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**Lab Dept:**                   **Anatomic Pathology**

**Test Name:**               **TGFBR1 GENE SEQUENCING**

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***General Information***

**Lab Order Codes:**       TGF

**Synonyms:**               Aortic aneurysm, familial thoracic 5 (AAT5) TGFBR1;Loeys-Dietz syndrome, type 1A (LDS1A) TGFBR1

**CPT Codes:**               81405 – Molecular Pathology procedure, Level 6

**Test Includes:**           Direct DNA sequencing of PCR products generated from genomic DNA, sequencing of exons and exon-intron boundaries, point mutations, splice site mutations and small exonic deletions, insertions and indels.

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***Logistics***

**Test Indications:**       For confirmation of symptoms and the clinical diagnosis related to Aortic aneurysm, familial thoracic 5 (AAT5) TGFBR1 or Loeys-Dietz syndrome, type 1A (LDS1A) TGFBR1.

AAT5 and AAT3 have been linked to mutations in the transforming growth factor Beta receptor type I and II genes (TGFBR1 and TGFBR2). Patients may have aneurysms of the aorta and other arteries. TGFBR2 mutations are currently estimated to be responsible for 5% of familial thoracic aortic aneurysms and dissections (TAAD).

Loeys-Dietz syndrome (LDS) is a recently described syndrome caused by mutations in one of two genes, TGFBR1 (Type 1A) or TGFBR2 (Type 1B). The reported phenotype is highly variable and overlaps considerably with Ehlers-Danlos syndrome IV or Marfan syndrome. A high percentage of patients have aortic root aneurysm or other arterial aneurysms. Some have cardiac abnormalities including patent ductus or atrial septal defects. Many display arterial tortuosity. Skeletal defects may include hypertelorism, pectus defects, joint laxity, craniosynostosis, arachnodactyly, scoliosis, talipes equinovarus, camptodactyly and malar hypoplasia. Some have dural ectasia. Additional features may include uterine, spleen or bowel rupture, thin, translucent, hyperextensible or velvety skin with atrophic scars and easy bruising. Blue sclera and a bifid uvula have also been observed.

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

**Referred to:**             Connective Tissue Gene Tests (CTGT)

**Phone Numbers:**       MIN Lab: 612-813-6280

STP Lab: 651-220-6550

<b>Test Availability:</b>	Daily, 24 hours
<b>Turnaround Time:</b>	1 – 2 weeks
<b>Special Instructions:</b>	Please include a completed CTGT <a href="#">Request form</a> with the patient or specimen to the laboratory.

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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: 3 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. Send via overnight shipping with a cold pack to reach CTGT Monday through Friday. If weekend or holiday when drawn, store at refrigerated temperatures. Please include a <a href="#">CTGT Shipment Packing Slip</a> with the shipment.
<b>Patient Preparation:</b>	None
<b>Sample Rejection:</b>	Mislabeled or unlabeled specimen

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### ***Interpretive***

<b>Reference Range:</b>	Interpretive report
<b>Critical Values:</b>	N/A
<b>Limitations:</b>	Published estimates of test sensitivity for genes linked to certain disorders can be very inaccurate, and that it is difficult to predict the probability of detecting a mutation in any single gene for one individual. The following factors contribute to this challenge: Many disorders have overlapping phenotypes; some disorders are linked to mutations in more than one gene; in some instances genes remain to be linked to specific disorders; for most disorders, proper diagnosis requires that clinical findings are considered along with genetic findings.
<b>Methodology:</b>	DNA sequencing

**References:**

[Connective Tissue Gene Tests](#) December 2014  
(484) 224-2900 Fax (484) 244-2904

**Updates:**

3/1/2011: CPT update  
2/11/2013: CPT update