

# Familial Thoracic Aortic Aneurysm and Dissection (TAAD)

Familial thoracic aortic aneurysm and dissection (TAAD) is an autosomal dominant inherited disease affecting the arteries. Variants have been discovered in 20% of TAAD cases. Genetic analysis is generally limited to the ACTA2 gene, which shows a 14% positivity rate. The MHI Molecular Diagnosis Laboratory reports only this gene for the TAAD profile. If no variant is identified and if the clinical situation warrants it, the Molecular Diagnosis Laboratory will then analyze the FBN1, SMAD3, TGFB2, TGFB3, TGFBR1 and TGFBR2 genes in second line and the MYH11, MYLK and SLC2A10 genes in third line.

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## Tested Genes:

First line	Second line	Third line
ACTA2	FBN1 SMAD3 TGFB2 TGFB3 TGFBR1 TGFBR2	MYH11 MYLK SLC2A10