SNP Microarray Testing Results

We found a deletion (loss) or duplication (gain) of genetic material. However, this result has UNCERTAIN CLINICAL SIGNIFICANCE.

My child's microarray result is:

Microarray testing can be ordered before or after a chromosome study. Chromosome studies can find:

- Extra or missing chromosomes
- Pieces of missing genetic material (large deletions)
- Extra pieces of genetic material (large duplications)

Microarray testing can find even smaller pieces of extra or missing genetic material (DNA).

What does this result mean?

- The loss or gain of genetic material (DNA) may lead to one or more broken genes. When the gene(s) cannot do their job, a patient may have developmental delays or other physical findings.
- Sometimes, pieces of extra or missing genetic material do not cause a broken gene. These changes are part of what makes people unique. They may be part of the reason people have different eye color or different heights. These genetic changes are called copy number variants (CNVs).
    - CNV's are common and are usually inherited from a parent
    - When a child with delays has a CNV, you will usually find the same CNV in one of the parents
    - If the parent with the CNV does not have any delays or health concerns, and the CNV is a common one, a child's delays or health problems are less likely caused by that CNV

Your child's results are uncertain. We do not know if the change we found is a CNV or a change that may be the cause for your child's health concerns.

- In some cases, we recommend testing both parents for the same CNV
- If one parent has this CNV, it is less likely to be the cause of your child's health problems
- Your child's doctor can order this test for both parents through our laboratory
- In some cases testing the parents will not help us to figure out if the CNV is causing the child's health problem. In these cases we will not recommend testing the parents.

This type of genetic testing is new. There is still a lot we do not know about how very small losses or gains of genetic material change us. This is why some results are not clear and are reported as UNCERTAIN CLINICAL SIGNIFICANCE.

What did this test look for?

This test may help diagnose the cause of health problems for a child who does not have a diagnosis. SNP microarray test can look for many different genetic syndromes. This test can find conditions known as:

- microdeletion syndromes (small missing piece of genetic material)
- microduplication syndromes (extra piece of genetic material)
- subtelomeric deletions (loss of material at end of chromosome).

More testing is sometimes needed to rule out a specific condition.

What did the test NOT look for?

- Some changes in genetic material do not result in missing or extra pieces of DNA. Microarray testing will not find rearrangements of genetic material.

- Genetic conditions are not always caused by extra or missing genetic material. Often a small change (point mutation) in a single gene can cause a health problem. These small changes cannot be found using microarray.

    - Other testing may help if a specific genetic syndrome is suspected

    - Syndromes are most common when there is more than one health problem

- When only a small number of cells carry extra or missing material, it is called mosaicism. Mosaicism will not be found if the number of cells with these changes is too small.

- It is not always possible to find genetic changes that explain a child's health problems.

What questions do I have?