
Reporting Title: Ehlers-Danlos Syndrome Gene Panel**Performing Location:** Rochester**Ordering Guidance:**

Customization of this panel and single gene analysis for any gene present on this panel are available. For more information see CGPH/ Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies.

Targeted testing for familial variants (also called site-specific or known mutations testing) is available for the genes on this panel. See FMTT / Familial Variant, Targeted Testing, Varies. To obtain more information about this testing option, call 800-533-1710.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Necessary Information:

Prior Authorization is available, but not required, for this test. If proceeding with the prior authorization process, submit the required form with the specimen.

Specimen Requirements:

Patient Preparation: 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

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant

Specimen Volume: 3 mL

Collection Instructions:

1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. Do not aliquot.

Specimen Stability Information: Ambient (preferred)/Refrigerated

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 30 days

Additional Information: Due to lower concentration of DNA yielded from saliva, it is possible that additional specimen may be required to complete testing.

Specimen Minimum Volume:

1 mL

Forms:

1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available:
 - Informed Consent for Genetic Testing (T576)
 - Informed Consent for Genetic Testing (Spanish) (T826)
2. Connective Tissue/Cerebrovascular Disease Genetic Testing Patient Information
3. Ehlers-Danlos Syndrome Gene Panel (EDSGG) Prior Authorization Ordering Instructions
4. If not ordering electronically, complete, print, and send a Cardiovascular Test Request Form (T724) with the specimen.

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
617254	Test Description	Alphanumeric		62364-5
617255	Specimen	Alphanumeric		31208-2
617256	Source	Alphanumeric		31208-2
617257	Result Summary	Alphanumeric		50397-9
617258	Result	Alphanumeric		82939-0
617259	Interpretation	Alphanumeric		69047-9
617260	Additional Results	Alphanumeric		82939-0
617261	Resources	Alphanumeric		99622-3
617262	Additional Information	Alphanumeric		48767-8
617263	Method	Alphanumeric		85069-3
617264	Genes Analyzed	Alphanumeric		48018-6
617265	Disclaimer	Alphanumeric		62364-5
617266	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:
Supplemental**CPT Code Information:**

81408 x2
81479
81479 (if appropriate for government payers)

Reference Values:

An interpretive report will be provided.