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 proposita was referred to our lab for cytogenetic exploration of a neurodevelopmental delay, microcephaly and facial abnormalities. He had also psychomotor retardation and microcephaly. The patient R-banding karyotype revealed a partial trisomy 8q captured by the p telomere of chromosome 8 in our patient. The clinical picture including psychomotor retardation and microcephaly. The patient R-banding karyotyping was performed followed by fluorescent in situ hybridization (FISH) using gene-specific probe TUPLE1 in 8q21.12. 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.

Abstracts

738 MOLECULAR CHARACTERIZATION OF DER (8) (QTERQ21.13:PTERP23.3) DN IN A CHILD ASSOCIATING PSYCHOMOTOR RETARDATION, HYDROCEPHALUS AND FACIAL DYSMORPHISM

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740 DUPLICATION OF THE SOX3 GENE IN A SRY NEGATIVE 46, XX MALE

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Case presentation An 11 old patient with hypoplasia of the right kidney and hypospadias was found to be SRY negative, 46, XX. His parents and younger sister were healthy. His intelligence was normal (IQ 92) and he had no other anomalies. The behavior, growth and development were all normal. His testes were >4ml and the penis was 5 cm. Ultrasound and MRI did not show internal female genitalis, while confirming right kidney hypoplasia (as did the DMSA scan).

ACTH test showed normal basal and stimulated 17OH-progesterone excluding a form of 46XX DSD due to 21-hydroxylase deficiency. 11-DOC and 11S were normal at both baseline and after ACTH stimulation, excluding 11-hydroxylase deficiency. Cortisol levels were in the mid normal range at baseline and responded to stimulation, excluding primary adrenal insufficiency. Androstenedione.

The hCG test found testosterone in the low normal range for male sex at age at baseline. It rised up to 146 ng/mL indicating the presence of functional Leydig cells targeted by hCG. The stimulated ratio TDHT was 5.6, not supporting 5 alpha-reductase deficiency.