



People with Down syndrome
helping to cure
congenital heart disease
in all people

- 40% of children born with Down syndrome (or trisomy 21) have a congenital heart defect (CHD).
- The most common form of CHD in Down syndrome is an atrioventricular septal defect (or AVSD)
- Children with Down syndrome are 2000 times more likely to have a complete AVSD than children without Down syndrome
- Researchers are conducting a multi-site study to identify genetic risk factors for

Who can participate in this study?

- Individuals with Down syndrome and a complete AVSD under the age of 18, and their parents
- Individuals with Down syndrome who were born with no heart defect and their parents may participate as control subjects

Participants in this study will be asked to:

- **Sign** a consent form
- **Provide** access to medical records such as a chromosome report and echocardiograms
- **Give** blood samples from the person with Down syndrome about 2 tablespoons from adolescents and about 1 teaspoon for smaller children. Children's blood samples may be taken at the time of routine blood work.
- **Give** saliva samples from the parents of the child with Down syndrome
- **Complete** telephone questionnaires (30-45 minutes) that covers health history, family health history, and environmental exposures.

Who is funding this study?

The study is funded by the National Institutes of Health (NIH).

How will you protect my privacy?

- Information you give us during the interview, as well as lab results from the samples you provide, will be kept confidential.
- Numbers instead of names will be used to identify participants' information.
- Personal information about you **will not** be available to anyone outside this study.
- We will **never** use your name or your family's name in any report or publication.

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