Lab Dept:	**Anatomic Pathology**
Test Name:	**EHLERS-DANLOS (EDS) TYPE I/11 SEQUENCING**

General Information

Lab Order Codes:	ED12
Synonyms:	COL5A1; COL5A2; EDSI/II Sequencing
CPT Codes:	81479 x2 – Molecular Pathology, Unlisted Procedure
Test Includes:	Sequencing of COL5A1 and COL5A2 genes.

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 EDS II (mitis type) are similar to, but less severe than, EDS I. The skin is less involved and joint laxity may be confined to hands and feet. Prematurity is not a feature. About 50% of the cases of classical EDS are caused by mutations in COL5A1 or COL5A2.
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.

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 7/10/2013: CPT update, previously listed as 81408.