Thoracic Aortic Aneurysm Panel

The Thoracic Aortic Aneurysm Panel offers Next Generation Sequencing of 14 genes associated with syndromic and non-syndromic forms of thoracic aortic aneurysm and dissections (TAAD). TAAD is characterized by aneurysm and dissection primarily of the thoracic aorta, but can involve other arteries including abdominal aortic aneurysms, cerebral aneurysms, and peripheral artery aneurysms. Even in the absence of obvious connective tissue disease, thoracic aortic aneurysms are often associated with genetic/familial predisposition. Most commonly, Familial TAAD is associated with dilation, aneurysm, and/or dissection of the ascending aorta. This is typically associated with an autosomal dominant inheritance pattern with reduced penetrance. ACTA2 is the gene most commonly implicated in Familial TAAD, accounting for up to 14% of cases. In total, the genetic basis of Familial TAAD is identifiable in approximately 20% of cases.

Several syndromic conditions are known to be associated with TAAD. This panel includes genes associated with connective tissue related disorders including Marfan syndrome, Loeys-Dietz syndrome, Ehlers-Danlos syndrome Type IV (Vascular Type EDS), Arterial Tortuosity syndrome, Shprintzen-Goldberg syndrome, Aneurysm-Osteoarthritis syndrome, Congenital Contractural Arachnodactyly and Homocystinuria. In addition to TAAD, additional cardiac features of these conditions can include arterial tortuosity, mitral valve prolapse, and congenital heart defects. Non-cardiac features may include skeletal anomalies, craniofacial manifestations, cutaneous findings, and in some instances, developmental delay.

TAA Panel Includes:
- ACTA2
- FLNA
- SMAD3
- CBS
- MYH11
- TGF2
- COL3A1
- MYLK
- TGFBR1
- FBN1
- SKI
- TGFBR2
- FBN2
- SLC2A10
Methodology:

Next Generation Sequencing: All coding exons, as well as their flanking regions, of the genes listed in the panel are enriched from the patient’s genomic DNA and sequenced using a solid-state sequencing-by-synthesis process. DNA sequences are assembled and compared to the published genomic reference sequences in Genome Reference Consortium Build 37. Dideoxy DNA sequencing is used to provide data for bases with insufficient coverage and to confirm the reported variants from next-generation sequencing. This assay does not detect variants in the promoter regions, deep intronic regions, or other regulatory elements, and does not detect large deletions or mosaicisms. Variants are reported according to HGVS nomenclature (www.hgvs.org/mutnomen).

Validation testing indicates an analytic sensitivity of greater than 99% and an analytic specificity of 100%.

References:


Specimen:

Peripheral blood in EDTA tube
Adult: 5-10mL
Child: 3-5mL
Infant: 1-3mL
For other specimen types, please contact us at 513-636-4474

Turnaround Time:

Full Panel Analysis 8-10 weeks
Known Mutation Analysis 1-2 weeks

CPT Codes:

Full Panel Analysis: 81410
Additional Family Members: 81403