



GEN30 - Genetic Risks for Bicuspid Aortic Valve Disease

OBJECTIVE: To identify genetic variants that contribute to BAV disease and clinical outcomes, including aortic stenosis, aortic regurgitation and TAAD.

ORGANIZATION

Lead Investigator: Siddharth Prakash MD, PhD
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Funding Source: PI Start up funds

CONCLUSIONS

Results: • *Results pending*

BACKGROUND AND RATIONALE

Bicuspid aortic valves (BAV) are among the most common congenital cardiovascular disorders with a prevalence of 1-2% in adults. Bicuspid valves result from a developmental defect in the separation of the cusps and are highly heritable. The majority of BAV patients develop clinically significant aortic stenosis or regurgitation by age 60. The clinical impact of BAV due to morbidity from progressive aortic valve dysfunction is substantial. Approximately half of all aortic valve replacements in the United States are due to BAV disease.

DESIGN

- Hypothesis*
- Previously we showed that isolated TAAD is caused by an allelic spectrum of variants ranging from rare, highly penetrant mutations that cause syndromic or familial forms of the disease to common variants with reduced penetrance. We hypothesize that the allelic architecture underlying BAV is similar.
- Inclusion criteria:*
- Adults (≥ 18 years old) with diagnosed BAV.
- Samples*
- Genetic material
- Data*
- Surgical, imaging and genetics data. Demographics and family history and medication use.

