ClinGenLab
Clinical Testing Laboratory, Inc.

Clinical Testing Lab of Washington
2150 Pennsylvania Avenue NW
Washington, DC 20037
Phone: 202-555-1212

Specimen Number	Specimen Type	Control Number	Account Number	Account Phone Number	Route
	Peripheral Blood				

Patient Last Name	Patient Barcode

Patient First Name	Patient Middle Name
Jonathan	

Patient SS#	Patient Phone	Total Volume

Age (Y/M/D)	Date of Birth	Sex	Fasting
35 y.o.		Male	

Patient Address	Indication / Family History / Ethnicity
	Indication: Thoracic aortic dissection, with clinical features and morphology suggestive of Marfan Syndrome **Family History: No known family history** **Ethnicity: Western European Caucasian**

Date and Time Collected	Date Entered	Date and Time Reported	Physician Name	NPI	Physician ID
			Jane Ferreiro, MD		

Tests Ordered
Marfan/TAAD Sequencing Panel

General Comments
Please send a copy of the final report to the Molecular Science/M1 Training office via Fax at (202) 555-1212

Clinical test results for Marfan/TAAD

9 conditions tested:

- Thoracic aortic aneurysm and aortic dissection (TAAD)
- Arterial tortuosity syndrome (ATS)
- Congenital contractural arachnodactyly (CCA)
- Ehlers-Danlos syndrome (EDS)
- Homocystinuria
- Loeys-Dietz syndrome (LDS)
- Marfan syndrome (MFS)
- Shprintzen-Goldberg syndrome (SGS)
- X-linked mental retardation with marfanoid habitus syndrome

GENE	TEST RESULTS	EXPLANATION
FBN1	Arg1790 Arg1790Ter	**This result confirms the diagnosis of or predisposition for Marfan Syndrome (MFS)**. This result should be interpreted in the context of clinical presentation and results of other laboratory tests. A PCR/sequencing study has confirmed one copy of the Arg1790Ter (FBN1: g.194098C>T, c.5368C>T or p.Arg1790Ter) variation. The Arg1790Ter mutation is caused by a C to T change at nucleotide position 194098 in the FBN1 gene and results in a change from an arginine to a stop (termination) codon at position 1790. This causes a premature termination of translation and produces an abnormally shortened protein. This individual's result has important implications for other family members. Clinical and laboratory evaluations should be considered for at risk individuals. Genetic counseling is recommended for at risk individuals.
ACTA2 MYH11 CBS SKI COL3A1 SLC2A10 COL5A1 SLC2A COL5A2 SMAD3 FBN2 TGFB2 FLNA TGFB1 MED12 TGFBR1	Negative	No known pathogenic variant detected in these genes

DISCLAIMER:

Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered.

ASSAY METHODS

Full-Gene Sequencing covers the full gene coding sequence, +/- 10 base pairs of adjacent intronic sequence, and other non-coding sequence positions containing select known pathogenic variants. Deletion/Duplication Analysis detects most intragenic deletions and duplications at single exon resolution. Rarely however, single-exon duplication events may be missed due to inherent sequence properties or isolated reduction in data quality.

CLINICAL DESCRIPTION

Familial thoracic aortic aneurysm and dissection (familial TAAD) involves problems with the aorta, which is the large blood vessel that distributes blood from the heart to the rest of the body. Familial TAAD affects the upper part of the aorta, near the heart. This part of the aorta is called the thoracic aorta because it is located in the chest (thorax). Other vessels that carry blood from the heart to the rest of the body (arteries) can also be affected. In familial TAAD, the aorta can become weakened and stretched (aortic dilatation), which can lead to a bulge in the blood vessel wall (an aneurysm). Aortic dilatation may also lead to a sudden tearing of the layers in the aorta wall (aortic dissection), allowing blood to flow abnormally between the layers. These aortic abnormalities are potentially life-threatening because they can decrease blood flow to other parts of the body such as the brain or other vital organs, or cause the aorta to break open (rupture).The occurrence and timing of these aortic abnormalities vary, even within the same affected family. They can begin in childhood or not occur until late in life. Aortic dilatation is generally the first feature of familial TAAD to develop, although in some affected individuals dissection occurs with little or no aortic dilatation. Aortic aneurysms usually have no symptoms. However, depending on the size, growth rate, and location of these abnormalities, they can cause pain in the jaw, neck, chest, or back; swelling in the arms, neck, or head; difficult or painful swallowing; hoarseness; shortness of breath; wheezing; a chronic cough; or coughing up blood. Aortic dissections usually cause severe, sudden chest or back pain, and may also result in unusually pale skin (pallor), a very faint pulse, numbness or tingling (paresthesias) in one or more limbs, or paralysis. Familial TAAD may not be associated with other signs and symptoms. However, some individuals in affected families show mild features of related conditions called Marfan syndrome or Loeys-Dietz syndrome. These features include tall stature, stretch marks on the skin, an unusually large range of joint movement (joint hypermobility), and either a sunken or protruding chest. Occasionally, people with familial TAAD develop aneurysms in the brain or in the section of the aorta located in the abdomen (abdominal aorta). Some people with familial TAAD have heart abnormalities that are present from birth (congenital). Affected individuals may also have a soft out-pouching in the lower abdomen (inguinal hernia), an abnormal curvature of the spine (scoliosis), or a purplish skin discoloration (livedo reticularis) caused by abnormalities in the tiny blood vessels of the skin (dermal capillaries). However, these conditions are also common in the general population. Depending on the genetic cause of familial TAAD in particular families, they may have an increased risk of developing blockages in smaller arteries, which can lead to heart attack and stroke.

-from GHR