## Clinical Dataset

### Presenting features triggering referral

### Age of onset of presenting symptoms

- [ ] Prenatal
- [ ] Neonatal (Birth to 28 days)
- [ ] Infantile (> 28 days month to 1 year)
- [ ] Childhood (>1 to 5 years)

### Evolution of symptoms

- [ ] Slowly progressive (> 6 months)
- [ ] Rapidly progressive (< 6 months)
- [ ] Acute-episodic (relapsing-remitting)

Deterioration with:

- [ ] Head injury
- [ ] Infection / Inflammation
- [ ] Anaesthetic

[ ] Non- progressive

### Pregnancy and Neonatal History

#### Prenatal

i. Pregnancy

- Normal
- Abnormal
- Unknown

Details:

a) Foetal movements  
- Normal  
- Abnormal

b) Foetal dysmorphology  
- Yes  
- No

c) Polyhydramnios  
- Yes  
- No

d) IUGR  
- Yes  
- No

ii. Labour

- Normal  
- Abnormal  
- Unknown

Specify:
- eg CTG abnormalities?  
- Meconium stained liquor?

#### Neonatal

i. Ventilatory support

- Yes  
- No

a) Improved over time?

- Yes  
- No

ii. Feeding assistance

- Yes  
- No

b) Improved over time?

- Yes  
- No

iii. Delayed head/neck control

- Yes  
- No

iv. Apgar scores

<table>
<thead>
<tr>
<th>1 minute</th>
<th>5 minutes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>Yes</td>
<td>No</td>
</tr>
</tbody>
</table>

v. Seizures

- Yes  
- No

vi. Hypoglycaemia

- Yes  
- No

Other neonatal issues:

- Delayed early motor milestones
- Delayed social development
- Delayed speech and language developmental
- Intellectual disability

- a. Severity  
- Mild  
- Moderate  
- Severe  
- Profound

- Not Assessed
Leukodystrophy Flagship
Optimal Clinical Data
Version 1, 24.9.19

<table>
<thead>
<tr>
<th>Plateau of motor skills</th>
<th>Yes</th>
<th>No</th>
<th>Age of onset:</th>
</tr>
</thead>
<tbody>
<tr>
<td>Regression of motor skills</td>
<td>Yes</td>
<td>No</td>
<td>Age of onset:</td>
</tr>
<tr>
<td>Highest motor milestone achieved:</td>
<td>Yes</td>
<td>No</td>
<td>Age of onset:</td>
</tr>
<tr>
<td>Regression of cognitive skills</td>
<td>Yes</td>
<td>No</td>
<td>Age of onset:</td>
</tr>
<tr>
<td>Age at last review:</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>Neuropsychiatric or behavioural symptoms</td>
<td>Yes</td>
<td>No</td>
<td>Age of onset:</td>
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</table>

viii. Comment:

<table>
<thead>
<tr>
<th>GMFCS</th>
<th>1</th>
<th>2</th>
<th>3</th>
<th>4</th>
<th>5</th>
<th>Not assessed</th>
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</thead>
<tbody>
<tr>
<td>MACS</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
<td>5</td>
<td>Not assessed</td>
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<tr>
<td>EDACS</td>
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<td>2</td>
<td>3</td>
<td>4</td>
<td>5</td>
<td>Not assessed</td>
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</tbody>
</table>

Cognitive assessment/ CFCS

**Epilepsy**

i. Seizure type [ ] Generalised [ ] Focal

ii. Comment:

<table>
<thead>
<tr>
<th>Sensorineural deafness</th>
<th>Yes</th>
<th>No</th>
<th>Not Assessed</th>
</tr>
</thead>
<tbody>
<tr>
<td>if present</td>
<td>congenital</td>
<td>later onset</td>
<td></td>
</tr>
<tr>
<td>if present</td>
<td>unilateral</td>
<td>bilateral</td>
<td></td>
</tr>
</tbody>
</table>

Audiology [ ] Normal [ ] Abnormal [ ] Not Assessed

Date of test:

Audiology common:

**Ophthalmological abnormalities**

i. Cortical blindness [ ] Yes [ ] No [ ] Not Assessed

ii. Optic atrophy/neuropathy [ ] Yes [ ] No [ ] Not Assessed

iii. Retinitis pigmentosa / retinal dystrophy [ ] Yes [ ] No [ ] Not Assessed

iv. Cataracts [ ] Yes [ ] No [ ] Not Assessed

v. Nystagmus [ ] Yes [ ] No [ ] Not Assessed

vi. Cherry red spot [ ] Yes [ ] No [ ] Not Assessed

vii. Glaucoma [ ] Yes [ ] No [ ] Not Assessed

vii. Vascular abnormalities [ ] Yes [ ] No [ ] Not Assessed

viii. Oculomotor apraxia [ ] Yes [ ] No [ ] Not Assessed

**Endocrinological abnormalities**

i. Diabetes mellitus [ ] Yes [ ] No [ ] Not Assessed

ii. Diabetes insipidus [ ] Yes [ ] No [ ] Not Assessed

iii. Hypopituitarism [ ] Yes [ ] No [ ] Not Assessed

iv. Hypothyroidism [ ] Yes [ ] No [ ] Not Assessed

v. Adrenal insufficiency [ ] Yes [ ] No [ ] Not Assessed

vi. Hypogonadotrophic hypogonadism [ ] Yes [ ] No [ ] Not Assessed

vii. Growth hormone deficiency [ ] Yes [ ] No [ ] Not Assessed

vii. SIADH [ ] Yes [ ] No [ ] Not Assessed

vii. Ovarian dysfunction [ ] Yes [ ] No [ ] Not Assessed

i. Comment:

**GI Tract abnormalities**

i. Gastro-oesophageal reflux [ ] Yes [ ] No

ii. NG/tube feeding [ ] Yes [ ] No Age at insertion: ____

iii. Gall bladder disease [ ] Yes [ ] No
### Leukodystrophy Flagship
#### Optimal Clinical Data
**Version 1, 24.9.19**

**Australian Genomics**

---

### v. Constipation
- Yes
- No

### vi. Diarrhoea
- Yes
- No

#### vii. Comment:

### Skin

**i. Photosensitivity**
- Yes
- No

**ii. Sensitivity to pressure sores and/or poor wound healing**
- Yes
- No

---

### Neurological Examination Findings

#### Age at Last Assessment:

#### Head Size (OFC):

#### Cranial Nerves:

- **EOM / gaze palsy**
  - Yes
  - No
  - Details:

- **Bulbar palsy**
  - Yes
  - No

- **Pseudobulbar palsy**
  - Yes
  - No

#### TONE

**i. Axial Hypotonia**
- Yes
- No

**ii. Appendicular hypotonia**
- Yes
- No

**iii. Hypertonia**
- Left upper limb
  - Yes
  - No
- Left lower limb
  - Yes
  - No
- Right upper limb
  - Yes
  - No
- Right lower limb
  - Yes
  - No

**iv. Details: Dystonia /Spasticity / Mixed (circle)**

---

### Strength

#### LEFT

- **Upper limb**
  - Normal
  - Reduced

- **Lower limb**
  - Normal
  - Reduced

#### RIGHT

- **Upper limb**
  - Normal
  - Reduced

- **Lower limb**
  - Normal
  - Reduced

#### MRC score

#### Reflexes

**i. Upper limb**
- Normal
- Hyper
- Hypo
- Not elicited

**ii. Lower limb**
- Normal
- Hyper
- Hypo
- Not elicited

**iii. Patellar**
- Normal
- Hyper
- Hypo

**v. Plantars**
- Up
- Down
- Absent

**vi. Plantars**
- Up
- Down
- Absent

#### vi. Comment:

- Clonus
  - Yes
  - No

---

### Sensation

#### Peripheral neuropathy
- Yes
- No

#### Details:

Other sensory deficits:

---

### Extrapyramidal Features

**i. Myoclonus**
- Yes
- No

**ii. Chorea-thetosis**
- Yes
- No

**iii. Tremor**
- Yes
- No

**iv. Parkinsonism**
- Yes
- No

---

### Cerebellar Dysfunction
### Other Neurological Abnormalities

**Comment:**

### General Examination Findings

**HEIGHT:**  
**WEIGHT:**

<table>
<thead>
<tr>
<th>Dysmorphic features</th>
<th>Yes</th>
<th>No</th>
<th>Comment:</th>
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<tbody>
<tr>
<td>Abnormal pigmentation</td>
<td>Yes</td>
<td>No</td>
<td>Comment:</td>
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<tr>
<td>Dental abnormalities</td>
<td>Yes</td>
<td>No</td>
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<tr>
<td>Genital abnormalities</td>
<td>Yes</td>
<td>No</td>
<td>Comment:</td>
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<tr>
<td>Tendinous xanthomas / xanthelasma</td>
<td>Yes</td>
<td>No</td>
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<tr>
<td>Hepatomegaly</td>
<td>Yes</td>
<td>No</td>
<td>Comment:</td>
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<tr>
<td>Splenomegaly</td>
<td>Yes</td>
<td>No</td>
<td>Comment:</td>
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<tr>
<td>Skin abnormalities</td>
<td>Yes</td>
<td>No</td>
<td>Comment:</td>
</tr>
<tr>
<td>Hair abnormalities</td>
<td>Yes</td>
<td>No</td>
<td>Comment:</td>
</tr>
<tr>
<td>Joint abnormalities</td>
<td>Yes</td>
<td>No</td>
<td>Comment:</td>
</tr>
<tr>
<td>Cardiac abnormalities</td>
<td>Yes</td>
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<tr>
<td>Digital anomalies</td>
<td>Yes</td>
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### Investigations

#### Previous Microarray and Genotype Data

### Laboratory Examinations

**TORCH SEROLOGY**

<table>
<thead>
<tr>
<th>Normal</th>
<th>Abnormal</th>
<th>Not Assessed</th>
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**Urine CMV**

<table>
<thead>
<tr>
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<th>Abnormal</th>
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</thead>
</table>

**CMV PCR from Guthrie card**

<table>
<thead>
<tr>
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<th>Abnormal</th>
<th>Not Assessed</th>
</tr>
</thead>
</table>

**Very long chain fatty acids (including phytic an / pristinic)**

<table>
<thead>
<tr>
<th>Normal</th>
<th>Abnormal</th>
<th>Not Assessed</th>
</tr>
</thead>
</table>

1. **Lysosomal enzymes**

2. **Plasma amino acids**

3. **Urine amino acids**

4. **Urine organic acids**

5. **Urine GAGs**

6. **Urine oligosaccharides**

7. **Urine polys**

8. **Urinary sulphatides**

9. **Vitamin B12**

10. **Creatine kinase**

11. **Thyroid function including reverse T3**

12. **Liver function tests**

13. **Full blood exam**

14. **Urine bile acids (Cholestanol)**

If abnormal is selected for any test, above, please describe:

15.
# Leukodystrophy Flagship
## Optimal Clinical Data
### Version 1, 24.9.19

<table>
<thead>
<tr>
<th>Test Description</th>
<th>Value</th>
<th>Normal Range</th>
<th>Date of Test</th>
<th>Notes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Highest blood lactate</td>
<td>_____ mmol/L</td>
<td>_____ normal range</td>
<td>Date of test:</td>
<td>sample – venous / arterial [nb: torniquet sample Y/N]</td>
</tr>
<tr>
<td>Venous pyruvate</td>
<td>_____ umol/L</td>
<td>_____ normal range</td>
<td>Date of test:</td>
<td></td>
</tr>
<tr>
<td>CSF lactate</td>
<td>_____ mmol/L</td>
<td>_____ normal range</td>
<td>Date of test:</td>
<td></td>
</tr>
<tr>
<td>CSF pyruvate</td>
<td>_____ umol/L</td>
<td>_____ normal range</td>
<td>Date of test:</td>
<td></td>
</tr>
<tr>
<td>CSF alanine</td>
<td>_____ umol/L</td>
<td>_____ normal range</td>
<td>Date of test:</td>
<td></td>
</tr>
<tr>
<td>CSF glycine</td>
<td>_____ umol/L</td>
<td>_____ normal range</td>
<td>Date of test:</td>
<td></td>
</tr>
<tr>
<td>CSF protein</td>
<td>_____ g/L</td>
<td>_____ normal range</td>
<td>Date of test:</td>
<td></td>
</tr>
<tr>
<td>CSF folate</td>
<td>_____ nmol/L</td>
<td>_____ normal range</td>
<td>Date of test:</td>
<td></td>
</tr>
<tr>
<td>CSF microscopy</td>
<td></td>
<td></td>
<td></td>
<td>WCC_____ RCC_____</td>
</tr>
<tr>
<td>CSF oligoclonal bands</td>
<td></td>
<td>Present Absent Not done</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Paired tests</td>
<td></td>
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</tr>
<tr>
<td>Venous lactate &amp; CSF lactate</td>
<td></td>
<td>Yes No</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Venous pyruvate &amp; CSF pyruvate</td>
<td></td>
<td>Yes No</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Plasma alanine &amp; CSF alanine</td>
<td></td>
<td>Yes No</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other (Biochemical / Histopath / Ultrastructural)</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

## Neurophysiology Testing

### a. Electromyography (EMG)

<table>
<thead>
<tr>
<th>Test Description</th>
<th>Assessed Not Assessed</th>
</tr>
</thead>
<tbody>
<tr>
<td>i. Myopathic</td>
<td>Yes No</td>
</tr>
<tr>
<td>ii. Mixed</td>
<td>Yes No</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Test Description</th>
<th>Assessed Not Assessed</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**EMG Comment:**

### b. Nerve conduction studies

<table>
<thead>
<tr>
<th>Test Description</th>
<th>Assessed Not Assessed</th>
</tr>
</thead>
<tbody>
<tr>
<td>Which nerves:</td>
<td></td>
</tr>
<tr>
<td>i. Motor:</td>
<td>Demyelinating Axonal Mixed Normal Not Assessed</td>
</tr>
<tr>
<td>ii. Sensory:</td>
<td>Demyelinating Axonal Mixed Normal Not Assessed</td>
</tr>
</tbody>
</table>

**Comment:**

### c. Electroencephalogram

<table>
<thead>
<tr>
<th>Test Description</th>
<th>Normal Abnormal Not Assessed</th>
</tr>
</thead>
<tbody>
<tr>
<td>Date of test:</td>
<td></td>
</tr>
</tbody>
</table>

**Findings:**

### Skeletal radiographs:

#### Psychology / Neuropsychology Assessment

**Comment:**
# Imaging Dataset

## Imaging Data

<table>
<thead>
<tr>
<th></th>
<th>Yes</th>
<th>No</th>
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<tbody>
<tr>
<td>Brain MRI</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Age at first MRI scan</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Age at last MRI scan</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

## Age at first / last MRI scan

- **Age at first MRI scan**
  - **Number of months / years**: _____
  - **Field strength**: 1.5T \(\square\) 3T \(\square\)
- **Age at last MRI scan**
  - **Number of months / years**: _____
  - **Field strength**: 1.5T \(\square\) 3T \(\square\)

## Signal Characteristics

<table>
<thead>
<tr>
<th>Characteristic</th>
<th></th>
<th></th>
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</thead>
<tbody>
<tr>
<td>T2 Hyperintensity</td>
<td></td>
<td></td>
</tr>
<tr>
<td>T1 Hypointensity</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mild T2 Hyperintensity</td>
<td></td>
<td></td>
</tr>
<tr>
<td>T1 Hyperintensity or T1 Isointensity or Mild T1 Hypointensity</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

## Affected white matter structures

1. □ Periventricular
2. □ Central
3. □ U-Fibres
4. □ Other – Specify: ________________________________

## Specified regions

- □ Frontal
- □ Parietal
- □ Occipital
- □ Temporal

## Specify predomiance:

- □ Hilus dentate nucleus
- □ Cerebellar peduncles
- □ Brain stem structures

## Is posterior fossa involvement predominant?

- □ Yes
- □ No

## Corpus callosum affected?

- □ Yes
- □ No

<table>
<thead>
<tr>
<th>Area</th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Anterior</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Middle</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Outer leaflet</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Posterior</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

## Internal capsule affected?

- □ Yes
- □ No

<table>
<thead>
<tr>
<th>Area</th>
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</thead>
<tbody>
<tr>
<td>Anterior Limb</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Posterior Limb</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

## External capsule affected?

- □ Yes
- □ No

## Distribution of the white matter signal abnormality

- □ Confluent
- □ Isolated/Multifocal
- □ Homogenous
- □ Symmetrical
- □ Low signal on flair (rarefaction)

## Is there contrast enhancement?

- □ Yes
- □ No
- □ Not done

## Is there restricted diffusion?

- □ Yes
- □ No
- □ DWI not done

## MR Spectroscopy

- □ Normal
- □ Abnormal
- □ Not done

## Spectroscopy Details: ________________________________
<table>
<thead>
<tr>
<th>State of myelination:</th>
<th>Adequate for age</th>
<th>Delayed</th>
<th>Arrested</th>
<th>Arrested in early stage</th>
<th>Abnormal irregular myelination</th>
</tr>
</thead>
<tbody>
<tr>
<td>Grey matter lesions:</td>
<td></td>
<td></td>
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</tr>
<tr>
<td></td>
<td>Cerebral cortex</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Caudate nucleus</td>
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<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Putamen</td>
<td></td>
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<td></td>
</tr>
<tr>
<td></td>
<td>Globus pallidus</td>
<td></td>
<td></td>
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<tr>
<td></td>
<td>Thalamus</td>
<td></td>
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<tr>
<td></td>
<td>Dentate nucleus</td>
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</tr>
<tr>
<td></td>
<td>Other nuclei, details</td>
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<tr>
<td></td>
<td>Cerebellar cortex, details</td>
<td></td>
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<td></td>
</tr>
<tr>
<td>Is there atrophy?</td>
<td>□ Yes □ No</td>
<td></td>
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<td></td>
<td></td>
</tr>
<tr>
<td>Cerebrum:</td>
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</tr>
<tr>
<td></td>
<td>White matter</td>
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<td></td>
</tr>
<tr>
<td></td>
<td>Cortex</td>
<td></td>
<td></td>
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<td></td>
</tr>
<tr>
<td></td>
<td>Ventricular enlargement</td>
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</tr>
<tr>
<td></td>
<td>Enlarged subarachnoid spaces</td>
<td></td>
<td></td>
<td></td>
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</tr>
<tr>
<td>Cerebellum:</td>
<td></td>
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</tr>
<tr>
<td></td>
<td>Vermis</td>
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<tr>
<td></td>
<td>Hemispheres</td>
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<tr>
<td>Extra Elements</td>
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</tr>
<tr>
<td>Cysts:</td>
<td></td>
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</tr>
<tr>
<td></td>
<td>Subcortical</td>
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<tr>
<td></td>
<td>Intraparenchymal</td>
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<tr>
<td></td>
<td>Frontal</td>
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<td>Parietal</td>
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<td>Occipital</td>
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<td>3T Scan Performed?</td>
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<td>Age at 3T scan</td>
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<td>Proton-MRS performed</td>
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<td>General comments</td>
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- Age at 3T scan:  [ ] Yes  [ ] No
- Proton-MRS performed:  [ ] Yes  [ ] No
- Best MRI scan quality:  [ ] Good  [ ] Adequate  [ ] Poor
- Differential diagnosis:  
- Likely genes:  
- General comments:  

*Australian Genomics Leukodystrophy Optimal Clinical Dataset, version 1 24 September 19*
Appendix 1: Beighton Score:
From: http://hypermobility.org/help-advice/hypermobility-syndromes/beighton-score/

The Beighton score is calculated as follows:

1. One point if while standing forward bending you can place palms on the ground with legs straight
2. One point for each elbow that bends backwards
3. One point for each knee that bends backwards
4. One point for each thumb that touches the forearm when bent backwards
5. One point for each little finger that bends backwards beyond 90 degrees.
Appendix 2: Bulbar Vs Pseudobulbar Palsy:

Bulbar palsy – clinical features:
- Gag reflex – absent
- Tongue – wasted, fasciculations
- “wasted, wrinkled, thrown into folds and increasingly motionless”.
- Palatal movement – absent.
- Jaw jerk – absent or normal
- Speech – nasal
- “indistinct (flaccid dysarthria), lacks modulation and has a nasal twang”
- Emotions – normal

Pseudobulbar palsy 0 clinical features:
- Gag reflex – increased or normal
- Tongue – spastic
- “it cannot be protruded, lies on the floor of the mouth and is small and tight”.
- Palatal movement – absent.
- Jaw jerk – increased
- Speech – spastic: “a monotonous, slurred, high-pitched, ‘Donald Duck’ dysarthria” that “sounds as if the patient is trying to squeeze out words from tight lips”.
- Emotions – labile