Autism Spectrum Disorders and Genetic Testing

Why is genetic testing important in Autism Spectrum Disorders?

The onset of ASD occurs in the first few years of life and affects approximately 1 in 88 children (MMWR Surveill Summ 2012 March 30; 61 (SS03):1-19). Autism Spectrum Disorders are more common among boys (1 in 54 boys) than girls (1 in 252 girls). Studies have shown that 10-20% of people with an ASD have an abnormal genetic result that explains the cause of their ASD. Early detection and accurate diagnosis are critical because:

- Early interventions can reduce challenges associated with ASD, lessen disruptive behavior, and provide some degree of independence.
- If the ASD is related to a genetic abnormality, family members can be properly counseled about their risk for having another child affected with ASD.

The UCLA Autism Evaluation Clinic strongly recommends that all individuals with a suspected developmental delay or ASD obtain genetic consultation and testing. The American College of Medical Genetics indicates that it is medically necessary and standard practice for individuals with an ASD and/or cognitive delays to undergo genetic testing.

What is genetic testing?

Genetic testing is a type of medical test that identifies changes in a person's DNA. Genes are made of DNA and provide the blueprint for how our bodies grow and develop. Thousands of genes are packaged into structures called chromosomes.

Genetic testing is used to find DNA changes associated with specific medical conditions. Genetic testing can help determine whether a person has a genetic disease, whether it has been inherited, and the chance for future children or other family members to have the same condition.

What genetic testing is available for ASD?

Three genetic tests detect the most common known genetic causes of ASD:

- Clinical Exome Sequencing: This genetic test simultaneously investigates all possible causative genes. Exome sequencing is most informative when the child’s sample is compared to DNA samples of each parent (a trio sample). Results are available approximately 16 weeks after the blood sample is drawn. This is our preferred genetic test at this point.
- Cytogenetic Array: This genetic test detects pieces of the chromosomes that are either missing (deletions) or extra (duplications). Deletions and duplications (also called copy number variations, or CNVs) lead to an atypical number of genes
being present in an individual. Recent news stories have highlighted two scientific studies that showed the cytogenetic array test can detect the cause of ASD in 6-7% of individuals.

- Fragile X: Fragile X Syndrome is the most common form of hereditary intellectual disability. Both males and females can be affected, but because the gene is on the X-chromosome, more males are affected than females. Fragile X syndrome is tested for because many of the non-physical symptoms of the disease are similar to those displayed in ASD.

**What will the results tell me?**

A normal result does not change the diagnosis of an ASD. It simply means the cause of the ASD is still unknown. Further developmental or genetic testing may be recommended.

An abnormal result indicates that a genetic difference has been found. This abnormality may be one that is associated with an ASD. When a genetic cause for the ASD is identified, other family members have the opportunity to seek genetic counseling.

In some cases, a genetic abnormality is difficult to interpret. In these cases, blood testing of the parents may be needed to clarify and interpret testing results.

**Where can I seek genetic testing?**

At UCLA, genetic counseling and testing may be scheduled by contacting Naghmeh Dorrani at (310) 206-6581. Counseling and testing is also available through private medical providers and developmental disability centers. The UCLA Clinical Genomics Center works closely with UCLA genetic counselors and is one of few places in the United States that offers clinical whole exome sequencing.

**Additional Information**

- [Autism](#)
- [Genetics & neurochemistry](#)
- Research Opportunities
  - [http://labs.genetics.ucla.edu/martinez-agosto/chome.html](http://labs.genetics.ucla.edu/martinez-agosto/chome.html)
  - 310-794-2405.