



## GEN64 - Cardiac Phenotype in Loeys Dietz Patients Enrolled in GenTAC

**OBJECTIVE:** To generate an analysis of the available cardiac features and compare them to related connective tissue disorders such as Marfan syndrome (MFS) and bicuspid aortic valve syndrome (BAV) as well as to unaffected individuals.

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### ORGANIZATION

*Lead Investigator:* Jennifer Pardo Habashi, MD

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*Funding Source:* GenTAC and JHU

### CONCLUSIONS

*Results:* • *Results pending*

### BACKGROUND AND RATIONALE

Patients with Loeys Dietz syndrome exhibit a more severe phenotype with aortic dissection occurring at a smaller dimension than in MFS, but with aneurysms and tortuosity throughout the vasculature as well. A comprehensive and up to date description will aid physicians in diagnosing and managing patients with Loeys Dietz syndrome.

### DESIGN

- Method:*
- We will review phenotype data on the 73 patients enrolled with confirmed Loeys Dietz syndrome and report prevalence. When appropriate, we will compare to patients with related diagnoses such as Marfan syndrome.
- Inclusion criteria:*
- Subjects with confirmed LDS diagnosis.
  - Subjects with confirmed MFS, BAV and EDS diagnosis for comparison
- Samples:*
- None
- Data:*
- Organ system review
  - Surgical
  - Quality of life
  - Genetic
  - Image
  - Demographics

