

a first-tier test for all pregnancies and stand-alone test for specific clinical.

13.P32

9q34.3 microdeletion by MLPA in a fetus with cardiac defects

*Bárbara Marques*¹, Cristina Ferreira¹, Filomena Brito¹, Cristina Alves¹, Lucilia Carvalho², José Furtado¹, Catarina Ventura¹, Catarina Silva³, Marisa Silva¹, Laurentino Simão¹, Joaquim Correia⁴, Hildeberto Correia¹

Instituto Nacional de Saúde Doutor Ricardo Jorge IP Unidade de Citogenética, Departamento de Genética Humana Lisboa-Portugal¹ Hospital Egas Moniz, Centro Hospitalar de Lisboa Ocidental, EPE Unidade de Fetopatologia, Serviço de Anatomia Patológica Lisboa-Portugal² Instituto Nacional de Saúde Doutor Ricardo Jorge IP Unidade de Tecnologia e Desenvolvimento, Departamento de Genética Humana Lisboa-Portugal³ Centro Hospitalar de Lisboa Central, EPE Centro de Diagnóstico Pré-natal, Maternidade Dr. Alfredo da Costa Lisboa-Portugal⁴

Submicroscopic deletion of 9q34.3 is a genomic rearrangement disorder that affects fetal development and results in mental retardation and multiple congenital anomalies. Heart defects are present in 40–45 % of patients and include structural anomalies, namely atrial septal defects, ventricular septal defects, valve anomalies comprising pulmonary valve stenosis and bicuspid aortic valve, persistent open foramen ovale and ductus arteriosus anomalies. The subtelomeric deletion syndrome caused by this chromosome rearrangement is one of the most common subtelomeric deletion syndromes and some authors suggest that its phenotypic features overlap with DiGeorge syndrome.

We report a prenatal diagnosis performed at 20⁺ weeks gestation due to fetal cardiac defect and absence of nasal bone. Molecular rapid aneuploidy testing yielded normal results but DiGeorge/Velocardiofacial Multiplex Ligation-dependent Probe Amplification (MLPA) analysis revealed a deletion on 9q34.3 comprising the EHMT1 gene, confirmed by fluorescence in situ hybridization with a subtelomeric probe for 9q34.3. Cytogenetic analysis results were also consistent with a terminal deletion. The couple opted for medical termination of pregnancy. Post-mortem examination of the 24-

weeks fetus revealed mild facial dysmorphism, bilateral suprarenal hypoplasia and a complex cardiopathy showing right implantation of truncus arteriosus, ventricular septal defect, large right cavities and hypoplastic left cavities. In order to establish a more accurate fetal genotype-phenotype correlation, breakpoint mapping by microarray is underway.

To our knowledge, this is the second case reported of prenatally diagnosed 9q34.3 deletion and the first associated with cardiac defects. This case reinforces the importance of using MLPA methodology in fetuses presenting with cardiopathy, since this approach allows analysing all known low copy repeats at 22q11.2 and several other regions associated with cardiac anomalies.

13.P33

Importance to realise the conventional cytogenetic study as a complement of the QF-PCR in cases of numerical anomaly suspicion.

*Jose Manuel Cabada*¹, Gemma Moreno¹, Alberto Alvarez¹, Adrian Alcalá¹, Jonathan García¹, Meritxell Piferrer¹, Yolanda Alvarez¹, Lorena Ripollés¹, Laia Puig¹, Montserrat Peris¹, Naima Alaoui¹, Cristina Fernandez¹, Rosella Soler¹, Mari Carmen Sanchez¹, Dolores Carnerero¹, Eulàlia Solsona¹

Balagué Center Cytogenetic Unit Hospitalet de Llobregat-Spain¹

The economic reasons combined with the improvement of the non invasive markers for the screening of fetal chromosomal anomalies has reduced the number of invasive determinations in the majority of European countries.

With the objective to reduce still more the costs, it has suggested the option to combine echography and QF-PCR without need to realise the conventional karyotype in cases of numerical chromosome alteration suspicion.

In the last three years we had in our laboratory 621 cases (of a total of 12730 determinations) with chromosomal alterations in amniotic fluid samples. All these cases referred by echographic alterations and or high risk in biochemical screening for Down's Syndrome.

Of the total of cases with chromosomal alterations, 497 were numerical and the rest of them were structural.