

## Genetic analysis of individuals and families with congenital heart disease

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### Synopsis:

Recent studies by Garg et al. have looked at human pedigrees with aortic valve disease. It was found that NOTCH 1, a signaling and transcriptional regulator, when mutated cause a variety of aortic valve anomalies. Other studies, using similar genetic analyses of human pedigrees with similar congenital heart defects, elucidated other genes that are related to congenital heart disease. In this study, we hope to understand how genetic variation causes congenital heart disease.

Certain congenital heart diseases will be caused by mutations in individual genes. However, other types may be caused by subtle changes in multiple genes. By performing a genotype association study, such genetic variation can be correlated with disease. We hope that this study will lead to the identification of genes that are related to the congenital heart disease.