THE UNIVERSITY OF ARIZONA
COLLEGE OF MEDICINE TUCSON
Genetic Counseling
Graduate Program

GENETIC TESTING FOR AUTISM SPECTRUM DISORDER

WHAT IS AUTISM SPECTRUM DISORDER (ASD)?

Autism spectrum disorder (ASD) is a developmental condition. It can affect both social communication and behavior. ASD should be diagnosed by a qualified clinician, such as a developmental pediatrician or psychologist.

People with ASD may have trouble looking people in the eye or have a hard communicating with others. Some people make the same body movements or sounds over and over (repetitive behaviors). Some may also have a strong interest in a specific topic (restrictive interests).

IS ASD GENETIC?

- ASD may be part of a pattern of signs and symptoms which are connected to each other (syndromic). Syndromic ASD is more likely to have a genetic diagnosis, like fragile X syndrome.
- Non-syndromic ASD does not have other signs or symptoms and is less likely to have a genetic diagnosis.
- In some cases, ASD can run in families.

IS GENETIC TESTING RECOMMENDED FOR ASD?

In some cases, genetic testing can:

- Help find a cause / make a diagnosis.
- Understand if there is a chance for future children in a family to have ASD.
- Determine if testing for family members is recommended.
- Guide patient care.

A recent study found that about 12% of patients who completed genetic testing had a genetic diagnosis. Of those with genetic findings, 72% had medical recommendations made based on those results1.

1Harris et al., 2020; Pathogenic Yield of Genetic Testing in Autism Spectrum Disorder. Pediatrics, 146(4).

WHAT GENETIC TESTS ARE RECOMMENDED FOR ASD?

The American College of Medical Genetics (ACMG) and the American Academy of Pediatrics (AAP) recommend a genetic evaluation be offered to all patients diagnosed with ASD. A chromosomal microarray (CMA) and fragile X testing may be offered first. If those tests are negative, whole exome sequencing (WES) may be recommended.

WHAT IS A CHROMOSOMAL MICROARRAY (CMA) AND FRAGILE X TESTING?

Our DNA is like an instruction book for how our bodies are put together and function every day. If our DNA is an instruction book, the chapters would be our chromosomes. The sections of the chromosomes, called bands, would be the pages. Our genes would be the words on the page. Missing or extra pages, or spelling mistakes on the page, can change the way our bodies and brains develop and work.

A CMA looks for extra or missing pages within each chapter. It can find pieces of chromosomes that are extra (duplicated) or missing (deleted). Sometimes when pieces of chromosomes are extra or missing, it can cause ASD.

Fragile X syndrome is another genetic change that can cause ASD. It is diagnosed with a test that looks for how many times a series of letters in a certain word are repeated. When the letters within this gene (word) are repeated more than 200 times, it can cause fragile X syndrome.

WHAT IS WHOLE EXOME SEQUENCING (WES)?

WES looks for spelling mistakes throughout the entire instruction book. It looks at all of the active DNA that we use every day and tells us if there are any changes. Sometimes a spelling change in a gene is not harmful (normal variant). Other times, spelling changes can cause ASD. If it is not clear if a spelling change is causing a symptom, the family may be asked to give more samples. For example, DNA samples from a biological parent or sibling may help better understand the genetic change.

WHAT ARE THE POSSIBLE RESULTS?

Results of genetic testing can be positive, negative, or inconclusive.

- A positive result means a change was found in the DNA that is related to the person's signs or symptoms. If a result is positive, your provider will discuss what it means and if it will change treatment or management. They will also discuss if it may affect other family members or future children.
- A negative result means no changes were found that are related to the person's signs or symptoms.
- An inconclusive result can also be called a "variant of uncertain / unknown significance". This means a change was found in a person's DNA, but it is not clear if the change is related to their condition. Some labs keep researching this change and update your provider when they learn what it means. Talk about this with your provider.

IS GENETIC TESTING FOR ASD COVERED BY INSURANCE?

Genetic testing for ASD may be or may not be paid for by insurance. If genetic testing is covered, insurance companies may cover some or all of the costs. Contact your insurance carrier to better understand what is and is not covered. A member of your healthcare team may also have more information to help you better understand your options.

IS GENETIC TESTING RIGHT FOR OUR FAMILY?

Every situation is different. Talk with your healthcare team about genetic testing to decide if it is right for you.