



GEN20 – Analysis of left ventricular size and systolic function in patients with Marfan syndrome and other genetically triggered thoracic aortic aneurysms

OBJECTIVE: To assess the presence of left ventricular size and systolic function in patients with different forms of genetically triggered thoracic aneurysms in the entire GenTAC population and to perform analysis

ORGANIZATION

Lead Investigator: William J. Ravekes MD

Co-Investigators: Kathryn Holmes MD, Jonathan Weinsaft MD, Michael Silberbach MD, Richard Devereux MD

Funding Source: GenTAC

BACKGROUND AND RATIONALE

Numerous prior studies have looked at left ventricular dimension and function in patients with Marfan syndrome. Patients with significant aortic and/or mitral valve regurgitation have been reported to have left ventricular dilation and decreased systolic function. However, there are conflicting reports as to whether there is impaired left ventricular function in patients without significant valvular regurgitation.

DESIGN

- Hypothesis:*
- Larger left ventricular cavity size (in SDs above the expected value for age and body surface area) is positively associated with the presence of $\geq 1+$ and $\geq 2+$ aortic regurgitation and/or $\geq 1+$ and $\geq 2+$ mitral regurgitation.
 - Decreased left ventricular systolic function (in SDs from the expected value for age) remains positively associated with the presence of left ventricular dilation.
 - While controlling for the degree of valve disease, the degree of left ventricular dilation and changes in systolic function will vary based on the subset genetically triggered thoracic aneurysms, Marfan syndrome vs. Loeys-Dietz, familial bicuspid aortic valve.

- Inclusion criteria:*
- The cardiac disease in patients with MFS correlates with age of diagnosis. Patients diagnosed earlier in life tend to have increased incidence of AV valve regurgitation, increased LVED and decreased ventricular function compared to older individuals.

- Exclusion criteria:*
- Subjects who are ≤ 50 years old and diagnosed with Marfan, LDS, BAV or FTAAD for whom imaging data is available.

- Samples Data:*
- Prior history of surgery
 - Prior history of myocardial
 - Prior history of aortic stenosis

- None
- Clinical evaluation and imaging data.

CONCLUSIONS

- Results:*
- *Results pending*