

Array Comparative Genomic Hybridization (Array CGH)

Understanding Your Genetic Test Result: Detection of a deletion and/or a duplication of genetic material

Your test result:

What does this test result mean?

Array CGH tests for imbalances in a person's genetic material (DNA). In the cells of the body, genetic material is organized into packages called chromosomes. Array CGH is a type of chromosome test that can determine whether or not there are any pieces of genetic material missing (deleted) or extra (duplicated). In most cases, we have two copies of our DNA - one copy is inherited from our mother and the other copy is inherited from our father. Finding an imbalance means there is either more or less than the usual amount of genetic material due to a deletion or duplication in the DNA. This result may also be called a "copy number variant."

When an imbalance (or copy number variant) is detected, the test result tells you:

- which chromosomes are involved
- the size of the deletion or duplication

What happens when there is deleted or duplicated genetic material?

When there is deleted or duplicated genetic material, it can affect a person's health and development. However, the type and severity of health issues varies. The effect of the imbalance depends on how much genetic material is missing or extra and which specific genes are involved. In some cases, a genetic imbalance can lead to physical birth defects, medical health problems, intellectual disabilities, and behavior difficulties. However, some deletions and duplications have few, if any, effects on a person's health and development.

How do I know what medical problems might happen based on my result?

Some deletions and duplications have been seen before and are known to cause specific medical problems. When a genetic imbalance is known to be related to health problems, it is called "*pathogenic*." This also means there is usually enough information to know what kind of health and development problems to expect. However, not everyone with the same genetic imbalance will have the exact same medical problems.

Other deletions and duplications are rare. There may be no one else known to have exactly the same imbalance or there may be very little information about health problems in others with the same result. This type of imbalance is called a "*variant of uncertain significance*". In this situation, it is hard to predict the type of medical problems or developmental problems to expect. However, we are still learning about deletions and duplications. So even if the information about your specific result is limited right now, we will continue to learn more as time goes on.

Where did the deletion or duplication come from?

Deletions and duplications usually happen as random changes in the DNA when an egg or sperm is being formed. It is permanent part of the genetic make-up of that person and cannot be fixed or changed. In some cases, the deletion or duplication is inherited from a parent. Whether an imbalance happened randomly or was inherited from a parent, it is **never** caused by exposure to any medication, injury, or illness either before or after birth.

What else should I think about?

It can help to recognize who else may benefit by knowing this result. There may be medical reasons to share this test result with others in your family. Your genetic counselor can review your family history and discuss what this result might mean for your relatives. Your genetic counselor can also help you figure out the best way to share the result with family members.

Resources:

The following websites provide support and information to families and individuals affected by a rare chromosome disorder.

Chromosome Disorder Outreach (CDO) - www.chromodisorder.org

P.O. Box 724

Boca Raton, FL 33429-0724

FamilyHelpline (561) 395-4252

Email: info@chromodisorder.org

Unique – www.rarechromo.org

PO Box 2189

Caterham

Surrey

CR3 5GN

United Kingdom

Tel: +44 (0)1883 330766

Email: info@rarechromo.org

Kaiser Genetics Departments

| | |
|---------------|----------------|
| Fresno | (559) 324-5330 |
| Modesto | (916) 614-4075 |
| Oakland | (510) 752-6298 |
| Sacramento | (916) 614-4075 |
| San Francisco | (415) 833-2998 |
| San Jose | (408) 972-3300 |