
Lab Dept: Anatomic Pathology

Test Name: EHLERS-DANLOS (EDS) SYNDROME TYPE I/II
DELETION/DUPLICATION 1st GENE

General Information

Lab Order Codes: EDSD1

Synonyms: COL5A1; COL5A2; EDS I/II Deletion/Duplication

CPT Codes: 81479 – Molecular Pathology, Unlisted Procedure

Test Includes: High Density Targeted Array of either COL5A1 or COL5A2 gene.

Logistics

Test Indications: Ehlers-Danlos syndrome (EDS) is a connective tissue disorder composed of numerous subtypes with distinct genetic and clinical findings. In general, EDS is characterized by joint hypermobility, skin hyperextensibility and tissue fragility.

The classical type of EDS includes EDS I and EDS II. They are autosomal dominant inherited disorders, and considered the most common types of EDS. Findings in patients with EDS I (gravis type) include marked skin involvement (hyperextensibility, atrophic scars) generalized joint hyperhypermobility and complications there of (dislocations, subluxations, pes planus), muscular hypotonia, and delayed motor development. Additional findings may also include rupture of the aorta or bowel. Prematurity is also common

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: CTGT (CTGT Test: 1131)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 2 - 3 weeks

Special Instructions: Request special forms for patient consent, physician signature and clinical history from Children's Laboratory or print [Request form](#) here. Select (HDT Array) and specify gene COL5A1 or COL5A2.

Note: These completed forms must accompany the specimen or come

with the patient when they present at the lab to be drawn.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube
Draw Volume:	6 mL (Minimum: 3 mL) blood
Processed Volume:	Same as Draw Volume
Collection:	Routine blood collection
Special Processing:	Lab Staff: Do Not centrifuge. Specimen should be sent in original collection container. Store and ship at refrigerated temperatures. Forward promptly. Note: For specimens collected Friday – Sunday (or on a holiday), they should be held in Children’s Laboratories at refrigerated temperatures and shipped on Monday (or the next business day Monday – Thursday).
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimen; inappropriate specimen type; frozen specimens

Interpretive

Reference Range:	No mutations detected
Critical Values:	N/A
Limitations:	N/A
Methodology:	High Density Targeted Array
References:	Connective Tissue Gene Tests February 2018 (484) 224-2900 Fax (484) 244-2904
Updates:	2/5/2013: CPT update 7/10/2013: CPT update, previously listed as 81407