

# NeuroDevelopmental Reflex Genetic Testing

## Information for Patients

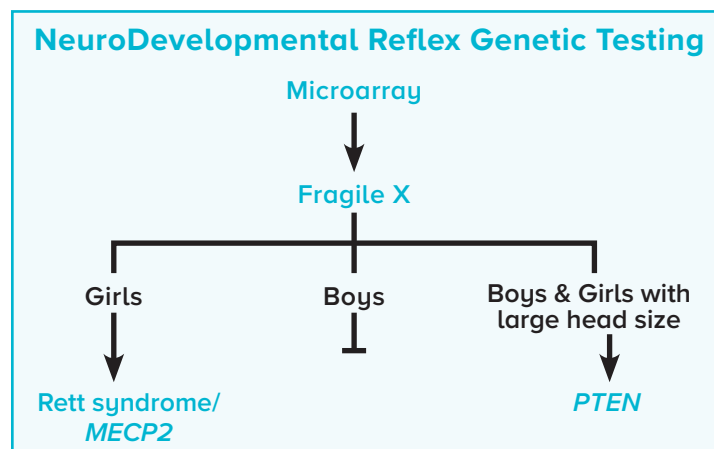
Genetic testing can be used to find a genetic cause for a patient's developmental delays, intellectual disability, and/or autism. This testing could:

- Give us more information about a patient's health now and in the future, allowing us to better care for that patient
- Answer questions about the chance that a future child in the patient's family could have similar health concerns

### What is DNA? Genes? Chromosomes?

DNA is all of the genetic material inside our cells. It provides the instructions that our bodies need to grow and develop. DNA is packaged into genes which are organized into structures called chromosomes. Genetic testing looks for changes in genes or for extra or missing pieces of DNA. Changes in genes or DNA can result in genetic disorders. Often these are known disorders with specific medical management recommendations. Sometimes genetic disorders are not as well known or understood.

For children with developmental delays and/or autism, we use a set of genetic tests in a particular order to give us the best chance of finding a genetic cause in the most efficient way possible. Testing options are determined based on the patient's sex (boy or girl) and head size.



If one of these tests finds a genetic cause for the patient's developmental delays, intellectual disability and/or autism, we will not need to continue to the next test. If a genetic cause is found for the patient's developmental delays, intellectual disability, and/or autism, the patient's health care provider will discuss this new diagnosis in more detail and/or make a referral to genetics or a specialty clinic.

These genetic tests are not able to detect all of the possible genetic changes that can lead to developmental delay, intellectual disability, and/or autism. If this testing is unable to find a genetic cause of the patient's developmental delay, intellectual disability, and/or autism, it does not mean that there is not a genetic cause. Further genetic testing may be indicated for the patient now or in the future. We encourage the patient and their health care team to continue consulting with genetics.

### Microarray

- A microarray is a genetic test that is used to find extra or missing genetic material (DNA)

- Too much or too little genetic material can cause health problems
- This test may find:
  - The cause of developmental delay or autism
  - Other health problems not related to developmental delay or autism
  - A close blood relationship between the patient's parents
  - A result that is uncertain or about which little is known

### Fragile X testing

- This test will tell us if the patient has Fragile X syndrome.
- Fragile X syndrome is the most common inherited cause of intellectual disability and results from a change in a single gene (*FMR1*) that is passed down in families.
- The *FMR1* gene is located on the X chromosome. This means that most boys have only one copy of *FMR1* and most girls have two copies of *FMR1*. Because of this, boys often have more severe symptoms of Fragile X syndrome than girls.
- If we find that the patient has Fragile X syndrome, we will offer testing to the patient's mother to see if this could happen again in future children. Parents can have changes in *FMR1* without having Fragile X syndrome.
- Women with a change in *FMR1* can have early ovarian failure. Some adult men with a change in this gene have problems with memory and muscle control later in life.

### PTEN testing

- This test will tell us if the patient has a change in the *PTEN* gene.
- Changes in *PTEN* can cause autism and a large head size. Changes in *PTEN* can also cause skin differences, growths in the intestines, and an increased chance for certain types of cancers.
  - *PTEN* changes that cause autism and a large head size have less cancer risk than changes that do not cause autism and a large head size.
- If we find that the patient has a change in the *PTEN* gene, we will offer testing to both parents to see if one of the parents also has the change. This will give us more information about whether this could happen again in future children.

### MECP2 testing

- This test will tell us if the patient has a change in the *MECP2* gene.
- Changes in the *MECP2* gene cause a condition known as Rett syndrome. This syndrome has many different forms and can cause developmental delay, autism, and a small head size as well as other health problems.
- The *MECP2* gene is located on the X chromosome. This means that most boys have only one copy of *MECP2* and most girls have two copies of *MECP2*.
- Boys with changes in *MECP2* are very rare. Therefore, we only recommend *MECP2* testing for girls.
- Almost every case (more than 99%) of Rett syndrome is new in the child and not inherited from a mother, but testing is available to confirm this.

Genetic testing can have three possible results: positive, negative, or a result of unknown significance. If the patient has a result of unknown significance, one or both parents may be offered testing to help us understand the result.