



GEN43- Association of bicuspid aortic valve with Marfan syndrome or Loeys Dietz: two strikes?

OBJECTIVE: To evaluate if dual diagnosis of BAV and either Marfan syndrome or Loeys Dietz effects outcome.

ORGANIZATION

Lead Kathy Holmes, MD

Investigator:

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Funding Source: GenTAC

Samples:

- None

Data:

- Demographic, genetic, clinical, imaging and surgical data.

BACKGROUND AND RATIONALE

Patients with Marfan syndrome (MS) and Loeys-Dietz (LDS) are known to have a prevalence of bicuspid aortic valve (BAV). In a recent Italian study, there is data to support increased prevalence of BAV in the Italian MFS population greater than the general population and that disease phenotype was more severe than MFS without BAV. In patients with LDS, the prevalence of BAV has been noted as a clinical finding; however, the impact of dual diagnosis in patients with LDS has not been described. The GenTAC database is a large repository of genetic, imaging and clinical information on patients with genetic predisposition for thoracic aortic aneurysms and related conditions. Therefore, it is a data repository designed for comparison between different types of syndromes associated with genetically triggered aneurysm.

CONCLUSIONS

Results:

- *Results pending*

DESIGN

Specific Aims:

- To evaluate the prevalence of BAV in patients with LDS and MFS
- To compare the severity and phenotype of disease in patients with MFS/BAV and LDS/BAV to those with MFS or LDS alone.

Inclusion criteria:

- Study participants with a diagnosis of LDS or MFS