
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

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: 4 mL) blood
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture
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 March 2012 (484) 224-2900 Fax (484) 244-2904
Updates:	2/5/2013: CPT update