

**DNM2 Sequencing
Clinical Questionnaire**
Please complete and return with sample

Name: _____ DOB _____ Date _____

Male Female

Suspected Diagnosis: Autosomal dominant centronuclear myopathy
 Autosomal dominant intermediate Charcot-Marie-Tooth disease

Features	Present:	Yes	No
Growth			
Gestational Age	_____ wks		
Birth weight	_____ gms		
Birth length	_____ cm		
OFC	_____ cm		
Age at Exam	_____ yrs & mos		
Height	_____ cm		
Weight	_____ kg		
OFC	_____ cm		

Cognitive Delay Yes No

Severity _____

Motor Delay Yes No

Never walked Yes No

Age Sat _____

Age Walked _____

Age Talked _____

Neuromuscular findings

- | | | |
|---------------------------|--------------------------|--------------------------|
| Hypotonia | <input type="checkbox"/> | <input type="checkbox"/> |
| Proximal weakness | <input type="checkbox"/> | <input type="checkbox"/> |
| Distal weakness | <input type="checkbox"/> | <input type="checkbox"/> |
| Myotonia | <input type="checkbox"/> | <input type="checkbox"/> |
| Facial weakness | <input type="checkbox"/> | <input type="checkbox"/> |
| Reduced eye movements | <input type="checkbox"/> | <input type="checkbox"/> |
| Ptosis | <input type="checkbox"/> | <input type="checkbox"/> |
| Foot drop | <input type="checkbox"/> | <input type="checkbox"/> |
| Dysarthria | <input type="checkbox"/> | <input type="checkbox"/> |
| Distal sensory loss | <input type="checkbox"/> | <input type="checkbox"/> |
| Depressed tendon reflexes | <input type="checkbox"/> | <input type="checkbox"/> |
| High-arched feet | <input type="checkbox"/> | <input type="checkbox"/> |
| Weak ankle dorsiflexion | <input type="checkbox"/> | <input type="checkbox"/> |
| Thin distal muscles | <input type="checkbox"/> | <input type="checkbox"/> |

Nerve conduction velocity _____

Other neuromuscular findings:
(please describe)

Features	Present:	Yes	No
Muscle biopsy done			
Increased central nuclei	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Dystrophic processes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Inflammatory processes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Other findings on muscle biopsy: (please describe)			

Facial findings

High arched palate Yes No

Other facial findings
(please describe):

Cardiac findings: Yes No

(please describe):

Additional findings:

- | | | |
|---------------------|--------------------------|--------------------------|
| Scoliosis | <input type="checkbox"/> | <input type="checkbox"/> |
| Joint contractures | <input type="checkbox"/> | <input type="checkbox"/> |
| NG-tube | <input type="checkbox"/> | <input type="checkbox"/> |
| G-tube | <input type="checkbox"/> | <input type="checkbox"/> |
| Respiratory support | <input type="checkbox"/> | <input type="checkbox"/> |

Please list type of respiratory support:
(vent, c-pap, etc)

Other findings:
(please describe)

Ethnicity:

Family History:

LAB USE ONLY (Do not write in this box):

MTM1:

DNM2: