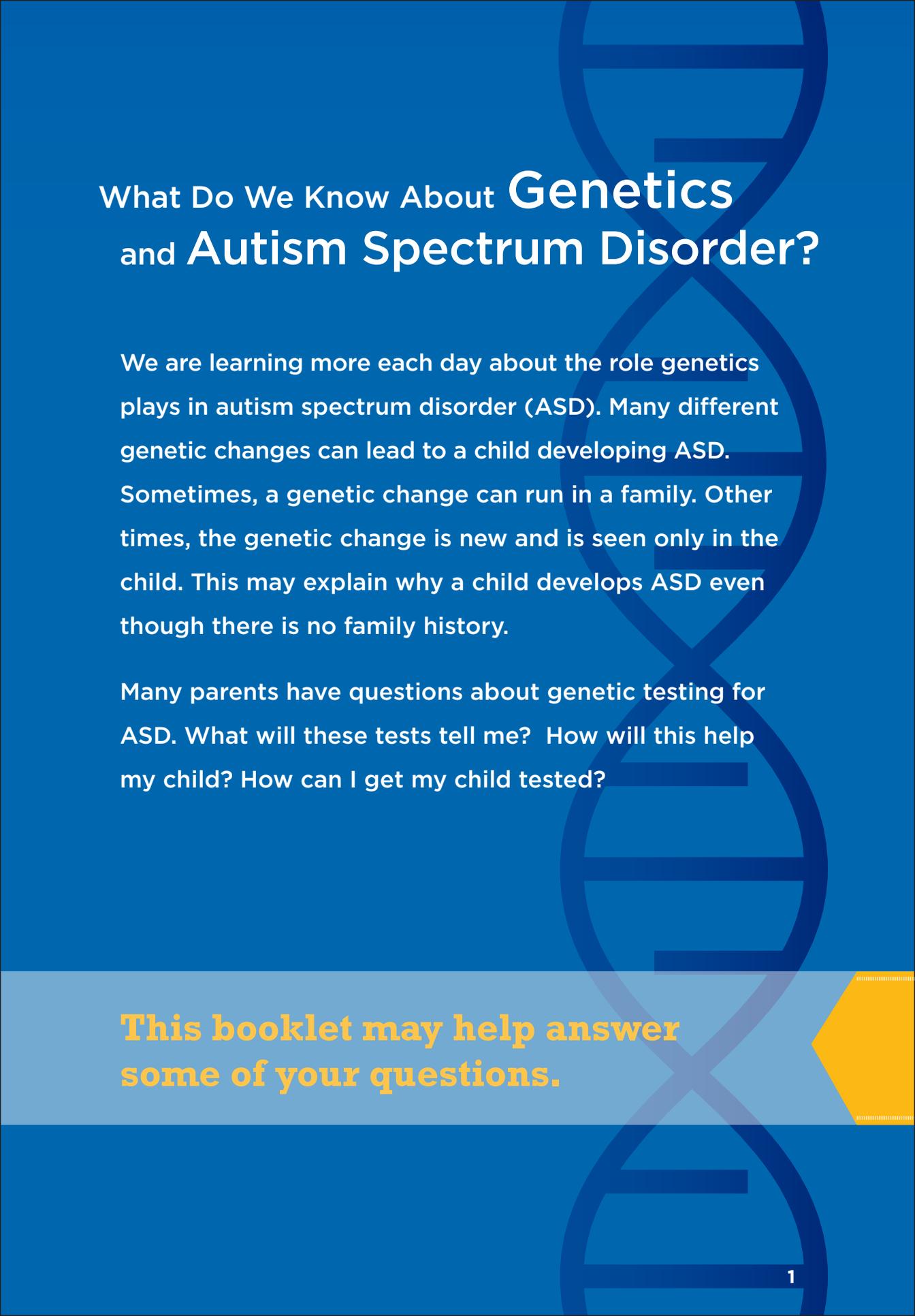


GENETIC TESTING

for Children with Autism Spectrum Disorder







What Do We Know About **Genetics** and **Autism Spectrum Disorder?**

We are learning more each day about the role genetics plays in autism spectrum disorder (ASD). Many different genetic changes can lead to a child developing ASD. Sometimes, a genetic change can run in a family. Other times, the genetic change is new and is seen only in the child. This may explain why a child develops ASD even though there is no family history.

Many parents have questions about genetic testing for ASD. What will these tests tell me? How will this help my child? How can I get my child tested?

**This booklet may help answer
some of your questions.**

Why Is Genetic Testing Recommended for Children with ASD?

Genetic testing for ASD can:

1. Provide an explanation of why your child developed ASD.
2. Help determine the chances that existing or future children will develop ASD. Depending on the test results, the risk to other family members ranges from as low as 5% to as high as 50%.
3. Help determine the types of services that your child may benefit from.
4. Help identify other health issues. For example, some genetic changes that lead to ASD can also lead to other health issues like eye problems. In this case, doctors could recommend that a child see an eye doctor.

Genetic testing for ASD cannot:

1. Tell you exactly how your child will develop.
2. Tell you how well treatments will work for your child.

Genetic testing for ASD may provide an explanation of why your child developed ASD.

Myths About Genetic Testing for ASD

Facts About Genetic Testing for ASD

All genetic changes found by testing will lead to health problems...



Some genetic changes are common and **do not** lead to health problems.

A negative test result means there is no genetic link for your child's ASD...



A single genetic test **does not** test for all possible genetic changes.

Genetic test results will impact your health insurance...



It is **against the law** for your health insurance to change based on the results of your genetic test.

It will show who the father is...



It is **not** a paternity test.

It will test for HIV or other viruses...



It **does not** test for HIV or other viruses.

It will test for substances or other drugs...



It **does not** test for substances or other drugs.



How Do I Get My Child Tested?

A genetic test usually involves a blood test. There are many different genetic tests available. Doctors often recommend two tests: Fragile X and chromosome microarray, or CMA. Doctors may also recommend other tests, depending on your child's development, medical history, and your family history.

Even if initial genetic tests for ASD do not find any genetic changes in your child, there may still be a genetic explanation. For this reason, more testing may be recommended.

There are two ways to get genetic testing for your child:

1. Your child's doctor can order the genetic test.

You can make a special appointment with your child's doctor for the genetic test, or schedule the test when your child is having blood drawn for routine screening.

2. Your child's doctor can refer your child to a genetics team.

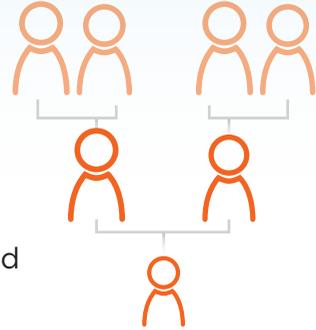
A genetics team consists of a **genetic counselor** and a **geneticist**. This service is usually covered by insurance, just as referrals to other specialists are covered. A **genetic counselor** has specialized training in explaining genetics. A **geneticist** is a doctor with expertise in genetic conditions including those that involve ASD.



What Does a Genetics Team Do?

The genetics team will gather information from your family to figure out the chances that a genetic change led to your child's ASD. At the visit, you will be asked about your child's medical history and your family history.

Your child will also have a physical exam. Based on what the team learns about your child and your family, they will discuss further steps that could help determine if there is a genetic explanation for your child's ASD. The visit with the genetics team will give your family time to ask questions and talk about any concerns you might have.



The Genetics Team Can:

- Give you a more detailed explanation of the genetic tests available and the possible results
- Act as a guide for your family during the genetic testing process
- Suggest the tests that are best for your child based on your family's history
- Help determine if your child has any additional medical problems that are related to ASD
- Talk with you about the chance for ASD to occur in other family members
- Work with you and your insurance company to get tests covered

Meeting with a genetics team is often helpful, even if you do not have genetic testing done.

What Should I Ask My Child's Doctor about Genetic Testing for ASD?

Here are some questions you might want to ask your child's doctor as you consider having genetic testing done for your child with ASD:

About Getting Tested

- How soon should I think about having testing done?
- Should I be thinking about talking with a genetics team?
- I'm not planning on having any more children, so will genetic testing still be useful for our family?

About the Results

- How long will it take to get the test results?
- Who will explain the test results to me?
- What exactly will the test results tell me?
- What does it mean if the test results are negative, positive, or of unknown significance or meaning?
- Who can see the test results besides the doctors and my family? Will information be kept private?

Will My Insurance Pay For Genetic Testing?

Many insurance plans cover the blood test needed for genetic testing, but you should talk to your child's doctor and your insurance company about your specific plan. You and your doctor or the genetics team can work with your insurance company to determine if recommended tests are covered and if there will be any cost to you.



Genetic testing may help you and your family. Talk to your doctor to see if testing is right for your child.

More Resources

For more information:

- Centers for Disease Control and Prevention (CDC):
<http://www.cdc.gov/genomics/resources/diseases/autism.htm>
- National Human Genome Research Institute (NHGRI):
<http://www.genome.gov/25522099>
- Genes in Life—Genetics 101:
<http://www.genesinlife.org/genetics-101>

Autism Speaks has several websites on related topics:

- Genetics and genomics:
<http://www.autismspeaks.org/science/initiatives/autism-genome-project>
- ASD genetic testing recommendations for children:
<http://www.autismspeaks.org/science/science-news/new-guidelines-genetic-testing-children>
- How to find specific information on specific genetic changes related to ASD:
<http://www.autismspeaks.org/blog/2013/02/08/finding-information-autism-related-genes>

For More information

© 2015 Boston University School of Public Health

We would like to acknowledge all the parents and health professionals who have provided invaluable feedback and contributed to the development of this booklet.

The content of this booklet was developed by a team from Boston University School of Public Health and Boston Medical Center (BMC), in collaboration with members of the Autism Consortium. For more information, please contact the Family Support Network and Autism Program at BMC: autismprogram@bmc.org.



This work was supported by The National Genetics Education and Consumer Network (NGECN) (Cooperative Agreement #U22MCO4100), funded by the Genetics Services Branch of the Maternal and Child Health Bureau, Health Resources and Services Administration (HRSA).



Designed by Codesign.