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

**Test Name:**                **FBN1 DELETION/DUPLICATION HDT ARRAY**

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***General Information***

**Lab Order Codes:**        FBDD

**Synonyms:**                Ectopia lentis FBN1; Marfan syndrome, type I (MFS1) FBN1; Weill-Marchesani syndrome (WMS), autosomal dominant FBN1

**CPT Codes:**                81479 – Molecular Pathology, Unlisted Procedure

**Test Includes:**            Deletion/Duplication High-Density Targeted (HDT) Array uses 60mer oligonucleotide sequences designed to selectively complement target areas with an extremely high degree of specificity. High resolution and sensitivity are achieved by assigning a number of probes to each exonic region to allow for a minimum of 3 probes within each 300-500 base section of the genomic DNA sequence. Non-coding intervening sequences are targeted at a minimum with approximately half of the density used for exonic regions. Many intervening sequences are more densely covered. The minimum CNV size detected by this high-density array is 300-500 nucleotides, which is the technical limit of the assay using these stringent parameters.

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***Logistics***

**Test Indications:**        Fibrillin-1 is the major structural component of the microfibrils that link together the various extracellular matrix components in most connective tissues, thus providing support for the organs. Microfibrils can also associate with elastin, forming elastic fibers that provide resilience and elasticity in tissues. Defects in the FBN1 gene compromise the fibrillin-1 function in these tissues and therefore, result in connective tissue weakness as seen in Marfan syndrome.

Ectopia lentis, isolated is a dominantly inherited disorder characterized by congenital lens dislocation. The lens may be dislocated in any direction. Ectopia lentis, isolated should be discriminated from the ectopia lentis associated with homocystinuria due to cystathionine beta-synthase deficiency. The latter disorder typically displays a downward dislocation of the lens. In some instances, identical FBN1 mutations have been described in both ectopia lentis, isolated and Marfan syndrome.

Weill-Marchesani syndrome, autosomal recessive is characterized by short stature, joint stiffness, brachydactyly, and several eye abnormalities including ectopia lentis, severe myopia, glaucoma and microspherophakia. Autosomal recessive WMS is caused by homozygous or compound heterozygous mutations in the ADAMTS10 gene which codes for a disintegrin-like and metalloproteinase with thrombospondin type 1 motif, 10. Weill-Marchesani syndrome, autosomal dominant is phenotypically similar to the recessive form. The autosomal dominant form is caused by mutations in the FBN1 gene, which codes for fibrillin 1.

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

**Referred to:** Connective Tissue Gene Tests (CTGT Test: 1188)

**Phone Numbers:** MIN Lab: 612-813-6280  
STP Lab: 651-220-6550

**Test Availability:** 24 hours

**Turnaround Time:** 2 weeks

**Special Instructions:** No transfusion within the past 30 days. Please include a completed CTGT [Request form](#) with the patient or specimen to the laboratory.

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### ***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 blood collection, mix specimen by gentle inversion

**Special Processing:** Lab Staff: **Do Not** 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 [CTGT Shipment Packing Slip](#) with the shipment.

**Patient Preparation:** None

**Sample Rejection:** Mislabeled or unlabeled specimens

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### ***Interpretive***

**Reference Range:** Interpretive report

**Critical Values:** N/A

**Limitations:** Partial exon deletion/duplications may not be detected unless they are located within the amplified region of the exon.

**Methodology:** High-Density Targeted Array

**References:**

[Connective Tissue Gene Tests](#) January 2018  
Phone (484) 224-2900 Fax (484) 244-2904

**Updates:**

2/5/2013: CPT update  
7/10/2013: CPT update CTGT, previously listed as 81407