Types of Genetic Testing

Karyotype test

- **About this test**
  - A karyotype is a picture of the chromosomes. Chromosomes are thread-like structures in the cells of the body that hold the genes.
  - This test is used to look for chromosomes that have pieces missing. It can also find extra pieces or pieces that are out of the normal order.
  - A karyotype can also show if one (1) or more entire chromosomes is missing or extra.
- **What does this test find?**
  - A karyotype shows large missing pieces of chromosomes, extra pieces of chromosomes, or pieces of chromosomes that are out of the normal order.

Fluorescence in situ hybridization test (FISH test)

- **About this test**
  - FISH testing uses fluorescent probes to look at specific areas of the chromosomes.
  - This test is used to find areas of one (1) or more chromosomes that are missing, extra, or out of the normal order. These changes may be too small to be seen on a karyotype.
- **What can this test find?**
  - FISH testing finds specific changes in chromosomes that the fluorescent probes are set to look for. It does not examine all the chromosomes, as a karyotype does.

SNP chromosome microarray test

- **About this test**
  - This test looks at all the chromosomes for any missing or extra pieces.
  - This test can show very small missing or extra chromosome pieces that the karyotype or FISH test might miss.
- **What can this test find?**
  - SNP chromosome microarray testing shows missing or extra pieces of chromosomes but it does not show chromosome pieces that are out of the normal order.

Deletion/Duplication test

- **About this test**
  - Deletion/Duplication testing looks for any missing or extra pieces within a specific gene.
• **What can this test find?**
  - Deletion/Duplication testing can find missing or extra pieces of DNA that are usually too small to be seen with karyotype, FISH or SNP chromosome microarray testing.

**Methylation analysis test**

• **About this test**
  - You inherit one (1) copy of each gene from your mother and one (1) from your father. Usually, these genes both work the same way. They are “turned on” or “turned off,” depending on the type of cell they are in. But for some genes, only one (1) copy is turned on or off. This happens in a process called “methylation.” Methylation testing checks to see if the process is happening correctly on a specific gene or chromosome.

• **What can this test find?**
  - Methylation testing can find changes in the pattern of methylation of a specific chromosome area or gene the test is set up to look at.

**Uniparental disomy test**

• **About this test**
  - Usually, you inherit one copy of each chromosome from your mother and one copy from your father. However, you can have 2 copies of an entire chromosome or two copies of a specific part of a chromosome that both come from one (1) parent. In this case, neither copy comes from the other parent. This is called “uniparental disomy” or “UPD”.

• **What can this test find?**
  - This test finds uniparental disomy on the specific chromosome or chromosome area being tested.

**DNA sequencing test**

• **About this test**
  - Sequencing reads the code for the DNA letters that make up the genes. It looks for any changes from the normal code.
  - DNA sequencing can be done on one (1) gene or on multiple genes.
  - Genomic sequencing is when all your genes are being tested at once.

• **What can this test find?**
  - This test can show if there is a change in the order of DNA letters that make up one (1) or more genes. Sequencing can also show if one (1) or a few of the DNA letters are lost or if there are extra letters.
  - Most of the time, this test does not show larger DNA areas that are missing, extra, or out of the normal order.