



## GEN09 - Recurrent chromosome 16p13.1 duplications are a risk factor for aortic dissection

**OBJECTIVE:** Using DNA from GENTAC patients with confirmed thoracic aortic disease and no identified mutation in a known gene, a dense SNP array will be done to identify insertions and deletions in the genome

---

### ORGANIZATION

*Lead Investigator:* Diana Milewicz, MD, PhD

*Co-Investigators:* Dongchuan Guo, PhD, Scott LeMaire, MD, John Belmont, MD, Siddharth Prakash, MD,

*Funding Source:* NHLBI SCCOR

### BACKGROUND AND RATIONALE

Research proposal GEN01 identified a number of copy number variants (CNVs) that are either present at increased frequency in the patients compared with 3000 controls or not present in the controls. In addition, these CNV regions remain promising as disease causing variants after assessment of the Database of Genomic Variants and Baylor's clinical database of nearly 12,000 cases tested on BAC and Agilent oligo arrays. Several of these CNVs involve known genes for TAA.

### DESIGN

- Hypothesis*
- A subset of early onset, genetically triggered aneurysms, either associated with syndromic features or not, will result from insertions and deletions in the human genome.
- Inclusion criteria:*
- Confirmed thoracic aortic disease
  - Mutations in known genes excluded by sequencing data
- Samples:*
- Genetic material
- Data:*
- None

### CONCLUSIONS

- Results:*
- This study shows that common genetic variants at 16q13.1 that likely act via FBN1 are associated with STAAD, suggesting a common pathogenesis of thoracic aortic disease in MFS and STAAD.
- Publication*
- Kuang, S.-Q., Guo, D.-C., Prakash, S. K., Johnson, R. J., Wang, M., Regalado, E., et al. (2011). [Recurrent chromosomes 16p13.1 duplications are a risk factor for aortic dissection](#). *PLoS Genetics*, 7(6), e1002118.

