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 intensity 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.

**MOLECULAR CHARACTERIZATION OF DER (8) (QTERQ21.13: PTERQ23.3) DN IN A CHILD ASSOCIATING PSYCHOMOTOR RETARDATION, HYDROCEPHALUS AND FACIAL DYSMORPHISM**

**DUPPLICATION OF THE SOX3 GENE IN A SRY NEGATIVE 46, XX MALE**