



GEN08 - Exome sequencing in cardiovascular phenotypes resulting from rare variants

OBJECTIVE: Use exome sequencing to identify causative genes for genetically triggered thoracic aortic disease.

ORGANIZATION

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Samples • Genetic Material

Data • Demographic and clinical data

CONCLUSIONS

Results: • *Results pending*

BACKGROUND AND RATIONALE

Non-syndromic forms of familial thoracic aortic aneurysms and dissections (TAAD) demonstrate significant genetic and clinical heterogeneity. Although positional cloning and candidate gene sequencing have successfully identified causative genes for this condition, the progress has been slow. Exome sequencing has the potential to rapidly identify causative genes for this disease. 250 – 300 familial TAAD patients with mutations in known genes excluded will undergo exome sequencing for the discovery phase, including subjects from GenTAC and other cohorts.

DESIGN

Hypothesis: • Exome sequencing will identify causative genes for genetically triggered thoracic aortic disease.

Inclusion criteria: • Confirmed thoracic aortic disease.
• Patients who are determine to NOT have mutations in known genes by clinical studies or resequencing
• Familial thoracic aortic aneurysm and/or dissection
• BAV/TAAD
• Patients presenting with TAAD under the age of 50 years.

