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
- CBS
- COL3A1
- FBN1
- FBN2
- FLNA
- MYH11
- MYLK
- SLC2A10
- SMAD3
- TGFB2
- TGFBR1
- TGFBR2

Please visit our website for more information on specific tests, how to request testing, submit samples, cost, and turnaround time: www.cincinnatichildrens.org/heartdx
Methodology:

Sensitivity & Accuracy:

References:

Specimen:

Peripheral blood in EDTA tube
Adult: 5-10mL
Child: 3-5mL
Infant: 1-3mL
For other specimen types, please contact Amy Shikany at 513-803-3317

Turnaround Time:

Full Mutation Analysis 2-4 weeks
Known Mutation Analysis 1-2 weeks

CPT Codes:

Full Gene Sequencing
Additional Family Members