



Instructions: The accurate interpretation and reporting of genetic results is contingent upon the reason for referral, clinical information, ethnic background, and family history. To help provide the best possible service, supply the information requested below and send this paperwork with the specimen, or return by fax to Mayo Clinic Laboratories, Attn: Molecular Genetics Lab Genetic Counselors at 507-284-1759.

Patient Information

Form with fields for Patient Name (Last, First, Middle), Birth Date (mm-dd-yyyy), Sex Assigned at Birth (Male, Female, Unknown, Choose not to disclose), and Legal/Administrative Sex (Male, Female, Nonbinary).

Referring Provider Information

Form with fields for Referring Provider Name (Last, First), Phone, Fax\*, Genetic Counselor Name (Last, First), Phone, and Fax\*.

\*Fax number given must be from a fax machine that complies with applicable HIPAA regulations.

Reason for Testing

Form with fields for Study purpose (Diagnostic, Presymptomatic/Family history) and Working Clinical Diagnosis (describe).

Ethnic Background

Form with checkboxes for various ethnic backgrounds: African American, Asian, Latinx, Northern European, Ashkenazi Jewish, French Canadian, Mixed European, Southern European, Indigenous; indicate tribe, Other (specify), Choose not to disclose, Unknown, None of the above.

Family History

If testing is being performed for an asymptomatic individual due to a family history, note that pretest genetic counseling is strongly recommended.

Form with field: Are other relatives known to be affected? No Yes. If Yes, indicate their relationship to the patient and list their symptoms:

Form with field: Have other relatives had molecular genetic testing? No Yes. If Yes, complete the information below: Genes, Mutations, Name of individual tested, Birth date of individual tested, Laboratory at which testing was performed.

## Patient Information

Patient Name (Last, First, Middle)	Birth Date (mm-dd-yyyy)
------------------------------------	-------------------------

## Clinical Information

 Check all that apply or fax most recent clinic note.

<b>Autonomic</b>	<input type="checkbox"/> Abnormal sweating	<input type="checkbox"/> Dysphagia	<input type="checkbox"/> Shortness of breath		
	<input type="checkbox"/> Abnormal temperature regulation	<input type="checkbox"/> Orthostatic dizziness/fainting			
<b>Cardiac</b>	<input type="checkbox"/> Abnormal heart rate	<input type="checkbox"/> Arrhythmia	<input type="checkbox"/> Cardiomyopathy	<input type="checkbox"/> Palpitations	
<b>Cognitive</b>	<input type="checkbox"/> Behavioral changes	<input type="checkbox"/> Difficulty concentrating	<input type="checkbox"/> Intellectual disability	<input type="checkbox"/> Speech/Language difficulties	
	<input type="checkbox"/> Cognitive decline	<input type="checkbox"/> Developmental delay	<input type="checkbox"/> Memory loss		
<b>Craniofacial</b>	<input type="checkbox"/> Blindness	<input type="checkbox"/> Eye movement disorder	<input type="checkbox"/> Ptosis	<input type="checkbox"/> Visual impairment	
	<input type="checkbox"/> Cataracts	<input type="checkbox"/> Hearing loss	<input type="checkbox"/> Retinitis pigmentosa		
	<input type="checkbox"/> Dysmorphic features	<input type="checkbox"/> Optic atrophy	<input type="checkbox"/> Tinnitus		
<b>Endocrine</b>	<input type="checkbox"/> Abnormal parathyroid function, check one:	<input type="checkbox"/> Hypo	<input type="checkbox"/> Hyper		
	<input type="checkbox"/> Abnormal thyroid function, check one:	<input type="checkbox"/> Hypo	<input type="checkbox"/> Hyper		
	<input type="checkbox"/> Diabetes mellitus				
<b>GI</b>	<input type="checkbox"/> Chronic diarrhea	<input type="checkbox"/> Cyclic vomiting	<input type="checkbox"/> Incontinence		
	<input type="checkbox"/> Constipation	<input type="checkbox"/> Gastroparesis	<input type="checkbox"/> Loss of appetite		
<b>Muscular</b>	<input type="checkbox"/> Easy fatigue	<input type="checkbox"/> Hypotonia	<input type="checkbox"/> Muscle wasting	<input type="checkbox"/> Myalgia	
	<input type="checkbox"/> Hypertonia	<input type="checkbox"/> Muscle stiffness	<input type="checkbox"/> Muscle weakness	<input type="checkbox"/> Myotonia	
<b>Neurological</b>	<input type="checkbox"/> Abnormal balance	<input type="checkbox"/> Cerebellar atrophy	<input type="checkbox"/> Foot drop	<input type="checkbox"/> Paraplegia	<input type="checkbox"/> Rigidity
	<input type="checkbox"/> Ataxia	<input type="checkbox"/> Chorea	<input type="checkbox"/> Gait abnormality	<input type="checkbox"/> Paresthesia	<input type="checkbox"/> Strokes
	<input type="checkbox"/> Bradykinesia	<input type="checkbox"/> Dysarthria	<input type="checkbox"/> Hallucinations	<input type="checkbox"/> Poor coordination	<input type="checkbox"/> Tremor
	<input type="checkbox"/> Brain malformation	<input type="checkbox"/> Dystonia	<input type="checkbox"/> Pain	<input type="checkbox"/> Recurrent headaches	
	<input type="checkbox"/> Deep tendon, check one:	<input type="checkbox"/> Absent	<input type="checkbox"/> Increased	<input type="checkbox"/> Decreased	<input type="checkbox"/> Vertigo
	<input type="checkbox"/> Neuropathy, check one:	<input type="checkbox"/> Motor	<input type="checkbox"/> Sensory	<input type="checkbox"/> Sensorimotor	<input type="checkbox"/> Autonomic
	<input type="checkbox"/> Weakness, check one:	<input type="checkbox"/> Distal	<input type="checkbox"/> Proximal		
<b>Psychiatric</b>	<input type="checkbox"/> Mood changes	<input type="checkbox"/> Psychiatric disturbance/diagnosis	<input type="checkbox"/> Sleep disturbances		
<b>Seizures/Epilepsy</b>	<input type="checkbox"/> Absence seizures	<input type="checkbox"/> Febrile seizures	<input type="checkbox"/> Generalized seizures	<input type="checkbox"/> Myoclonus	
	<input type="checkbox"/> Epileptic encephalopathy	<input type="checkbox"/> Focal seizures	<input type="checkbox"/> Infantile/Epileptic spasms		
<b>Skeletal/Limb Abnormalities</b>	<input type="checkbox"/> Club foot	<input type="checkbox"/> Hammer toe	<input type="checkbox"/> Pes cavus	<input type="checkbox"/> Scoliosis	
	<input type="checkbox"/> Contractures	<input type="checkbox"/> Painless foot ulcers	<input type="checkbox"/> Pes planus		
<b>Other Manifestations</b>	<input type="checkbox"/> Other, specify:				

## At what age did symptoms present?

Has previous testing been performed for this patient?  No  Yes If Yes, complete information below.

- Sequencing for genes: \_\_\_\_\_
- Deletion/Duplication for genes: \_\_\_\_\_
- Electromyography/nerve conduction study (EMG/NCS); describe: \_\_\_\_\_
- Ulnar motor forearm nerve conduction velocity (m/s) and distal amplitude (mV) and/or R1 blink latency (ms): \_\_\_\_\_
  
- Imaging [eg, brain magnetic resonance imaging (MRI)]: \_\_\_\_\_
- Muscle biopsy; describe: \_\_\_\_\_
- Creatine kinase (CK) level; describe: \_\_\_\_\_