Lab Dept: Anatomic Pathology

Test Name: TGFBR1 &TGFBR2 GENE SEQUENCING

General Information

Lab Order Codes: LDAB

Synonyms: Loeys-Dietz syndrome, Type 1A (LDS1A) TGFBR1; Loeys-Dietz syndrome, Type 1B (LDS1B); Loeys-Dietz Types 1A/1B; LDS Types 1A/1B

CPT Codes: 81405 x2 – 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.

Logistics

Test Indications: For confirmation of symptoms and the clinical diagnosis related to Loeys-Dietz syndrome, type 1A (LDS1A) TGFBR1 and Loeys-Dietz syndrome, type 1B (LDS1B) TGFBR2.

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

Test Availability: Daily, 24 hours

Turnaround Time: 1 – 2 weeks

Special Instructions:	Please include a completed CTGT Request form with the patient or specimen to the laboratory.

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 venipuncture
Special Processing:	Lab Staff: **Do Not** centrifuge. Specimen should be sent in original collection container. Ship at ambient temperature. Send via overnight shipping to reach CTGT Monday through Friday. If weekend or holiday when drawn, store at refrigerated temperatures. Please include a CTGT Shipment Packing Slip with the shipment.
Patient Preparation:	None
Sample Rejection:	Mislabled or unlabeled specimen

Interpretive

Reference Range:	Interpretive report
Critical Values:	N/A
Limitations:	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.
Methodology:	DNA sequencing
References:	Connective Tissue Gene Tests June 2011 (484) 224-2900 Fax (484) 244-2904
Updates:	2/11/2013: CPT update 7/9/2013: CPT update CTGT, previously listed as 81406