



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, Birth Date, Sex, Referring Provider Name, Phone, Fax, and Genetic Counselor Name.

*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 European Caucasian, African American, Hispanic, Asian, and Other specify.

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 for Are other relatives known to be affected? and instructions to list relationships and symptoms if Yes.

Form with field for Have other relatives had molecular genetic testing? and instructions to complete information below if Yes.

Patient Information

Patient Name <i>(Last, First, Middle)</i>	Birth Date <i>(mm-dd-yyyy)</i>
---	--------------------------------

Clinical Information Check all that apply or fax most recent clinic note.

Autonomic	<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	
Cardiac	<input type="checkbox"/> Abnormal heart rate	<input type="checkbox"/> Arrhythmia	<input type="checkbox"/> Cardiomyopathy
		<input type="checkbox"/> Palpitations	
Cognitive	<input type="checkbox"/> Behavioral changes	<input type="checkbox"/> Difficulty concentrating	<input type="checkbox"/> Intellectual disability
	<input type="checkbox"/> Cognitive decline	<input type="checkbox"/> Developmental delay	<input type="checkbox"/> Memory loss
Craniofacial	<input type="checkbox"/> Blindness	<input type="checkbox"/> Eye movement disorder	<input type="checkbox"/> Ptosis
	<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
Endocrine	<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		
GI	<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
Muscular	<input type="checkbox"/> Easy fatigue	<input type="checkbox"/> Hypotonia	<input type="checkbox"/> Muscle wasting
	<input type="checkbox"/> Hypertonia	<input type="checkbox"/> Muscle stiffness	<input type="checkbox"/> Myalgia
		<input type="checkbox"/> Muscle weakness	<input type="checkbox"/> Myotonia
Neurological	<input type="checkbox"/> Abnormal balance	<input type="checkbox"/> Chorea	<input type="checkbox"/> Gait abnormality
	<input type="checkbox"/> Ataxia	<input type="checkbox"/> Dysarthria	<input type="checkbox"/> Hallucinations
	<input type="checkbox"/> Brain malformation	<input type="checkbox"/> Dystonia	<input type="checkbox"/> Pain
	<input type="checkbox"/> Cerebellar atrophy	<input type="checkbox"/> Foot drop	<input type="checkbox"/> Paresthesia
	<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
Psychiatric	<input type="checkbox"/> Mood changes	<input type="checkbox"/> Psychiatric disturbance/diagnosis	<input type="checkbox"/> Sleep disturbances
Seizures/Epilepsy	<input type="checkbox"/> Absence seizures	<input type="checkbox"/> Febrile seizures	<input type="checkbox"/> Generalized seizures
	<input type="checkbox"/> Epileptic encephalopathy	<input type="checkbox"/> Focal seizures	<input type="checkbox"/> Infantile/Epileptic spasms
Skeletal/Limb Abnormalities	<input type="checkbox"/> Club foot	<input type="checkbox"/> Hammer toe	<input type="checkbox"/> Pes cavus
	<input type="checkbox"/> Contractures	<input type="checkbox"/> Painless foot ulcers	<input type="checkbox"/> Pes planus
Other Manifestations	<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: _____