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Opinion

What are the Indications for Microarray Test?

Jorge Sales Marques*

Pediatric Department, Centro Hospitalar Conde S. Januário, Macau, China

***Corresponding Author**: Jorge Sales Marques, Pediatric Department, Centro Hospitalar Conde S. Januário, Macau, China.

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Microarray or array CGH, allows the detection of gains or losses of genetic material of smaller quantity (compared to the karyotype) - submicroscopic changes (microdeletions and microduplications).

It explores the ability of the DNA molecule to specifically bind to another DNA molecule (A = T and G = C). It consists of thousands of small DNA sequences ("probes") arranged on a plate called a chip. The patient's DNA is "digested" (separated into small fragments) and these fragments being associated with a flourishing marker. DNA reference (from a pool of people without genetic alterations) is also associated with a flourishing marker of different color. The reference DNA and the patient's DNA are mixed and applied on the chip and hybridization is performed (Figure 1).

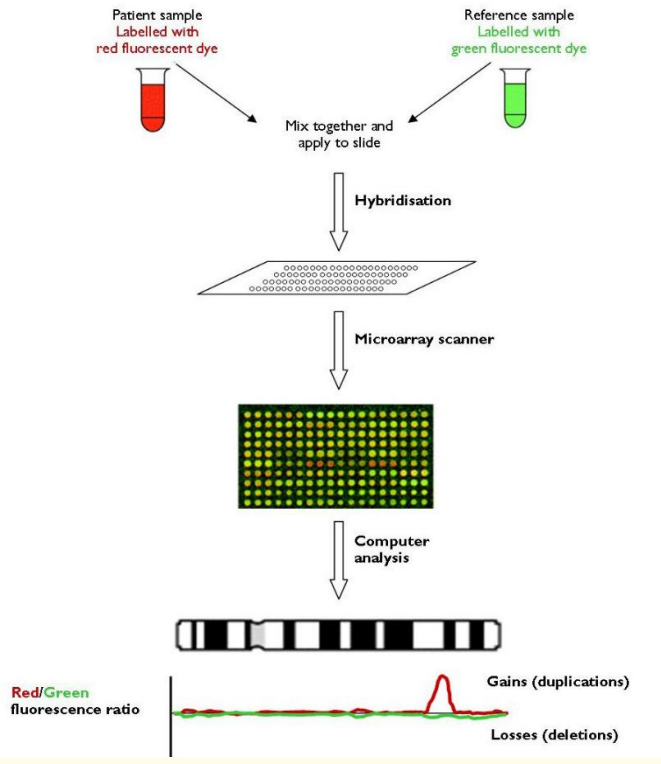

Figure 1: *How microarray works.*

There are different types of microarray:

1- BAC (bacterial artificial chromosome clones) - small human DNA sequences 80-200 kbs, may not detect small changes. Less likely to detect changes of uncertain clinical significance.
2- oligo (oligonucleotide probes)-small sequences of artificial DNA 60kbs, detect smaller changes. Most likely to detect changes of uncertain clinical significance.

What are the advantages and disadvantages to do microarray?

The advantages are ability to explore the 46 chromosomes in a single test and detect gains or losses of genetic material more accurately than the karyotype, being able to detect which specific genes are included in these duplications or deletions. It allows detecting these changes when there are no clues to which chromosome may be affected (therefore not applying other techniques, such as FISH). It allows to detect some cases of mosaicism and more precisely specify genetic imbalances detected in the karyotype.

Microarray, does not detect: balanced chromosomal rearrangements (such as balanced translocations and inversions), since it does not result in loss or gain of material. Also, cannot detect some types of polyploidy, such as triploidy. In these cases, the karyotype for detection is still required. Other situations that doesn't detect are: punctual mutations of base pairs that may also be responsible for genetic pathologies, mitochondrial DNA alterations, chromosomal changes in mosaic (only less than 30%).

What are the different between karyotype, FISH for subtelemeric rearrangements and microarray?

1 - Karyotype: it can detect genomic changes of 3 million base pairs (Mb). It is often not to detect changes of 5-10 Mb depending on the region involved and conditions of the sample. Excluding trisomy 21, the karyotype only detects genetic changes in <3% of individuals with intellectual deficit. It is based on the subjective analysis of gain or loss of material (interpersonal and interlaboratory variation). Is better than the microarray for detection of balanced rearrangements and low grade mosaicism.
2 - FISH for subtelomeric rearrangements: it detects changes in 2.4-2.6% of the patients studied. It detects changes of 100 kb (10 to 100 times smaller than the karyotype)
3 - Microarray: it detects changes of 100-250 kb (10 to 100 times smaller than the karyotype) throughout the genome. It detects changes of 20-50kb when directed to a specific region. Also, detect changes in 11-15% of patients with normal karyotype.

According to the American College of Medical Genetics – 2010, is the first line of postnatal research for: development delay, mental retard, autism spectrum disorders and multiple congenital anomalies that do not characterize known genetic syndromes (Figure 2).

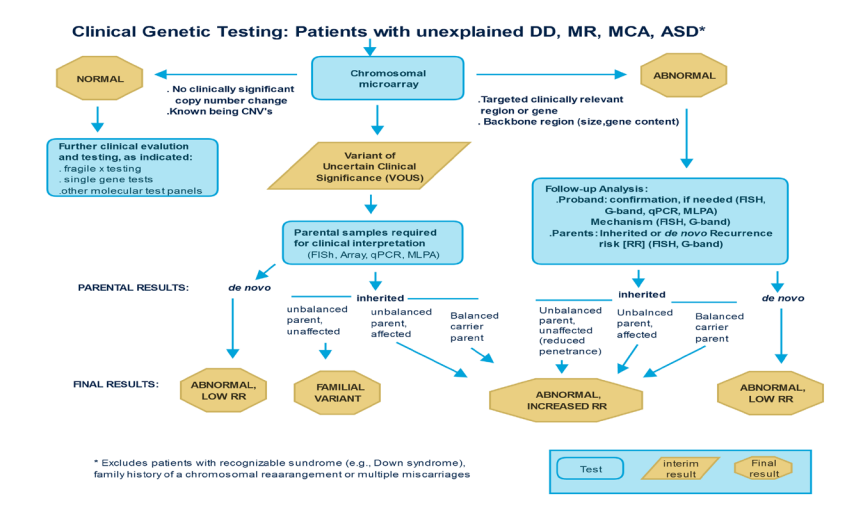

Figure 2: *Flow chart of microarray in development delay (DD), mental retard (MR), autism spectrum disorders (ASD) and multiple congenital anomalies (MCA).*

If we find the etiology, we can establish a cause, predict the functional impact and prognosis, change approach, influences prevention strategies, identifies conditions that require treatment and provide genetic counseling to the family (Table 1).

Age (years)	Gender	Main features	Diagnosis
3	M	Global delay	Duplication 13q12.12
16	F	Autistic features, mental retardation	Mosaic trisomy 8
4	F	Global delay	Williams syndrome
4	F	Global delay, VSD, PDA	Deletion 15q25.2
1	F	Global delay, short stature	Mos46, Xidic(X)/45, X
4	F	Global delay, aortic supravalvular stenosis	Williams syndrome
1	F	Global delay	47 XXX
4	F	Global delay, short stature, obesity	Prader Willi syndrome
4	F	Global delay	Duplication 21q11.2
15	F	Global delay, mental retardation, deeply set eyes, microbrachycephaly	1p36 syndrome

Table 1: *Some examples of cases diagnosed by microarray.*

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