



GEN01 - Sequencing of Known Genes for Thoracic Aortic Aneurysms and Dissection in the GENTAC Cohort

OBJECTIVE: The majority of patients in the GenTAC cohort do not have identified mutations in the six genes known to cause TAAD. Sequencing of the known genes in the first 500 patients with TAAD will be completed to identify mutations in the known genes leading to TAAD.

ORGANIZATION

Lead Investigator: Diana Milewicz, MD, PhD

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Funding Source: GenTAC

CONCLUSIONS

Results:

- The results from this resequencing has spurred additional research described in other proposals (e.g., GEN03, GEN08, GEN09, GEN16). Please refer to these proposal for more information

BACKGROUND AND RATIONALE

Thoracic aortic aneurysms predispose patients to acute aortic dissections, which are catastrophic events that often cause sudden death (TAAD). Mutations in single genes can lead to a predisposition to TAAD inherited as part of a syndrome, such as Marfan syndrome (MFS), Loeys-Dietz syndrome (LDS) or vascular Ehlers-Danlos syndrome (vascular EDS). The majority of patients with TAAD do not have an identified syndrome but approximately 20% of these patients have a family history of TAAD, indicating a strong genetic component to non-syndromic TAAD

DESIGN

Inclusion criteria:

- First 500 subjects enrolled into the GenTAC Registry with a known TAAD.

Exclusion criteria:

- Patients with Turner syndrome (45,XO)
- Patients with an identified mutation in a known gene.
- Relatives of individuals already included in the resequencing samples.

Samples:

- Genetic material

Data:

- None

