



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

National Down Syndrome Study Research Announcement!!

WHY: We are inviting families of children with Down syndrome to participate in an NIH funded study to identify genetic and environmental factors that increase the chance of congenital heart defects. This study will help us understand why some children with Down syndrome have heart defects while others do not.

WHO: We are planning to enroll 600 children born with Down syndrome and atrioventricular septal defect (AVSD), commonly called AV canal, and 600 children born with Down syndrome and **without** any structural heart defect. The children must be under the age of 18 years old. **This is an expansion of previous studies started in 2001. If you have participated before, you will not need to enter this study again.**

WHERE: This study is a collaborative effort between multiple sites, including Kennedy Krieger Institute and Johns Hopkins University, both in Baltimore, MD.

WHAT: Parents will be asked to complete a telephone interview (30 – 45 minutes) about their pregnancy and reproductive histories and maternal and family health related factors. Additionally, parents will be asked to donate a saliva sample for DNA analysis. Adolescents will be asked to donate a small blood sample (1.5-4 teaspoons), and younger children will be asked to donate a smaller blood sample (minimum $\frac{1}{5}$ teaspoon) for DNA analysis. **The blood samples can be drawn during a regular visit to one of the sites or by arrangement with your primary care physician.** We will ask to obtain your child's medical records that relate to their diagnosis (such as a karyotype) and their heart status (such as an echocardiogram report). There are no direct medical benefits or significant risks for participating in the study.

HOW: If you are interested in participating or have questions, please contact us!

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APPROVED

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