Lab Dept: **Anatomic Pathology**

**Test Name:** EHLERS-DANLOS (EDS) TYPE IV DELETION/DUPLICATION

### General Information

**Lab Order Codes:** EDS4D

**Synonyms:** COL3A1; EDS IV Deletion/Duplication

**CPT Codes:** 81479 – Molecular Pathology unlisted procedure

**Test Includes:** High Density Targeted Array of the COL3A1 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 vascular type, EDS IV, represents the most severe form of the disorder. Patients frequently suffer rupture of the arteries and intestines. The vascular type is an autosomal dominant disorder caused by mutations in COL3A1.

**Lab Testing Sections:** Anatomic Pathology - Sendouts

**Referred to:** CTGT (CTGT Test: 1136)

**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) COL3A1.

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