

Leukodystrophy Molecular Diagnostics Part I

Last Name:
First Name:
DOB:
Gender:

Date of first symptoms: ___/___/___
 Date of Clinical Diagnosis: ___/___/___

Please √ if present:

- BHx** Full Term Prematurity
 Poor feeding Perinatal depression
 Respiratory distress
 hypoglycemia
 Small for GA Large for GA
 Comments: _____

- FHx** MR /DD autism spectrum
 Seizures cerebral palsy
 Demyelinating disease- MS, etc
 Comments: _____

- PMHx** febrile seizure FTT/ growth delay
 delay - motor delay -cognitive
 Comments: _____

Symptoms	previous	now
Encephalopathy	<input type="checkbox"/>	<input type="checkbox"/>
Dementia	<input type="checkbox"/>	<input type="checkbox"/>
Seizures	<input type="checkbox"/>	<input type="checkbox"/>
Gait disturbance	<input type="checkbox"/>	<input type="checkbox"/>
Frequent Emesis	<input type="checkbox"/>	<input type="checkbox"/>
Paroxysmal deterioration	<input type="checkbox"/>	<input type="checkbox"/>
With Fever	<input type="checkbox"/>	<input type="checkbox"/>
With Trauma	<input type="checkbox"/>	<input type="checkbox"/>

Clinical Exam

- Organomegaly
 Skin findings _____
 macrocephaly microcephaly
 hyperexcitability exaggerated startle

Cranial Nerves/ Bulbar

- normal
 optic atrophy visual impairment
 nystagmus strabismus
 facial weakness deafness
 dysphonia dysarthria
 dysphagia respiratory failure

Motor

- normal
 spasticity hypotonia
 focal weakness proximal weakness
 dystonia tremor
 choreoathetosis

- Sensory Exam** normal
 vibratory loss proprioceptive loss

- Deep tendon reflexes** normal
 hyperreflexia hyporeflexia

- Gait/ Cerebellar** normal
 ataxia dysmetria
 walks with assistance wheelchair bound

Magnetic Resonance Imaging

Please include copies of available studies

Laboratory data collected: please √ if normal

- | | | |
|--|--|---|
| <input type="checkbox"/> Karyotype | <input type="checkbox"/> Cholesterol | <input type="checkbox"/> Cholestanol |
| <input type="checkbox"/> CK | <input type="checkbox"/> vit B12 | <input type="checkbox"/> folic acid |
| <input type="checkbox"/> vit E | <input type="checkbox"/> TSH | <input type="checkbox"/> NH3 |
| <input type="checkbox"/> VLCFA | <input type="checkbox"/> phytanic acid | <input type="checkbox"/> pipecolic acid |
| <input type="checkbox"/> Lactate | <input type="checkbox"/> Pyruvate | <input type="checkbox"/> L/P ratio |
| <input type="checkbox"/> Cu | <input type="checkbox"/> ceruloplasmin | <input type="checkbox"/> amino acids |
| <input type="checkbox"/> carnitine | <input type="checkbox"/> acylcarnitine | <input type="checkbox"/> TORCH |
| <input type="checkbox"/> Isoelectric focusing of transferrin (CDG) | | |
| <input type="checkbox"/> Mitochondrial DNA | | |

Urine :

- OrganicAcids N acetylaspartic acid
 sialic acid Oligosaccharides
 sulfatide purine pyrimidine

CSE:

- cell count protein glucose
 lactate glycine amino acids
 neurotransmitters

Lysosomal enzymes:

- hexosaminidase A
 arylsulfatase A
 β galactosidase
 Galactosylcerebroside β galactosidase

Muscle biopsy:

- respiratory chain enzymes
 pyruvate dehydrogenase assay
 histology

If any abnormalities please specify:

Thank you!