



GEN32 - Genetic Associations with Bicuspid Aortic Valve

OBJECTIVE: To identify genetic, and non-genetic, predictors and pathways of bicuspid aortic valve disease (BAV).

ORGANIZATION

Lead Investigator: Simon Body, MD, MPH

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Samples

- Genetic Material

Data

- Demographic, clinical, surgical, imaging and family history data

BACKGROUND AND RATIONALE

Bicuspid aortic valve (BAV) is the most frequent congenital cardiac malformation, occurring in 0.5-1.2% of the population. In adults, it can be a benign abnormality; but it is associated with thoracic aortic aneurysm or dissection in 20-30% of patients, and early development of aortic valve incompetence and calcification in >50% of patients. Bicuspid aortic valve disease accounts for ~40% of the >30,000 aortic valve replacements (AVR) performed in the US each year and is performed on a younger population than those presenting with calcific tricuspid aortic valve (TAV) disease. BAV is also a principal risk for thoracic aortic aneurysm and dissection in younger age groups. Yet, we know little of the etiology, cellular events and modifiers of disease progression for BAV to aortic valve stenosis and incompetence, and thoracic aortic aneurysm/dissection.

CONCLUSIONS

Results:

- *Results pending*

DESIGN

- Specific Aims*
- Genetic etiology and molecular biology of BAV.
 - Access the cellular pathophysiology of BAV disease
 - Identify factors that determine disease progression of BAV
- Inclusion criteria:*
- All BAV subjects without exclusion for other diseases or syndromes such as coarctation or Turner's syndrome.
 - Subjects > 18 years of age

