

Genetic Testing for Developmental Delays and Behavioral Problems

Genetic Testing has been recommended for your child.

- Some children have a genetic cause for developmental delays and/or behavioral concerns.
- Finding a genetic cause can be important to your child and your family.
 - A genetic result can give more information about your child's health now and in the future. This allows us to take better care of your child because we know what to expect.
 - Genetic testing can also answer questions about the chance a future child in your family may have similar health issues.
 - If your child has a positive ("abnormal") result, a visit with a genetics doctor may be recommended
- This test cannot identify all genetic conditions.
 - Even if your child's test is normal, there may still be a genetic cause for his/her delays. This is more likely if the child has delays AND other health problems. If your doctor thinks more genetic testing is needed, he/she may send you to a genetics doctor.
- A negative ("normal") genetic test result does NOT change your child's diagnosis. Your child should still be seen by his/her doctors and therapists.

What is DNA? Genes? Chromosomes?

DNA is genetic material we all have. It provides information our body needs to grow and develop. DNA is packaged into genes. Genes are like ingredients in a recipe. If there are extra or missing ingredients- or if the wrong ingredient is added to a recipe- there will be a difference in the final product. Many genes together are packaged on a chromosome. Most people have 46 chromosomes. If there is extra or missing information on a chromosome, it can cause delays in a child.

What are we testing for?

Fragile X testing

- Fragile X syndrome is the most common cause of mental retardation (severe delays) that results from a change in a single gene and is passed down in families.
- Fragile X syndrome is caused by a change in the *FMR1* gene. Mental delays usually affect boys more severely than girls. Women with a change in this gene can have early ovarian failure. Some adult men with a change in this gene have problems with memory and muscle control later in life.
- If your child has Fragile X, we will offer testing to other family members to see if this could happen in future children and to find out if anyone else is at risk for these health concerns.

Chromosome studies

- These studies can find extra or missing chromosomes, large pieces of missing genetic material (deletions), large pieces of extra genetic material (duplications), and large pieces of DNA that have been moved around (translocations, inversions or insertions).
- If your child has an abnormal chromosome result, we will offer testing to both parents to see if this could happen again in future children.

Microarray testing

- Microarray can find smaller pieces of missing (microdeletions) or extra (microduplications) genetic material (DNA) that is not seen by chromosome studies.
- Possible results are normal, abnormal, or a result of unknown significance. If your child has a result of unknown significance, both parents will be offered testing to help understand the result. Genetic changes that cause health problems are often new in a child. They are less likely to be passed down from an unaffected parent (a parent who does not have health concerns).