

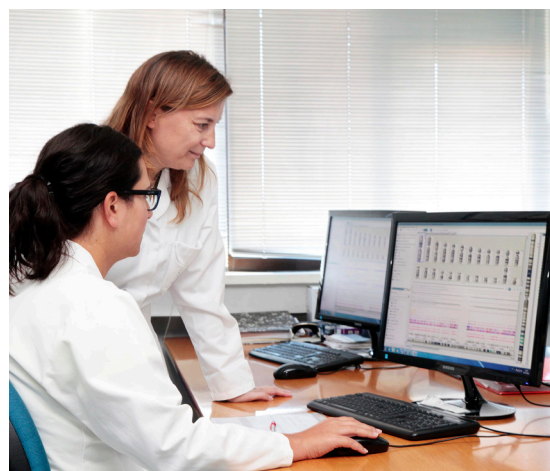
ARRAY CGH

Information for health professionals



CHROMOSOMAL MICROARRAY ANALYSIS USING ARRAY CGH

Chromosomal abnormalities are the major cause of psychomotor development delay. Traditional cytogenetic techniques such as karyotyping or FISH have been used for decades but are limited by low resolution qualitative results. The microarray chromosomal analysis is a very high resolution technique that provides a genome-wide approach. The implementation of CGH testing in standard clinical practice started in 2007. Since 2009 the scientific evidence, brought consensus for the current use of chromosomal microarray analysis as a first tier approach for psychomotor development delay, autism or polymalformative syndromes, when the clinical observation does not suggest a specific genetic syndrome. Array CGH testing is replacing karyotyping, due to its increased diagnostics rates (15% CGH vs 2% karyotype).



CGC Genetics, a medical genetics laboratory with over 20 years of experience in cytogenetics, provides chromosomal microarray analysis using the most comprehensive and robust technology available (CytoScan®, Affymetrix®).

The solutions offered by CGC Genetics include a very high analytical resolution together with the clinical interpretation of results carried out by our Genetic Medical Team. This alliance of different resources allows us to meet the higher European quality standards in the field of Medical Genetics.

This technology is characterized by:

- High resolution, with a total number of markers of 750.000 and 2.700.000
- High density of markers in regions of clinical interest (ISCA, OMIM and RefSeq genes)
- Highest gene coverage: 100% of ISCA genes, 98% of OMIM genes and 96% of RefSeq genes
- Combination of non-polymorphic and SNP marker
- High density SNPs allow the detection of low level mosaicism, loss of heterozygosity (LOH) and uniparental disomy (UDP)
- Double confirmation of the copy number variants (CNV) by the combined analysis of both types of markers

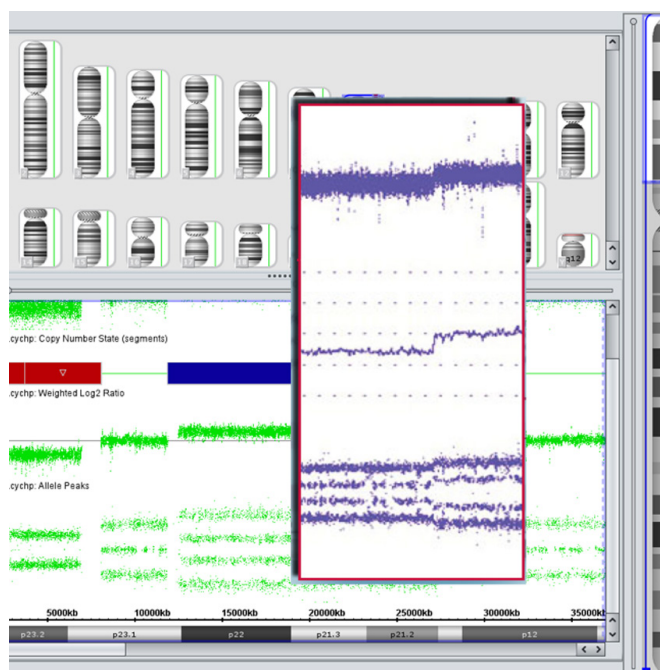
CGC Genetics can offer two different resolutions, where the highest resolution has the greatest diagnostic capability:

• **ARRAY CGH CYTOSCAN® 750K (HIGH RESOLUTION)**

- 550.000 oligonucleotides for detection of DNA losses and gains
- 200.000 SNPs for detection of loss of heterozygosity (LOH) and uniparental isodisomy (UPD)

• **ARRAY CGH CYTOSCAN® HD (VERY HIGH RESOLUTION)**

- 1.950.000 oligonucleotides for detection of DNA losses and gains
- 750.000 SNPs for detection of loss of heterozygosity (LOH) and uniparental isodisomy (UPD)



Array CGH available at CGC Genetics uses Affymetrix platform, a combined platform which allows to detect, besides DNA losses and gains, LOH which are highly important in diagnosis of recessive disorders and in cases of uniparental disomy.

Affymetrix platform used by CGC Genetics is the only FDA-validated platform for use in diagnosis of psychomotor delay, autism and polymalformative syndromes.

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