



GEN34 - The Turner Syndrome aortopathy in the context of other genetically triggered aortic diseases

OBJECTIVE: To investigate and better understand the aortopathy of Turner Syndrome within the context of other genetically-triggered aortic diseases.

ORGANIZATION

Lead Investigator: Michael Silberbach, MD

Co-Investigators: Cheryl Maslen PhD, Kathy Holmes MD

Funding Source: GenTAC

CONCLUSIONS

Results: • *Results pending*

BACKGROUND AND RATIONALE

Aortic dissection occurs in approximately 1-2 % of individuals with Turner syndrome. The aortopathy of Turner syndrome (TS) has never been viewed from the perspective of other syndromes and genetic conditions where aortic aneurysm and dissection are likely to occur. In particular no one has ever compared the TS individual with BAV and BAV in non-TS. We propose to mine the rich clinical data that comprises the GenTAC in order to understand the aortopathy of TS within the context of other genetically-triggered aortic diseases.

DESIGN

- Hypothesis:*
- TS is an incremental risk factor for aortic dilation in the setting of BAV.
 - The clinical characteristics and medical/surgical treatment of the TS population within GenTAC are different than other GenTAC diagnostic subgroups.
- Inclusion criteria:*
- Subjects enrolled into GenTAC with Turner Syndrome, BAV, and TAAD.
- Exclusion criteria:*
- Subjects with Marfan syndrome, Ehlers-Danlos, genetic mutations and Loeys-Dietz
- Samples:*
- None
- Data:*
- Surgical, imaging and genetic data. Organ system review and medication use.

