SNP microarray Pre-Test Counseling Information

What is a chromosome SNP microarray?

Our genetic material, which is called DNA, is organized into about 20,000 genes, which can also be called our "genome." These genes are what provide instructions for our body's growth and development by producing substances like proteins. Our genes are packaged into chromosomes with each of us typically having 23 pairs of chromosomes, or 46 chromosomes in total. One chromosome from each pair is inherited from the mother and the other from the father.

A SNP (Single nucleotide polymorphisms) microarray is a genetic test that can detect changes in a person's chromosomes. The SNP microarray test looks for changes in specific areas of a person's chromosomes, such as gains (duplications) or losses (deletions). These gains or losses result in extra or missing copies of genetic material. Changes in a person's chromosome may be associated with known genetic conditions or may cause problems with their health and development.

In addition, it allows us to identify areas in our genome that have loss of heterozygosity (LOH). Specifically, it can detect if areas in the genome are the same—such as two copies of a gene, parts of a chromosome, or two entire copies of the same chromosome –and may indicate that these are inherited from the same parent. This may increase the risk for a recessive condition. A recessive condition is when it takes alterations (mutations) in both copies of a gene to cause the condition. In addition, it may indicate the presence of uniparental disomy (UPD)—when both copies of a chromosome pair or segments of a chromosome pair are inherited from one parent and neither is inherited from the other parent. Of note, this testing may also reveal whether parents are related like cousins (consanguinity).

The SNP microarray does not detect all differences in chromosomes or DNA. For example, it cannot detect rearrangements in the chromosome that do not cause extra or missing material (balanced chromosome rearrangements) and cannot detect small spelling errors in the DNA (point mutations).

What are the possible results of a chromosome SNP microarray?

- A normal test result means that there are no detectable gains or losses of genetic material that were seen. There are also no unusual patterns of genetic similarity that can arise when two parents are closely related. However, many genetic conditions cannot be found by this test, therefore if normal, your care team will let you know if additional genetic testing is recommended

- An abnormal result means that a gain or loss of specific genetic material was found, or an area of genetic similarity was found. Sometimes this result will diagnose a well-

described genetic condition. Sometimes the size or location of the material will lead us to think that this is the cause of a person's condition. Abnormal results that are not related to the initial reason for testing can be found (for example finding a deletion associated with short stature when testing was done for structural heart disease).

- A variant of unknown significance (VUS) could mean that a gain or loss of material was found but there is not enough information about the specific genetic material to know whether this is part of normal human variation or will cause a genetic condition. If a person has this test result, we sometimes will recommend genetic testing for the parents as well to see if it was inherited or is new to the patient. This information is used to help decide if the variant is likely the cause of a person's condition.

For patients with structural heart disease

- If the patient has other physical differences, in addition to the congenital heart defect, additional genetic testing and a consult to genetics will be recommended.
- The likelihood of a positive (abnormal) genetic test result for isolated structural heart disease is estimated to be about 10%.
- Genetic testing is a choice. Your team may offer genetic testing for you or your child. The decision to proceed with testing is personal and we want families to be comfortable with possible test results and reason for testing prior to having blood drawn.