416

THE CLINICAL IMPLICATION OF ARRAY-CGH (AGILENT 244K AND 4X180K) ANALYSIS IN PEDIATRIC SERVICES

K. Kosma, M. Tzetis, S. Kitsiou - Tzeli, H. Fryssira, K. Giannikou, V. Oikonomakis, A. Syrmou, E. Kanavakis

Medical Genetics, University of Athens, Medical School, Aghia Sophia Childrens Hospital, Athens, Greece

Background and aim: Array comparative genomic hybridisation (aCGH) is used to detect small copy number changes within the genome that are not always visible by conventional karyotyping.Patients with mental retardation (MR), developmental delay (DD), autism, or/and multiple congenital anomalies (MCA) may have normal standard karyotype and not always can be classified as distinctive syndromes. To detect submicroscopic chromosomal aberrations 253 patients with various degrees of MR, DD, autism, and/or single or multiple congenital abnormalities and normal previous conventional karyotype, many of which had also received a variety of other genetic tests (FRAX, RETT, single FISH tests or metabolic screens), were analyzed with aCGH.

Methods: Agilent 244K & 4x180K was performed allowing a theoretical resolution of >50Kb.

Results: Clinically significant submicroscopic imbalances were detected in 92 (~30%) patients. Some patients presented with more than one aberration relevant to their phenotype. The high percentage of positive patients is probably due to the strict criteria of patient selection.

number of Patients	Chromosome	Gain/ Loss	start/ end UCSC hg18	Length Range (Mb)
7	1p36.33-p36.32	4 L/ 3 G	554268->6141121	0.69->5.6
1	1p31.2-p22.3	1 L	69154817->87595434	18.44
1	1q21.1	1 G	145009467->146212234	1.2
1	1q44	1 L	242886313->244525547	1.64
1	2p16.3	1 L	51090504->51167934	0.077
2	2q24.2-q31.1	2 L	163364586->170720574	2.1->7.4
1	2q37.3	1 L	238465588->242690037	4.2
1	3p25.3	1 L	9499872->11406036	1.9
3	3p14.1	3 L	70598263->71601477	0.083->1.2
3	5p15.33	3 L	934536->2209449	1.01->1.26
4	5p15.2	4 L	11095452->11368310	0.034->0.052
1	5q23.2-q31.1	1 L	124232611->135251538	11
2	5q33.1	2 L	149716211 ->150082767	0.015->0.37
1	7p22.2-22.3	1 L	250232->2801328	2.55
3	7p22.2-p22.1	2 G; 1 L	4340544->7025368	1.7->2.7
2	7q11.23	2 L	72022051->73923281	0.029->1.9
5	8q24.23-q24.3	4 L, 1G	138221526->146250824	3.7-8
1	8q24.3	1 L	145944589->146264902	0,413
1	8q21.11-q21.13	1 L	77016764->80392452	3.4
1	9p24.3-p22.1	1 G	194193 ->19203881	11,0
1	9q31.2	1 L	107304497->109588754	2.3
4	9q34.2-34.3	4 L	134831651->140241935	0.7-5.4
1	13q33.1-q34	1 L	100510439 ->114114568	13.6
4	15q11.2-q14	2 L; 2 G	18362555->36837570	5->17.8
7	15q11.2	5 L; 2G	18683110->25373779	0.02->2.1
2	15q21.3-q22.31	2 L	54118678->62432440	8.13
5	16p13.3	4 L; 1 G	70350->3229290	0.08->2.9
1	16q21-q22.1	1 L	64772843->66806006	2.03
2	17q12	2 L	31474518->34217217	1.1->2.7
5	17q21.31-q21.32	3 L, 1 G	41288843->42142422	0.25->0.8
2	18p11.32-p11.21	2 L	4316->15370683	9.6->15.3
2	20q13.3	2 L	61433519->62419593	0.29->0.99
1	22q11.1-q11.21	1 G	15438946->17041773	1.6
1	22q11.21-q11.23	1 L	20128705->21984222	1.86
1	22q11.21	1 L	17299942->19794119	2.5
1	Xp22.32	1 G	5818688->5918660	0.1
3	Xp22.31	3 G	6477006->8124803	1.6
1	Xp11.3	1 L	43208140->43765770	0.56
1	Xp11.22	1 L	53238335->53251848	0.013
2	Xq13.3	2 L	74410307->74516096	0.041->0.1
2	Xq28	2 L	151370278->153127086	1.5
1	Xq28	1 L	153248667-> 153335811	0.087

[Abnormal Chromosomal Microarray Results]

Conclusions: Array CGH is a powerful tool for the identification of novel chromosomal syndromes and for more accurate prognosis and phenotype-genotype correlations.