

Test Definition: EDSGG

Ehlers-Danlos Syndrome Gene Panel, Varies

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:

<u>Prior Authorization</u> 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.

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



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- 3. Ehlers-Danlos Syndrome Gene Panel (EDSGG) Prior Authorization Ordering Instructions
- 4. <u>If not ordering electronically, complete, print, and send a Cardiovascular Test Request Form</u> (T724) with the specimen.

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Туре	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.