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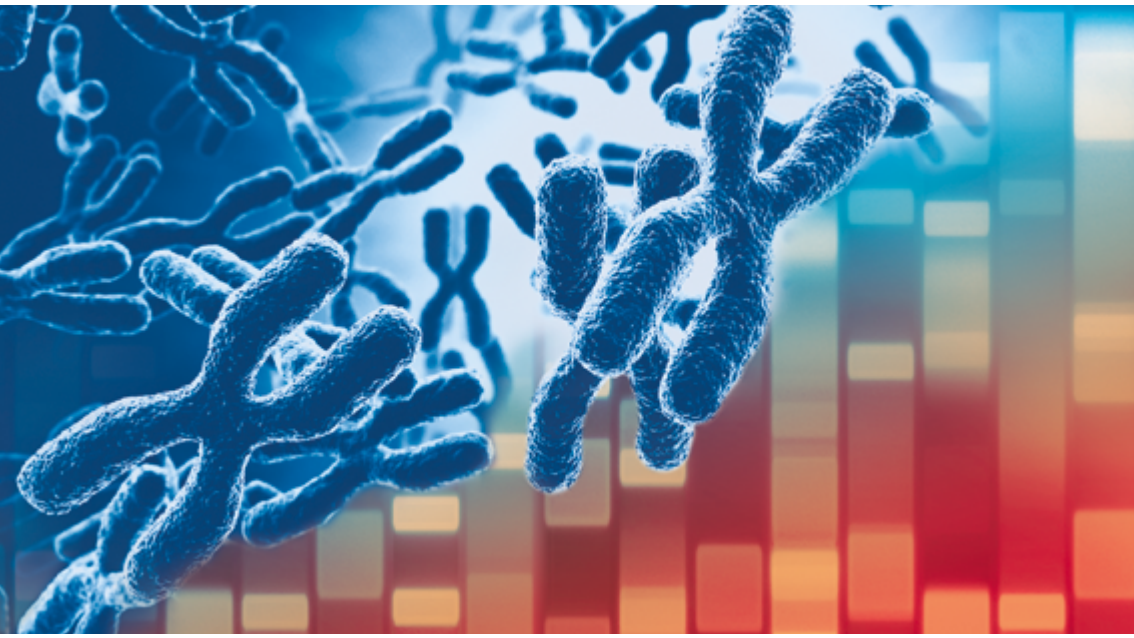

# Chromosomal microarray analysis by SNP-array

*(single nucleotide polymorphism - array)*

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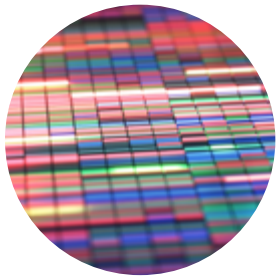


Chromosomal Microarray Analysis (CMA) is one of the techniques for studying chromosomes. It can detect unbalanced chromosomal abnormalities (copy number gain or loss, known as “copy number variation” or CNV) across the whole genome.

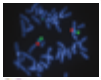
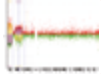
Compared to the conventional karyotype, the resolution is higher, which increases the diagnostic sensitivity in the search for CNV’s.

Unlike FISH (*Fluorescence in Situ Hybridization*), CMA allows the study of the whole genome with a single test.

Two types of chips exist:

- **Array CGH** *(comparative genomic hybridization array)* allows analysis of signal intensity, to detect CNV’s
- **SNP array** *(single nucleotide polymorphism array)* allows analysis of signal intensity and genotyping data

The table below summarises and compares the main characteristics of the conventional karyotype, FISH, array CGH and SNP array.

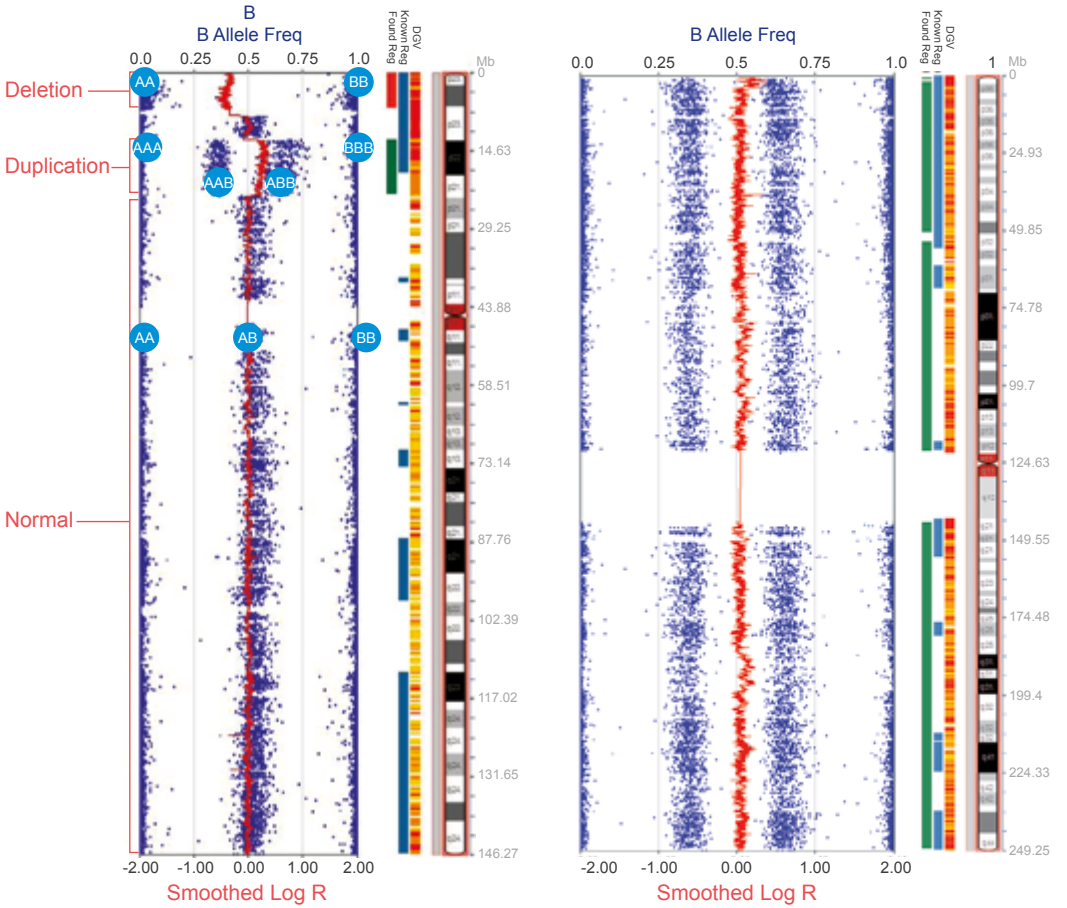
| | Method | Resolution | Coverage | Detection of unbalanced anomaly | Detection of balanced anomaly | LOH | Detection of triploidies |
|---|---|---|---|---|---|---|---|
|  | Karyotype | 5-10 Mb | Whole genome | YES | YES | NO | YES |
|  | FISH | 150-200 kb | Specific probe | YES | YES | NO | YES |
|  | Array CGH | 30-100 kb | Whole genome | YES | NO | NO | NO |
|  | SNP array | 30-100 kb | Whole genome | YES | NO | YES | YES |

# SNP array or array CGH

Like array CGH, SNP array is used in DNA-chip based chromosomal analyses (CMA). The SNP array allows the whole genome to be studied.

Unlike array CGH which uses patient versus control competitive hybridization (gains or losses compared to the control), in SNP array, each probe binds to a patient's complementary DNA sequence, without competition. This generates a signal, which will be decreased in the event of copy loss (deletion) and increased in the event of copy gain (duplication, triplication, etc.). In addition, SNP array also uses genotyping data: the probes are located at polymorphic nucleotides of the genome and undergo single base extension (SNP: *single nucleotide polymorphism*). This confirms copy losses and gains but also detects loss of heterozygosity (LOH) and triploidies.

**Example of profiles (complex CNVs and triploidies)**



The copy number variations (CNV) thus identified are then classified into:

pathogenic - probably pathogenic - **VOUS** - probably benign - benign

Benign and possibly benign CNVs are not indicated in the reports. Whether the VOUS are reported depends on whether it is an antenatal or postnatal study, according to their size and gene content and according to the clinical context.

## Advantages of SNP array

- **Study of the whole genome** in the same way as the standard karyotype
- Covers regions of the genome responsible for **recurrent syndromes** (DiGeorge, Prader-Willi / Angelman, Wolf-Hirschhorn…)
- Identifying **small variations** (starting from 30 kilobases, compared to 5-10 megabases for the standard karyotype)
- Identification of **loss of heterozygosity** (LOH)
- **Detection of triploidies**
- **Direct DNA extraction** possible without cell culture step (recurrent culture failures of miscarriage products for the conventional karyotype)
- **Quick result** (DNA extraction possible on fresh sample, turn around time 3 days)
- **Visualisation** of partial maternal contamination.

**VOUS : Variant Of Unknown Significance*

# Practical details

| Indication | | Type of sample |
|---|---|---|
| **Prenatal (resolution 1Mb)** | | |
| First line analysis | Characterisation of chromosomal alteration identified by a standard karyotype:<br>• chromosome marker<br>• apparently balanced de novo chromosomal alteration or inherited with abnormal ultrasound findings<br>• unbalanced or complex chromosomal alteration<br>Abnormal ultrasound findings | • amniotic fluid<br>• chorionic villi<br>• foetal blood<br>• product of conception<br>• foetal tissue<br>+<br>• maternal EDTA blood sample (maternal contamination) |
| Other | • maternal serum markers<br>• NIPT unsuccessful<br>• fetal deaths / Product of conception | |
| **Postnatal (resolution 200kb)** | | |
| First line analysis | • neurodevelopmental disorders<br>• malformation syndromes | EDTA blood |
| Other | • reproductive disorders<br>• growth abnormalities<br>• isolated malformation<br>• verification of CNV detected by another technique<br>• characterisation of chromosomal alteration identified by standard karyotype | |
| Turnaround | 3 weeks | |
| Documents and information to be provided | • Order form B3-INTGB (Prenatal) or order form B12-INTGB (Postnatal)<br>• Informed consent form signed by the patient and the prescriber D44-INTGB<br>• Clinical details | |
| Cost | Contact us | |
| Other resources | Information sheet for patients - N24-INTGB | |

Find all the information and documents relating to the analyses offered by the laboratory at **www. eurofins-biomnis.com > Test Guide.**

## For more information, get in touch with your designated contact persons

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DS21-INTGB – February 2020