Online Appendix

International Bicuspid Aortic Valve Consortium (BAVCon)

BAVCon is a collaborative registry that consists of 16 institutions and also includes data from the National Registry of Genetically Triggered Thoracic Aortic Aneurysms and Cardiovascular Conditions (GenTAC, https://gentac.nhlbi.nih.gov). The authors thank Barbara Kroner, PhD, for assistance with GenTAC data. Demographic, clinical and imaging data on all patients who are enrolled at participating sites are collected using standardized forms. All data elements were prospectively defined, and study protocols were approved by institutional review boards at each site. Images from each enrolled patient are reviewed by local principal investigators to ensure data accuracy. Imaging modalities and medications are determined by treating physicians according to clinical guidelines and institutional policies.

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Glossary of Terms

Penetrance : the proportion of individuals with a mutation who exhibit clinical symptoms

Linkage analysis : the formal study of the association between the inheritance of a condition in a family and a particular chromosomal locus

Pedigree : genetic representation of a family tree that diagrams the inheritance of a trait or disease though several generations

Heritability : the proportion of observable differences in a trait between individuals within a population that is due to genetic differences

Epigenetic : the study of changes in gene expression or cellular phenotype, caused by mechanisms other than changes in the underlying DNA sequence

LOD score : logarithm (base 10) of odds, a statistical test often used for linkage analysis that compares the likelihood of obtaining the test result if the phenotype and genetic locus are indeed linked, to the likelihood of observing the same data purely by chance

Expressivity : variations in a phenotype among individuals carrying a particular genotype

Positional cloning : a method of gene identification in which a gene for a specific phenotype is identified only by its approximate chromosomal location

Proband : the first affected family member who seeks medical attention for a genetic disorder

Alleles : variant forms of the same gene

Genome-wide association study : an approach that involves rapidly scanning markers across the entire genomes of many people to find genetic variations associated with a particular disease

Intron : nucleotide sequence within a gene that does not code for a protein

Segregation analysis : statistical test to determine the pattern of inheritance of a trait

Exome : the protein-coding portion of the genome, approximately 1% of the total genomic sequence