

# Project 2

Driscoll Lab

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# Primary Aim

To investigate the genetic component of outflow tract malformations

# Subjects

- Trios consisting of probands with outflow tract anomalies and their parents

# Genotyping

- Candidate genes
- Single nucleotide polymorphisms (SNPs)

# Methods

- High throughput single nucleotide polymorphism (SNP) genotyping
- Statistical analysis
  - transmission disequilibrium test (TDT)

# Jag1

- Expressed in the outflow tract in human (day 32)
- Notch signaling pathway
- JAG1 mutations have been associated with CHD

# RARb

- Belongs to retinoic acid receptor (RAR) family
- Expressed during early heart development in mouse (TS 13)
- Imperative for vitamin A utilization
- Double mutant RAR mice exhibit cardiac defects including outflow tract abnormalities

# Next stage

- 120 trios from 2004/2005 have been added
- 12 new SNPs have passed in-house QC
- We have a new list of candidate genes to consider