GEISINGER
AUTISM & DEVELOPMENTAL MEDICINE

Seeking volunteers for research into developmental, behavioral and medical features associated with 17q12 deletion syndrome

The chromosomes contain genetic material that tells our bodies how to grow and develop. They come in pairs, one set inherited from each parent. When a person has extra or missing genetic information (a duplication or deletion), they may have an increased chance for health or developmental concerns.

When a deletion of chromosome 17q12 occurs, an individual has an increased risk for neurodevelopmental disorders (such as developmental delay/intellectual disability, autism spectrum disorder, and schizophrenia) and medical issues (kidney problems and diabetes). The purpose of this research study is to identify and better understand the developmental, behavioral and medical features found in individuals with a 17q12 deletion. This information will be useful in aiding future diagnosis and clinical decision-making for individuals with unexplained neurodevelopmental disorders.

Overall study aims:

- To identify individuals who are at high risk for carrying a 17q12 deletion
- To collect information about developmental, behavioral, and medical features associated with 17q12 deletion

Your participation will involve:

- Completing several online surveys (or we can mail paper copies if you are unable to complete online)
- A medical record review
- In-person direct cognitive assessment (available to local U.S. families only)

Who can participate in online surveys:

- Children, adolescents, or adults aged 3-55 who:
    - have confirmed 17q12 deletion, and speak/read English as their primary language

For questions about your rights as a research participant, contact the Human Research Protection Program (HRPP) staff of the Geisinger Institutional Review Board (which is a group of people who review the research to protect your rights) at (570) 271-8663. The principal investigator of this study is Dr. David H. Ledbetter.

If you would like to learn more about taking part in this study, please contact:

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