SNP Microarray Testing Results

We found a deletion (loss) or duplication (gain) of genetic material. This is an ABNORMAL result.

My child's microarray result is:

Microarray testing is often ordered 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.
- The microarray report will give you information about your child's specific genetic change.
    - Look for this information under "Clinical Significance" in your report.
- Some genetic changes are common and your doctor may give you a general idea of the health concerns related to this change.
- Not all children with the same or similar genetic change have the exact same health issues.
- Some genetic changes are new or unique and not much information is known.

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.

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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.

- 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.

- 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.

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

What questions do I have?