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**Lab Dept:** Anatomic Pathology

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

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***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.

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***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

**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.

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### ***Specimen***

<b>Specimen Type:</b>	Whole blood
<b>Container:</b>	Lavender top (EDTA) tube
<b>Draw Volume:</b>	6 mL (Minimum: 4 mL) blood
<b>Processed Volume:</b>	Same as Draw Volume
<b>Collection:</b>	Routine venipuncture
<b>Special Processing:</b>	Lab Staff: <b>Do Not</b> 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).
<b>Patient Preparation:</b>	None
<b>Sample Rejection:</b>	Mislabeled or unlabeled specimen; inappropriate specimen type; frozen specimens

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### ***Interpretive***

<b>Reference Range:</b>	No mutations detected
<b>Critical Values:</b>	N/A
<b>Limitations:</b>	N/A
<b>Methodology:</b>	High Density Targeted Array
<b>References:</b>	<a href="#">Connective Tissue Gene Tests</a> March 2012 (484) 224-2900 Fax (484) 244-2904
<b>Updates:</b>	2/5/2013: CPT update 7/10/2013: CPT update, previously listed as 81407