morphogenesis. Their prevalence is estimated between 0.4 and 0.6% of live births. CHD is essentially multifactorial. Among the genetic causes, chromosomal aberrations are involved in congenital heart disease. Indeed, among patients who carry chromosomal abnormalities, 30% have cardiovascular problems. 22q11.2 microdeletion is the most common cause.

The purpose of this study was to determine whether subtle chromosomal anomalies previously undetected by conventional cytogenetic banding methods could be identified by array-CGH in children with isolated CHD. We reported 30 unrelated newborns recruited from Neonatology service for genetic exploration.

Genetic investigations are essentially based on the techniques of cytogenetics and molecular cytogenetics. At first intension banded karyotyping was performed followed by fluorescent in situ hybridization (FISH) using gene-specific probe TUPLE1 in 22q11.2. As a last resort comparative genomic hybridization CGH-array 44K (Agilent® Technology) has been performed for 4 patients.

FISH showed normal hybridization to the DiGeorge syndrome critical region for all patients and no copy number variations was detected by array-CGH.

Our analysis was limited by a small and heterogenous study population. Also Increasing resolution arrays are needed to detect cryptic rearrangements.

We propose this strategy to explore a wider group of patients to identify new genetic factors involved in the development of cardiac malformations. The identification of genetic etiologies for CHD is important to provide genetic counseling and to establish a report genotype phenotype for every type of heart disorder.

Complex but balanced chromosomal rearrangements can give rise, through recombination during meiosis, to complex unbalanced rearrangements. Here, we report on the case of a 21 months old child associating a 8q21.13 duplication and 8p23.3 microdeletion. The proposal was referred to our lab for cytogenetic exploration of a hydrocephalus associated with facial dysmorphism. He had also psychomotor retardation and microcephaly. The patient R-banding karyotype revealed a partial trisomy 8q captured by the p telomere of the same chromosome, whereas the parents' karyotypes were normal. CGH-array technique characterized breakpoints and estimated its size to 61.8 Mb. Interestingly, an additional cryptic loss of 15.3Mb associated with partial trisomy 8q24.12q24.23 of 22.7Mb region. These results were confirmed by FISH using telomeric 18q probe.

Choringal atresia and skeletal malformation are in agreement with the monosomy 18q. Interestingly, the deletion includes GALR1 gene in 18q23 witch encodes galanine receptor. Galanine is a neuromodulator that stimulates growth hormone secretion. MBP, and adjacent genes, are implicated in myelination process and haptinsufficiency explains partially developmental delay. Otherwise, the haptinsufficiency of the 18q22.3-q23 gene region is suggested to be a critical region for the immunoglobuline A deficiency which is significantly associated to celiac disease. Our patient has not until now immunological disorders.

The association deletion 18q22- GH deficiency and decreased myelination is now well established. The real therapeutic impact of GH treatment is discussed.

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