Are you interested in participating?

To learn more please call us at (215) 906-5071. We can also be reached by email at bblresearch@upenn.edu.



Find us on Facebook!

How else can we get involved?

Please join us at our free monthly support and education group for family members and those diagnosed with 22q11.2DS

Where are we located?

Hospital of the University of Pennsylvania – Neuropsychiatry Section

3400 Spruce Street Philadelphia, PA 19104

Principal Investigator: Raquel E. Gur, MD, PhD.

CHOP – "22q and You Center"

34th Street and Civic Center Blvd. Philadelphia, PA 19104

Principal Investigator: Beverly S. Emanuel, PhD

Director, Clinical Genetics: Elaine Zackai, MD

Program Director: Donna McDonald-McGinn, MS, CGC

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For more information: Call 215-906-5071

www.med.upenn.edu/bbl/ projects/22q



Brain-Behavior & Genetic Studies of the 22q11.2DS



Please contact us for more information and to participate in our research studies.

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About Our Study

Our mission

This study is a combined effort of the Hospital of the University of Pennsylvania and the Children's Hospital of Philadelphia "22q and You" Center. We are aiming to better understand brain and behavioral differences in individuals ages 8 years and older with the chromosome 22q11.2 deletion syndrome. In addition, by looking at individuals with the deletion, we may begin to better understand risk factors associated with brain and behavior changes, as people grow older.

Who is eligible?

This study might be a good fit if:

- You or your child has a diagnosis of 22q11.2 deletion syndrome
- You or your child is age 8 years or older

What will the study involve?

If you or your child decides to take part in the study, participation will include:

- A clinical interview
- Solving memory and puzzle-like games on a computer
- Providing a small blood sample
- Ages 12 or older may be asked to consider taking part in a brain fMRI

Compensation

Participants will be compensated for participation, time, and travel.



What does it all mean?

Clinical Evaluation involves clinical interviews and standard tests performed by doctors and trained research staff.

Neurobehavioral Assessment includes computerized tests of attention, learning, memory, language skills, and sensorimotor integration.

Neuroimaging is a comprehensive evaluation of brain anatomy and physiology in relation to behavior.

Functional MRI (fMRI) shows changes in brain activity as participants perform cognitive tasks while in an MRI machine.

Blood Samples give us genetic and molecular information about how the brain works. We may ask to obtain a blood sample from your arm, hand or wrist.