



## GEN53 - Investigation the Role of Notch Signaling in Marfan Syndrome

**OBJECTIVE:** To determine if the Notch1 signaling pathway is altered in the aneurysmal ascending aorta in either the setting of Marfan syndrome, bicuspid aortic valve or from other etiologies

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### ORGANIZATION

*Lead Investigator:* Vidu Garg, MD  
*Co-Investigators:* Sara Koenig, BA  
*Funding Source:* Nationwide Children's Hospital

### CONCLUSIONS

*Results:* • *Results pending*

### BACKGROUND AND RATIONALE

Consistent with the aortic valve and aneurysm phenotypes, Notch1 transcripts are found in the developing and adult aortic valve and wall. Our initial report along with studies by other investigators have demonstrated an association between NOTCH1 mutations and malformations affecting the left-sided cardiac structures (BAV, aortic coarctation and hypoplastic left heart syndrome) and suggest that loss of NOTCH1 predisposes to cardiovascular disease.

### DESIGN

*Method:* Perform immunohistochemistry to determine if there is a change in the expression of Notch1 and its downstream targets, Hey1 and Hey2

*Inclusion criteria:*

- Subjects with MFS, BAV and TAAD < 50 years old.

*Samples:*

- Aortic Tissue

*Data:*

- Imaging
- Surgical

