

NIMGenetics Array-CGH

What is the Array-CGH?

The Array CGH is a technique used in genetic diagnosis that allows the analysis of an individual's complete genome in the search for alterations of gains or losses of genetic material, in less than two weeks.

Array-CGHs used by NIMGenetics are backed by Agilent technologies, world leader in the development of oligonucleotide platforms. These Array-CGHs allow the detection of deletions or duplications of genomic material with an approximate resolution of 200Kb.

Why use this technology for Genetic diagnosis?

Approximately 0.6% of the babies born have unbalanced chromosomal changes detected through analysis of the conventional karyotype. The Array-CGH has been recently introduced to identify chromosomal alterations in patients with pathology, but without alterations in the karyotype. During this time it has been shown that this technology is more sensitive and efficient than the conventional karyotype, in the detection of chromosomal duplications or deletions, since the resolution of Array-CGHs is 10 times higher than the one for a conventional karyotype (which has a maximum resolution of 5 Mb).

NIMGenetics provides platforms with 44,000, 105,000 or 244,000 oligonucleotide probes that cover the complete genome. The selection of the most appropriate platform must be done individually, depending on the objective of the study indicated for each patient. The designs of Agilent Technologies's Array-CGHs are specially geared towards the detection of alterations with clinical repercussions. NIMGenetic's experience in the identification of variations of a polymorphic origin (variations that show up in the general population) minimizes the risk of obtaining results of difficult clinical interpretation.

From the cost/benefit point of view, it is necessary to keep in mind that this technology is equivalent to hundreds of combined FISH trials, throughout the whole genome. Thus, the Array-CGH must be considered as a first alternative in the study of gains or losses of genomic material.

How does the Array-CGH work?

The Array-CGH allows the comparison of the number of DNA copies between the sample to be tested and a healthy control.

Both DNAs are marked with different colours and hybridized jointly in the array. The analysis is done quantitatively, calculating the proportion of DNA of each colour for each genomic region of the case under study with respect to the healthy control.

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Gains or losses detected are analyzed to rule out the presence of these variations in the healthy population. In addition, different databases are consulted to determine the relationship between the patient's clinical profile and the alterations observed. In some cases, the presence of alterations not previously described calls for a study of the parents to rule out the possibility of an inherited alteration that is not responsible for the patient's clinical profile.



Finally, before requesting this test, it is important to take into account the limitations of this technology. With the Array-CGHs it would not be possible to diagnose those alterations that do not imply a loss or gain of genomic material, such as: specific mutations, translocations or balanced inversions. In addition, those duplications or deletions lower than the resolution range of the platform used or alterations present in mosaic (in less than 40% of the cell population), which could be the cause of the patient's pathology, would also be non-detectable.

Indications for the use of the Array-CGH

The Array-CGH is indicated in patients with a normal karyotype and with a clinical profile with:

- Unexplained mental or developmental retardation.
- Congenital anomalies or dysmorphic traits.
- Autistic disorders or clinical presentations that suggest a specific chromosomal syndrome.

In addition, it is indicated in altered karyotypes:

- In patients with apparently balanced translocations with an abnormal clinical phenotype. The Array-CGH may detect cryptical deletions or duplications in those regions.
- In the presence of duplications or deletions in the karyotype to determine the limits of the altered region.
- When marker chromosomes are identified, to determine their origin.

An expert team, a safe value.

NIMGenetics guarantees a quick and safe diagnosis, made possible by a multidisciplinary team with experts in different fields within genomics and clinical genetics.

The staff at NIMGenetics is at your disposal to resolve queries or provide any further information.
info@nimgenetics.com