

Will It Happen Again? Autism, Genetics and Genetic Counseling

Evidence from many studies suggests that inherited factors are largely responsible for the occurrence of autism spectrum disorders (ASD). In some cases, autistic disorder is a feature of an identifiable genetic condition. More frequently, no underlying specific cause can be determined (this is called idiopathic autism, meaning autism caused by unknown factors). There is some evidence that idiopathic autism is caused by changes or mutations in genes. Currently, there have been approximately 30 genes suggested in association with ASD. However, no specific genes or combination of genes have been consistently associated with autism to date.

Will it happen again to our future children?

For parents with one child affected by idiopathic autism, the risk of autism in a subsequent child is approximately 5%. When more than one child is affected, the recurrence risk is greater than 8%, and possibly as high as 25%.

Could autistic disorder still be genetic in my family even if no other family members have it?

Yes. The genetic changes that cause autistic disorder may be new in the family. These changes could occur when DNA, the genetic material we got from both of our parents, was copied to make sperm or eggs, and then passed down generations.

Is a female child less likely to be affected?

Yes. A male fetus is three to four times more likely than a female fetus to develop autism. Therefore, the risk of having another affected child according to sex is closer to 2% for a female fetus and 8% for a male fetus. Currently, there is no known reason why more males have autistic disorder than females.

Are there environmental causes to ASD?

Although ASD has a strong genetic basis, for some at risk individuals, a possible cause might involve environmental triggers. The link between vaccines and ASD is probably the most debated environmental cause of autism. However, to date, there has been no definitive, scientific evidence that any vaccine or combination of vaccines can cause ASD. More information about autism and the MMR vaccine is available at the website of National Institute of Child Health and Human Development: www.nichd.nih.gov/publications/pubs/autism/mmr/.

Are there increased risks for autism-like symptoms that are not Autism?

Immediate relatives, meaning parents, siblings and offspring, of individuals with ASD are at increased risk of having autistic-like behaviors that fall below the threshold for a diagnosis of ASD. The risk of a sibling exhibiting one or more features of the broader autistic-like behaviors might be as high as 30%.

Are there genetic disorders associated with autism?

Autism tends to occur more frequently than expected among individuals who have certain medical conditions, including fragile X syndrome, Rett syndrome, tuberous sclerosis, neurofibromatosis type1 (NF1), and untreated phenylketonuria (PKU). Some chromosomal changes have also been associated with an increased risk of autism.

Why genetic counseling? What can a genetic counselor do for us?

Genetic counselors are health professionals who provide information and support to families who have members with birth defects or genetic disorders. They investigate the medical problem present in the family, interpret information about the disorder, analyze inheritance patterns and recurrence risk, and discuss available options with the family.

If a medical reason is determined, risk of having another child with ASD may be refined. Meanwhile, if the condition is caused by certain medical conditions, early intervention and treatments can be tailored to the

person according to the nature of the disorder.

What Happens at a Genetic Counseling Session?

When you go to see a genetic counselor, he or she will:

- collect your family medical history and your medical history as well.
- evaluate how your family's health may affect you or your children, and what the risk is for a genetic condition to happen again in the family.
- help you make decisions if you want further tests by discussing pros and cons with you.
- help you understand the test results and discuss how the results may affect your health and your life with you.

If there is a genetic condition revealed by the test results, the counselor will discuss available options with you and help you make decisions. If there is a specific genetic condition identified, the counselor or the doctor can refer you to other resources or to medical specialists, educational specialists, or support groups that you may wish to know about.

What forms of prenatal genetic tests are available?

If there is a medical reason, prenatal genetic tests for some genes or chromosomal changes are available. Amniocentesis is a medical procedure in which a small amount amniotic fluid is extracted from the amniotic sac. Amniotic fluid is the fluid that surrounds your baby in the uterus and contains cells shed by the baby. Chorionic villus sampling (CVS) is another type of medical procedure involves taking a sample of tissue from the placenta. From the amniotic fluid or the placenta tissue, the baby's DNA is extracted and examined for genetic abnormalities.

Where Can I Find a Genetic Counselor?

You can ask your local hospitals or go to the website of National Society of Genetic Counselors (www.nsgc.org) to locate genetic counseling services.

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