SNP Array

Pre-Test Counseling Information

A chromosome SNP array is a genetic test that is able to detect changes in a person's chromosomes.

What is a chromosome SNP array?

A chromosome SNP (single nucleotide polymorphism) array is a genetic test that is able to detect changes in a person's chromosomes. Chromosomes are the packages within cells that contain a person's genetic information (called "genes" or "DNA"). This genetic information tells a person's body how to develop and function properly. The SNP array 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 chromosomes may be associated with known genetic conditions or may cause problems with health and development. The SNP array test does not detect all differences in the chromosomes or DNA. For example, it cannot detect rearrangements in the chromosomes that do not cause extra or missing copies of genetic material (balanced chromosome rearrangements) and cannot detect small changes in the DNA (point mutations).

Chromosomes are packages in cells that contain genetic information which tells a person's body how to develop and function properly.

A chromosome SNP array looks for gains or losses in this genetic information that could cause problems with health and development.

What is the difference between this and a CGH test?

A chromosome SNP array test replaces the CGH (oligonucleotide comparative genomic hybridization) test. Just like the CGH test, a chromosome SNP array test looks for gains or losses in a person's chromosomes. Unlike the CGH test, a chromosome SNP array also looks for genetic similarity (runs of homozygosity). Genetic similarity is an area of the chromosome that does not show the normal differences we expect to see between the material passed down (inherited) from the mother and the father.

What are the possible results of a chromosome SNP array?

- A normal result means that there are no detectable gains or losses of the genetic material screened in the test panel. There are also no unusual patterns of genetic similarity. However, many genetic conditions cannot be found by this test, such as most single-gene disorders because they are often due to small changes in the DNA (point mutations).
- 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 it is the cause of a person's condition.

To Learn More

- Genetic Counseling Clinic 206-987-2056, option 1
- Your child's healthcare provider
- seattlechildrens.org

Free Interpreter Services

- In the hospital, ask your child's nurse.
- From outside the hospital, call the toll-free Family Interpreting Line 1-866-583-1527. Tell the interpreter the name or extension you need.

- A variant of unknown significance could mean that a gain or loss of specific genetic material was found. But in this case, there is not enough information about the specific genetic material to know whether or not it will cause a genetic condition. If a person has this test result, we sometimes recommend testing for parents to find out if this variant is new for the person or inherited from a parent. This information is used to help decide if this is a likely cause of a person's condition.
- A variant of unknown significance could also mean that a region of genetic similarity was found. Sometimes a genetic abnormality called uniparental disomy (UPD) is suspected. UPD happens when more chromosome material is passed down (inherited) from one parent than the other. If multiple regions of genetic similarity are found by the SNP array, that person's parents might be more closely related than originally thought (called consanguinity). For example, the parents might be related like cousins. Additional testing of the parents may help us to better understand a person's result. If a region of genetic similarity is found, it could provide a clue to a specific genetic condition and more genetic testing may be recommended.
- A SNP array could also show results that are not directly related to the reason the test was ordered. For example, the results could show that a person is at risk to develop a genetic condition that happens later in life. The results could also show that a person is a carrier for a genetic condition.

Insurance pre-authorization

Genetic tests, like the SNP array, are expensive tests that may or may not be covered by a person's insurance. Insurance plans are interested in knowing whether the SNP array is medically necessary and if or how it will affect the medical management of a person. Because of this, it is important for the healthcare provider ordering the test to clearly document the reasons for performing the SNP array in the medical record. Insurance pre-authorization is recommended for genetic testing. Without pre-authorization, some patients may have to pay for the SNP array test.

Genetic Counseling

A genetic counselor is available to discuss this information in more detail your family. To reach a clinical genetic counselor at Seattle Children's, call 206-987-2056.

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