



# Clinical Information Form

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

## Clinical (Check All That Apply)

- |   |   |  |                                 |
|---|---|--|---------------------------------|
| <b>Eye</b><br><input type="checkbox"/> Retinitis Pigmentosa<br><input type="checkbox"/> Optic Atrophy<br><input type="checkbox"/> Other | <b>Hearing</b><br><input type="checkbox"/> Sensorineural <input type="checkbox"/> Stickler <input type="checkbox"/> Usher | <b>Neuronal Migration</b><br><input type="checkbox"/> Meckel <input type="checkbox"/> Joubert <input type="checkbox"/> Other | <input type="checkbox"/> Stroke |
|---|---|--|---------------------------------|

- Cognitive/Neurobehavioral**  Intellectual Disability (ID)  Syndromic ID  Nonsyndromic ID  Autism  Dementia

- Movement Disorders**  Ataxia  Episodic Ataxia  Dystonia  Chorea/Athetosis  Parkinson Disease  L-Dopa Response

- |   |  |  |
|---|--|--|
| <b>Epilepsy</b><br><input type="checkbox"/> Myoclonic <input type="checkbox"/> Other<br><input type="checkbox"/> Absence <input type="checkbox"/> Tonic Clonic<br><input type="checkbox"/> Epileptic Encephalopathy | <b>Spasticity</b><br><input type="checkbox"/> Spastic Paraplegia <input type="checkbox"/> Other<br><input type="checkbox"/> Spastic Quadriplegia | <b>Connective Tissue &amp; Bone</b><br><input type="checkbox"/> Ehlers Danlos <input type="checkbox"/> Marfan <input type="checkbox"/> Aneurysms<br><input type="checkbox"/> Other |
|---|--|--|

- |   |  |
|---|--|
| <b>Neuromuscular</b><br><input type="checkbox"/> Distal <input type="checkbox"/> Proximal <input type="checkbox"/> Muscle Atrophy <input type="checkbox"/> Contractures<br><input type="checkbox"/> Malignant Hyperthermia <input type="checkbox"/> Arthrogryposis <input type="checkbox"/> Rhabdomyolysis<br><input type="checkbox"/> Periodic Paralysis <input type="checkbox"/> Statin Use <input type="checkbox"/> Myasthenia | <b>Nerve/Anterior Horn Cell</b><br><input type="checkbox"/> Neurofibromas <input type="checkbox"/> Charcot-Marie-Tooth <input type="checkbox"/> Sensory<br><input type="checkbox"/> Autonomic <input type="checkbox"/> Pain <input type="checkbox"/> Motor <input type="checkbox"/> Nerve Conduction<br><input type="checkbox"/> Other |
|---|--|

- |   |   |  |  |
|---|---|--|--|
| <b>Cardiomyopathy</b><br><input type="checkbox"/> Dilated <input type="checkbox"/> Hypertrophic<br><input type="checkbox"/> Noncompaction | <b>Arrhythmias</b><br><input type="checkbox"/> Ventricular Tachycardia <input type="checkbox"/> Brugada<br><input type="checkbox"/> Long or Short QT <input type="checkbox"/> Conduction Defect | <b>Congenital Heart Defects</b><br><input type="checkbox"/> Heterotaxy<br><input type="checkbox"/> Other | <b>Endocrine</b><br><input type="checkbox"/> Hypothyroidism <input type="checkbox"/> Other<br><input type="checkbox"/> Diabetes Mellitus |
|---|---|--|--|

## Imaging (Check All That Apply)

- |   |   |   |
|---|---|---|
| <b>Brain MRI</b><br><input type="checkbox"/> Leigh Disease<br><input type="checkbox"/> Basal Ganglia Calcification<br><input type="checkbox"/> Stroke<br><input type="checkbox"/> Cerebellar Atrophy<br><input type="checkbox"/> Abnormal Myelin (describe) | <b>EEG (Describe Findings)</b><br>_____ | <b>EMG/NVC (Describe Findings)</b><br>_____ |
|---|---|---|

## Laboratory

- |   |  |
|---|--|
| <b>Metabolic (Describe Findings)</b><br>_____ | <b>Genetic (Describe Findings)</b><br>_____  |
| <b>CPK</b> Maximum _____<br>Minimum _____     | <input type="checkbox"/> Chromosomal Microarray<br><input type="checkbox"/> Deletion/Insertion Testing<br><input type="checkbox"/> Other (comment) |

## Family History

- Ethnicity (please check)**
- |                                    |   |   |                                       |
|------------------------------------|---|---|---------------------------------------|
| <input type="checkbox"/> Caucasian | <input type="checkbox"/> Sephardic Jewish | <input type="checkbox"/> African American (or Black)          | <input type="checkbox"/> Asian        |
| <input type="checkbox"/> Hispanic  | <input type="checkbox"/> Ashkenazi Jewish | <input type="checkbox"/> Native American (or American Indian) | <input type="checkbox"/> Other: _____ |
- |                                  |                                  |                         |
|----------------------------------|----------------------------------|-------------------------|
| <b>Affected Maternal Lineage</b> | <b>Affected Paternal Lineage</b> | <b>Siblings</b>         |
| Relationship to Proband          | Relationship to Proband          | Number (specify gender) |
| Symptoms                         | Symptoms                         | Healthy/Affected        |

**Additional Comments**  
\_\_\_\_\_