



Genetic Testing for Autism

Genetic testing involves the direct examination of the DNA molecule and testing that DNA for genetic disorders. They are used for several reasons such as carrier screenings, newborn screenings, prenatal diagnosis testing, forensic/identity testing, etc. The American Academy of Pediatrics (AAP) and American College of Medical Genetics and Genomics (ACMG) recommend that the decision to offer genetic testing should be in the best interest of the child. Clinical geneticists, genetic counselors or any other health care professional with specific training can offer genetic testing. Research has expanded on genetic testing since this issue has grown. Debate about formal parental consent has been a large opposition for newborn genetic screening as well. An early diagnosis can prepare families for the challenges that are ahead for their child. Families can prepare their plan of action and identify the risk of diseases that may be preventable.

When is it appropriate?

- Distinguishing between autism spectrum disorder and other behavioral or developmental disorders
- Assessing your child for medical concerns
- Determining genetic causes
- Outlining the child's strengths and challenges

Pros

- Early diagnosis and treatment can greatly outweigh the risks
- Carrier screening can increase integration status into life plans
- Avoidance of the shock and resentment that may occur when screening is delayed
- Greater opportunity for parental guidance in available management options
- Tests may discover conditions like Fragile X Syndrome, tuberous sclerosis and epilepsy
- Reoccurrence risk (the risk of having another child with ASD is approximately 4 to 7%)

Cons

- Cost of tests (Not all families are covered by insurance)
- Results of tests are not always a yes or no answer, may be difficult to interpret
- Some families may feel a sense of guilt or blame once the genetic variant is found in one half of the family
- Treatment recommendations rarely change for a child with ASD based on genetic test results

Resources

- <http://pediatrics.aappublications.org/content/early/2013/02/17/peds.2012-3680.full.pdf+html>
- <http://pediatrics.aappublications.org/content/early/2013/02/17/peds.2012-3680.full.pdf+html>
- <http://www.childrenshospital.org/az/Site613/mainpageS613P3.html>