

## Neurology-Associated Genetic Assays

<b>Test ID:</b>	<b>Published Name:</b>
AFTDP	Inherited Frontotemporal Dementia and Amyotrophic Lateral Sclerosis Gene Panel, Varies
ATAXP	Inherited Ataxia Gene Panel, Varies
CMSP	Inherited Congenital Myasthenic Syndrome Gene Panel, Varies
DMDZ	DMD Gene, Full Gene Analysis, Varies
DWPAN	Comprehensive Distal Weakness Gene Panel, Varies
EDMDP	Inherited Emery-Dreifuss Gene Panel, Varies
IMNP	Inherited Motor Neuropathy Gene Panel, Varies
IMSNP	Inherited Motor and Sensory Neuropathy Gene Panel, Varies
ISNP	Inherited Sensory Neuropathy Gene Panel, Varies
ISPP	Inherited Spastic Paraplegia Gene Panel, Varies
LGCMP	Inherited Limb-Girdle Muscular Dystrophy and Congenital Myasthenic Syndrome Gene Panel, Varies
MDYSP	Inherited Muscular Dystrophy Gene Panel, Varies
MNDP	Inherited Motor Neuron Disease Gene Panel, Varies
MUPAN	Comprehensive Neuromuscular Gene Panel, Varies
PARDP	Inherited Parkinson Disease Gene Panel, Varies
PEPAN	Comprehensive Peripheral Neuropathy Gene Panel, Varies
RABMP	Inherited Rhabdomyolysis and Metabolic Myopathy Panel, Varies
SEP9Z	SEPTIN9 Gene, Full Gene Analysis, Varies
SMCP	Inherited Skeletal Muscle Channelopathy Gene Panel, Varies
SOD1Z	SOD1 Gene, Full Gene Analysis, Varies

**Explanation:** On the effective date, the Specimen Required section for the assays listed above will be updated to exclude *any anticoagulant* for whole blood specimens. Additionally, new specimen types will now be accepted.

<b>Current Specimen Required</b>	<b>New Specimen Required</b>
<p><b>Patient Preparation:</b> A previous bone marrow transplant from an allogenic donor will interfere with testing. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.</p> <p><b>Specimen Type: Whole blood</b>  <b>Container/Tube:</b> Lavender top (EDTA) or yellow top (ACD)  <b>Acceptable:</b> Any anticoagulant  <b>Specimen Volume:</b> 3 mL  <b>Collection Instructions:</b>            1. Invert several times to mix blood.            2. Send specimen in original tube. <b>Do not aliquot.</b>  <b>Specimen Stability Information:</b> Ambient (preferred)/Refrigerated</p>	<p><b>Patient Preparation:</b> A previous bone marrow transplant from an allogenic donor will interfere with testing. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.</p> <p><b>Submit only 1 of the following specimens:</b></p> <p><b>Specimen Type: Whole blood</b>  <b>Container/Tube:</b>  <b>Preferred:</b> Lavender top (EDTA) or yellow top (ACD)  <b>Acceptable:</b> None  <b>Specimen Volume:</b> 3 mL  <b>Collection Instructions:</b>            1. Invert several times to mix blood.            2. Send whole blood specimen in original tube. <b>Do not aliquot.</b></p>

**Additional Information:** To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted. Testing may be canceled if DNA requirements are inadequate.

**Specimen Stability Information:** Ambient (preferred) 4 days/Refrigerated 4 days/Frozen 4 days

**Additional Information:**

1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed.
2. To ensure minimum volume and concentration of DNA is met, the requested volume must be submitted. Testing may be canceled if DNA requirements are inadequate.

**Specimen Type: Saliva**

**Patient Preparation:** Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.

**Supplies:** Saliva Swab Collection Kit (T786)

**Specimen Volume:** 1 Swab

**Collection Instructions:** Collect and send specimen per kit instructions.

**Specimen Stability Information:** Ambient (preferred) 30 days/Refrigerated 30 days

**Additional information:** Saliva specimens are acceptable but not recommended. Due to lower quantity/quality of DNA yielded from saliva, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When applicable, specific gene regions that were unable to be interrogated will be noted in the report. Alternatively, additional specimen may be required to complete testing.

**Specimen Type: Extracted DNA**

**Container/Tube:**

**Preferred:** Screw Cap Micro Tube, 2mL with skirted conical base

**Acceptable:** Matrix tube, 1mL

**Collection Instructions:**

1. The preferred volume is at least 100 mcL at a concentration of 75 ng/mcL.
2. Include concentration and volume on tube.

**Specimen Stability Information:** Frozen (preferred) 1 year/Ambient/Refrigerated

**Additional Information:** DNA must be extracted in a CLIA-certified laboratory or equivalent and must be extracted from a specimen type listed as acceptable for this test (including applicable anticoagulants). Our laboratory has experience with Chemagic, Puregene, Autopure, MagnaPure, and EZ1 extraction platforms and cannot guarantee that all extraction methods are compatible with this test. If testing fails, one repeat will be attempted, and if unsuccessful, the test will be reported as failed and a charge will be applied. If applicable, specific gene regions that were unable to be interrogated due to DNA quality will be noted in the report.

## Questions

Contact Michelle Raths, Laboratory Resource Coordinator at 800-533-1710.