Hospital ☐
Infusion at Home ☐

If Home: pick list:
Nurse infuses ☐
Nurse cannulates and leaves ☐
Patient cannulates and infuses ☐

Is the patient currently experiencing:

Febrile reactions?
No ☐
Yes ☐

Analphylactoid reactions?
No ☐
Yes ☐

Does the patient require any pre-medication: No ☐
Yes ☐

**Antibody status**

Measure date: __ __ / __ __ / __ __ __ __

Antibody status to infused product, Tested No ☐
Yes ☐ If Yes, Positive No ☐
Yes ☐

**Stopped Treatment**

Final treatment on ERT ☐
clinical trial ☐

Date of last infusion: __ __ / __ __ / __ __ __ __

Final type of treatment

Final dose: ___mg

Final frequency: weekly ☐
every 2 weeks ☐

Hospital/ home infusion: Infusion in Hospital ☐
Infusion at Home ☐ If Home: pick list: Nurse infuses ☐
Nurse cannulates and leaves ☐
Patient cannulates and infuses ☐

Why was treatment stopped?

# Appendix 11f Clinical Record Form for NPC patients

NPC Patient ID:

Date of visit ___/ ___ / _____

## NPC CRFs

## Inclusion Criteria

Has Clinician given consent for the patient or their carer to be approached?

No ☐ Do not continue
Yes ☐

Has the patient given informed signed consent?

No ☐ Do not continue
Yes ☐

Has the parent/carer given informed signed consent?

No ☐ Do not continue
Yes ☐

Reason for not giving consent (if given):

_____

Hospital ID: _____

Patient Identification number _____

Date of consent __ __ / __ __ / __ __ __ __

Version of consent form: _____

# Patient Information

**General Patient Information for MPS1 Adults and Children**

**Date of birth** __ __ / __ __ / __ __ __ __

**Gender**
Male ☐
Female ☐

**Does the patient have a carer**
Yes ☐
No ☐

**Onset of first symptoms**
Roughly ☐
exactly ☐
can't remember ☐ __ __ / __ __ / __ __ __ __

**Any other family members affected?**

No ☐
Yes ☐
Unknown ☐
If so, how many? __
Relationship to patient:
Mother ☐
Father ☐
Sibling ☐
Other ☐ Please Specify_____

**Month and Year of Diagnosis** Month and year: ___ / ____

**Method of diagnosis**
Recorded ☐
Not recorded ☐

**Cultured skin fibroblasts:**

Studies of filipin staining
☐ Normal
☐ Abnormal

LDL-induced cholesteryl ester Formation
☐ Normal
☐ Abnormal

NPC1 cDNA
☐ Done
☐ Not done

NPC1 cDNA results as amino acid change _____
Please use the following format e.g. Thr1036Met/Gln928Pro

NPC2 cDNA
☐ Done
☐ Not done

NPC2 cDNA results as amino acid change _____
Please use the following format e.g. Glu20Ter/Ser67pro

**Reason for diagnosis**

**Was patient detected by family screening?** ☐ No
☐ Yes

**Did the patient present with symptoms?** ☐ No
☐ Yes
If so, what? _____

**What symptoms did they present with?** ☐ Hepatic
☐ Splenic
☐ Pulmonary
☐ Neurological
☐ Psychiatric
☐ Ophthalmic
☐ Haematological
☐ Other
Please specify_____

**Physician of initial presentation:**

**Please specify_____**

☐ GP
☐ Neonatal obstetrician / specialist
☐ Psychiatrist
☐ Haematologist
☐ Other

**Current Employment Status** Paid Employed ☐
Unemployed ☐
Unpaid Employment ☐

Select as appropriate

Full time ☐
Part time ☐

# Neonatal Symptoms

**Cholestatic jaundice/ hepatomegaly**

☐Present
☐Absent
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

This was detected
☐Antenatally from scan
☐Postnatally

Is treatment required? ☐No
☐Yes

Liver volume (ml) _____ from MRI/CT

**And / or:**

Liver size (cm) _____ No. of cm below the costal margin on the mid clavicular line

**Splenomegaly**

☐Present
☐Absent
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

This was detected
☐Antenatally from scan
☐Postnatally

Is treatment required? ☐No
☐Yes

Spleen volume (ml) _____ from MRI/CT

**And / or:**

Spleen size (cm) _____ No. of cm below the costal margin on the mid clavicular line

**Hydrops fetalis** (Abnormal accumulation of fluid)

☐Present
☐Absent
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

Ascites present ☐No
☐Yes

Oedeama present ☐No
☐Yes

**Is Special Care Baby Unit (SCBU) Care required?**
☐No
☐Yes

**Liver function/ failure - is the Prothrombin Time (PT) greater than 100?**
☐No
☐Yes
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

# CNS measures

**Learning Difficulties / Cognitive impairment**
☐Recorded
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

**Severity of learning difficulties:**

☐**0** No learning difficulties
☐**1** Mild language and/or memory impairment, school or work difficulties, but still attending school or working
☐**2** Moderate to severe impairment of school or work function, but still at school or work, able to converse. Obvious memory impairment
☐**3** Unable to attend school or work, but able to perform most self care and function at home. Severe language and memory impairment
☐**4** No functional memory or language.

**Seizures / Epilepsy**
☐Recorded
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

On anti epileptic medication?
☐Yes
☐No
☐Not recorded

**Severity of Seizures**:

☐**0** No seizures
☐**1** Provoked seizures only (i.e. with fever or intercurrent illness)
☐**2** One seizure per month or less frequently
☐**3** One seizure per week to one seizure per month
☐**4** More than one seizure per week

**Other Cortical Signs:**
☐Present
☐Absent
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

Please specify: _____

**Vertical supranuclear gaze palsy**

☐Recorded
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

**Severity of Vertical supranuclear gaze palsy**:
☐**0** Normal
☐**1** Prolonged saccadic latency only
☐**2** Mildly to moderately slowed saccades
☐**3** Marked slowing of saccades
☐**4** Absent saccades

**Ataxia**
☐Recorded
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

**Severity of ataxia**:
☐**0** Normal
☐**1** Apparent only on tandem walking
☐**2** Ataxia on straight gait
☐**3** Able to walk only with assistance
☐**4** Unable to walk

**Pyramidal tract dysfunstions** (sic!) **(movement disorders)**
☐Recorded
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

**Severity of Pyramidal tract dysfunction**:
☐**0** Normal
☐**1** Normal tone with increased reflexes
☐**2** Mildly to moderately increased tone and reflexes
☐**3** Marked spasticity with gait impairment (scissoring)
☐**4** Severe spasticity with inability to walk.

**Dystonia / difficulty in positioning limbs**
☐Recorded
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

**Severity of Dystonia**:
☐**0** Normal
☐**1** Dystonia apparent only with action (e.g. posturing of hands or feet with walking, stressed or unstressed)
☐**2** Dystonia of one limb, apparent at rest
☐**3** Dystonia of two or more limbs, sparing axial muscles
☐**4** Generalised dystonia

**Speech – Dysarthia "slurred or irregular"**
☐Recorded
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

**Severity of Speech problems**:
☐**0** Normal
☐**1** mild dysarthria, not impairing intelligibility
☐**2** moderate dysarthria, impairing intelligibility of more than 50% of spoken words to independent observer
☐**3** Severe dysarthria, with most speech unintelligible to independent observer
☐**4** Anarthria

**Swallowing difficulties/Dysphagia**
☐Recorded
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

**Severity of swallowing difficulties/Dysphagia:**
☐**0** Normal
☐**1** choking/regurgitation with thin liquids
☐**2** occasional choking on dry solids
☐**3** choking/regurgitation with thick liquids
☐**4** unable to swallow

**Cataplexy:**
☐Recorded
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

**Severity of Cataplexy:**
☐**0** Normal
☐**1** head nodding episodes only
☐**2** Episodes impairing tone beyond head nodding, with or without falls, not more than three times weekly
☐**3** Episodes impairing tone beyond head nodding, with or without falls, more than three times weekly to daily
☐**4** Episodes impairing tone beyond head nodding, with or without falls, one or more per day

**Myclonic jerks present**
☐No
☐ Yes
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

# Visceral

**Splenomegaly (Abnormal enlargement of the spleen)**
☐Recorded
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

**Severity of Splenomegaly:**
☐**0** No spleen enlargement
☐**1** just palpable at costal margin
☐**2** up to 5 cm palpable
☐**3** 5-10 cm palpable
☐**4** > 10 cm palpable

**Hepatomegaly (Abnormal enlargement of the liver)**
☐Recorded
☐Not recorded

Measure Date: __ __ / __ __ / __ __ __ __

**Severity of Hepatomegaly:**
☐**0** No liver enlargement
☐**1** Palpable just below costal margin, not accounted for by hepatoptosis
☐**2** Up to 5 cm palpable below costal margin, not accounted for by hepatoptosis
☐**3** Enlarged > 5 < 15 cm below costal margin, not accounted for by hepatoptosis
☐**4** Enlarged > 15 cm below costal margin, not accounted for by hepatoptosis

# Audiometry

Evidence of hearing loss: No ☐
Yes ☐

Hearing Test: Done ☐
Not Done ☐

Measure date: __ __ / __ __ / __ __ __ __

**Type of hearing loss (pick one)**

Conductive ☐
Sensorineural ☐
Mixed (combination of conductive & sensorineural ☐

**Degree of hearing loss (average decibels over tested frequencies) (pick one)**

Mild (0-40 decibels) ☐
Moderate (41-70 decibels) ☐
Severe (71-95 decibels) ☐
Profound (over 95 decibels) ☐

Note: to calculate the degree of hearing loss, add all the decibels and divide by the number of data points.

Hearing Aid Used No ☐
Yes ☐
Not Recorded ☐

Type of hearing aid_____

Measure date: __ __ / __ __ / __ __ __ __

# Other measures

**Does the patient have Inflammatory Bowel Disease?**
☐Present
☐ Absent
☐Not recorded

Measure date: __ __ / __ __ / __ __ __ __

**Does the patient have Psychiatric Symptoms?**
☐Present
☐ Absent
☐Not recorded

Measure date: __ __ / __ __ / __ __ __ __

Please specify: _____

**Does the patient have urinary incontinence?**
☐ Yes
☐ No
☐Not recorded

Measure date: __ __ / __ __ / __ __ __ __

**Does the patient have faecal incontinence?**
☐ Yes
☐ No
☐Not recorded

Measure date: __ __ / __ __ / __ __ __ __

# Body Measurements

**Height**
☐ Recorded current age _____
☐Not recorded

Measure date: __ __ / __ __ / __ __ __ __

Height (cm) _____

**Weight**
☐ Recorded
☐Not recorded

Measure date: __ __ / __ __ / __ __ __ __

Weight (kg) _____

# Significant Co-morbidity

**Does the patient have any co-morbidities? Yes / No**

**How many? ____**

Please enter significant co-morbidities in the spaces below, and then answer yes or no to the organs affected

1

2

3

4

5

6

| | **1** | **2** | **3** | **4** | **5** | **6** |
|---|---|---|---|---|---|---|
| CNS | Yes / No | Yes / No | Yes / No | Yes/No | Yes / No | Yes/No |
| Visceral | Yes / No | Yes / No | Yes / No | Yes/No | Yes / No | Yes/No |
| Audiometry | Yes / No | Yes / No | Yes / No | Yes/No | Yes / No | Yes/No |
| Bowels | Yes / No | Yes / No | Yes / No | Yes/No | Yes / No | Yes/No |
| Psychiatric symptoms | Yes / No | Yes / No | Yes / No | Yes/No | Yes / No | Yes/No |
| Incontinence | Yes / No | Yes / No | Yes / No | Yes/No | Yes / No | Yes/No |

**Children Only:**

| | | | | | | |
|---|---|---|---|---|---|---|
| Neonatal Symptoms | Yes / No | Yes / No | Yes / No | Yes/No | Yes / No | Yes/No |
| Growth | Yes / No | Yes / No | Yes/No | Yes/No | Yes / No | Yes/No |

# Con-meds

**Is the patient taking any con-meds?**

| Medication | Prescription / Non prescription | Dose | Frequency | Date started |
|---|---|---|---|---|

# Withdrawal from the study

**Is the patient still in the full study?** No ☐
Yes ☐

Withdrawal date: __ __ / __ __ / __ __ __ __

Reason for Withdrawal

☐ Patient has life threatening disease
☐ Failure to comply (due to medication)
☐ Evidence of disease progression
☐ Patient is pregnant
☐ Death
☐ Patient is on a clinical trial
☐ Questionnaire burden
☐ Patient turned 16 and didn't re-consent
☐ Other Please specify_____

Continue in the study notes only? No ☐
Yes ☐

**Record of death**

Date of death: __ __ / __ __ / __ __ __ __

Did clinician certify this death as condition related? No ☐
Yes ☐

Please record the wording from the dearth certificate:

Part 1a ---------- Free text (Disease or condition directly leading to death)

Part 1b ---------- Free text (Other disease or condition, if any, leading to 1a)

Part 1c ---------- Free text (Other disease or condition, if any, leading to 1b)

Part 2 ---------- Free text (Other significant conditions CONTRIBUTING TO THE DEATH but not related to the disease or condition causing it)

# Record of Therapy (Enzyme Replacement Therapy, substrate reduction therapy etc):

Is patient on SRT No ☐
Yes ☐

**Initial Treatment**
Initially on SRT ☐
clinical trial? ☐

Date of first Infusion: __ __ / __ __ / __ __ __ __

Age when first infusion received: _____

Initial type of treatment: _____

Initial dose: ___(units)

Initial frequency: weekly ☐
every 2 weeks ☐
Please specify:_____

Initial type of treatment (substrate reduction therapy)

Initial dose: ___mg

Initial frequency: 3 times per week ☐
2 times per week ☐
1 time per week ☐
2 weekly ☐
Other ☐ Please Specify_____

**Current treatment**
Currently on SRT or clinical treatment? (drop down menu)

Date started current treatment? _____

Current type of treatment

Current dose: ___units

Current frequency: 3 times per week ☐
2 times per week ☐
1 time per week ☐
2 weekly ☐
Other ☐ Please Specify_____

Hospital/ home infusion: Infusion in Hospital ☐
Infusion at Home ☐

If Home: pick list:
Nurse infuses ☐
Nurse cannulates and leaves ☐
Patient cannulates and infuses ☐

Is the patient currently experiencing:

Febrile reactions? No ☐
Yes ☐

Anaphylactoid reactions? No ☐
Yes ☐

Does the patient require any pre-medication: No ☐
Yes ☐

Current type of treatment (substrate reduction therapy) _____

Current Dose _____(mg)

Current frequency: 3 times per week ☐
2 times per week ☐
1 time per week ☐
2 weekly ☐
Other ☐ Please Specify_____

**Antibody status**

Measure date: __ __ / __ __ / __ __ __ __

Antibody status to infused product, Tested No ☐
Yes ☐ If Yes,

Antibody Status Test Result Positive No ☐
Yes ☐
Don't know ☐

# Record of Therapy (Enzyme Replacement Therapy, substrate reduction therapy etc):

Is patient on SRT No ☐
Yes ☐

**Initial Treatment**
Initially on SRT ☐
clinical trial? ☐

Date of first Infusion: __ __ / __ __ / __ __ __ __

Age when first infusion received: _____

Initial type of treatment: _____

Initial dose: ___(units)

Initial frequency: weekly ☐
every 2 weeks ☐
Please specify:_____

Initial type of treatment (substrate reduction therapy)

Initial dose: ___mg

Initial frequency: 3 times per week ☐
2 times per week ☐
1 time per week ☐
2 weekly ☐
Other ☐ Please Specify_____

**Current treatment**
Currently on SRT or clinical treatment? (drop down menu)

Date started current treatment? _____

Current type of treatment (ERT)

Current dose: ___units

Current frequency: 3 times per week ☐
2 times per week ☐
1 time per week ☐
2 weekly ☐
Other ☐ Please Specify_____

Hospital/ home infusion: Infusion in Hospital ☐
Infusion at Home ☐ If Home: pick list:
Nurse infuses ☐
Nurse cannulates and leaves ☐
Patient cannulates and infuses ☐

Is the patient currently experiencing:

Febrile reactions? No ☐
Yes ☐

Anaphylactoid reactions? No ☐
Yes ☐

Does the patient require any pre-medication: No ☐
Yes ☐

Current type of treatment (substrate reduction therapy) _____

Current Dose _____(mg)

Current frequency: 3 times per week ☐
2 times per week ☐
1 time per week ☐
2 weekly ☐
Other ☐ Please Specify_____

**Antibody status**

Measure date: __ __ / __ __ / __ __ __ __

Antibody status to infused product, Tested No ☐
Yes ☐ If Yes,

Antibody Status Test Result Positive
No ☐
Yes ☐
Don't know ☐

**Stopped Treatment**
Final treatment on SRT or clinical trial (drop down menu)

Date of last Infusion: __ __ / __ __ / __ __ __ __

Final type of treatment _____

Final weight ____kg

Final dose: ___units

Final frequency: 3 times per week ☐
2 times per week ☐
1 time per week ☐
2 weekly ☐
Other ☐ Please Specify_____

Hospital/ home infusion: Infusion in Hospital ☐
Infusion at Home ☐ If Home: pick list: Nurse infuses ☐
Nurse cannulates and leaves ☐
Patient cannulates and infuses ☐

Final type of treatment (substrate reduction therapy)_____

Final dose: ___mg

Final frequency: 3 times per week ☐
2 times per week ☐
1 time per week ☐
2 weekly ☐
Other ☐ Please Specify_____

Why was treatment stopped?
Please specify:_____