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Integrative Review

Review of 117 Microarray Tests During a Period of 18 Months

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Abstract

Microarray or array CGH, detected gains or losses of genetic material of smaller quantity.

We reviewed 117 microarray tests that were requested by pediatric doctors during a period of 18 months, with the objective to verify the number of positive results and later analyze each of those results.

We found 20 tests (17%) with chromosome abnormalities. Microarray is a first line postnatal research of development delay, mental retardation, autism spectrum disorder and non-syndromic multiple congenital disorder.

Keywords: *Microarray; Genetic; Indications*

Introduction

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).

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 and chromosomal changes in mosaic (only less than 30%).

Purpose

The aims of this study are two: 1 - verify the number of positive results in microarray test after a period of 18 months 2 - review all positive results during this period.

Material

We checked all microarray tests that were requested during a period of 18 months (1/2/2016 to 31/8/2017).

Criteria of inclusion: All test requested by pediatrics doctors.

Criteria of exclusion: All test that were asked by Prenatal Diagnostic Centre (PDC).

Methods

All microarray tests were done in Tsan Yuk hospital, Prenatal laboratory, using Perkin Elmer CGXTM V2.0 (60k oligonucleotide array), resolution: 200 kb.

Results

In 2016, a total of 30 tests were done and in 2017, these numbers increased to 87.

The total array tests during 18 months were 117 (Figure 1).

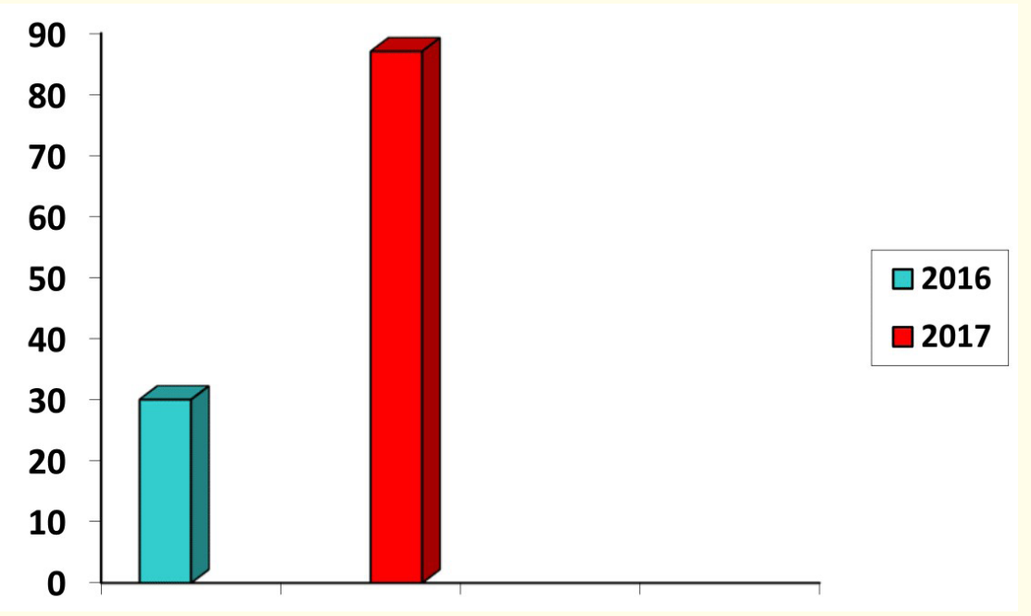

Figure 1: *Year distribution of microarray test.*

We found 20 cases (17%) with positive results in microarray test (Figure 2).

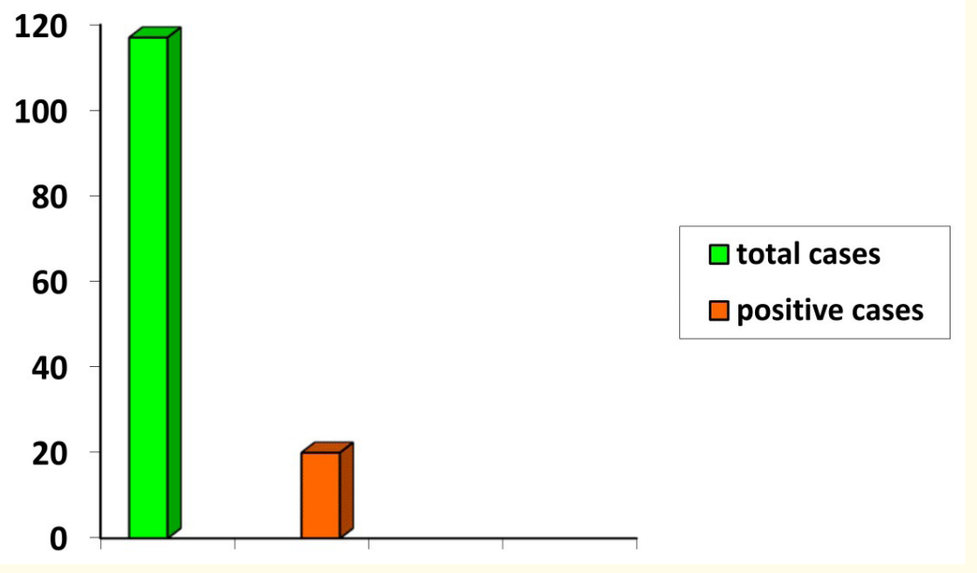

Figure 2: *Total of positives cases.*

All 20 cases fulfill the ACMG criteria (Table 1).

Cases	Development delay	Mental retardation	Autism spectrum disorder	Non-syndromic multiple congenital disorder	Microarray test result
1	X	X			duplication 13q12.12
2	X			X	duplication 14q32.2-q32.3
3	X			X	1p22.3-p13.3
4	X	X	X		15q11.2-q13.1
5	X				47XXX
6	X	X	X		trisomy 8
7	X	X	X		16p13.11-p12.3
8	X				7q11.23
9	X			X	46,xx,der(18)
10	X				mos46,X,idic(X)(p22.3)(44)/45,X(6)
11	X				7q11.23
12	X				45,X(1)/46,XX(59)
13	X				47XXX
14	X			X	deletion 10q22.1-q22.3
15	X				7q11.23
16	X	X		X	deletion 7q21.3-q22.1
17	X				Ap4B1 heterozygous
18	X				monosomy X
19	X	X	X		duplication 6p21.3
20	X				duplication 21q11.2

Table 1: *All cases detected by microarray.*

Discussion

Microarray detects changes of 100 - 250 kb (10 to 100 times smaller than the karyotype) throughout the genome. It detects changes of 20 - 50 kb when directed to a specific region. Also, detect changes in 11 - 15% of patients with normal karyotype.

Excluding trisomy 21, the karyotype only detects genetic changes in < 3% of individuals with intellectual deficit.

If the patient presented with mental retardation with suspicious of X-fragile syndrome, we need to performed molecular study of X-fragile for the confirmation.

Nowadays, the indications for microarray tests are according to the American College of Medical Genetics - 2010 (ACMG), in which there are 4 indications as first line of postnatal research for: development delay, mental retard, autism spectrum disorders and multiple congenital anomalies that do not characterize known genetic syndromes.

If positive, we can offer a prenatal diagnosis and genetic counseling for the couple in the future pregnancy.

Our data showed that the numbers of request has triple in 2017 and only we included the first six months of the year. These results means that our pediatric doctors are using regularly the request of microarray tests according to the ACMG recommendations.

In our study, we found 17% of cases with positive results, some of them could be diagnosed by simple karyotype or FISH study. These happened because microarray is the first indication for the above clinical signs. On the other hand, is important to remember that microarray cannot diagnose balanced chromosomal rearrangements (such as balanced translocations and inversions). In these situations, karyotype is required for the diagnosis.

All of the 20 cases diagnosed by microarray, showed in their presentation development delay. Around 33% presented with mental retardation, 25% with autism spectrum disorder and 20% of cases with non-syndromic multiple congenital disorder [1,2].

Conclusion

Our study confirmed that microarray is an important test for genetic diseases of patients with development delay, mental retard, autism spectrum disorders and multiple congenital anomalies that do not characterize known genetic syndromes.

Bibliography

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