IDENTIFICATION OF A MOSAIC NON-INHERITED SMALL SUPERNUMERARY RING CHROMOSOME 2: CYTOGENETIC-MOLECULAR STUDIES AND GENOTYPE-PHENOTYPE CORRELATION– Poster.

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Introduction: The identification of supernumerary marker chromosomes (SMCs) derived from all the autosomes is currently possible, but rarely by conventional cytogenetics alone. Supernumerary ring chromosomes (SRCs) account for about 10% of these cases. SRCs derived from chromosome 2 are unusual, and there are only a few cases reported in the literature. The severity of the phenotype depends on the type of the mosaicism, the percentage of cells affected by the genetic change and the chromosome involved.

Methods: The authors report the case of a boy aged 8 referred for cytogenetic studies, presenting with behavior and learning problems, mental retardation with uncoordenated speech, attention deficit and hyperactivity (PHDA), as well as small slanting palpebral fissures.

The karyotype was obtained from peripheral blood lymphocyte cultures using high resolution GTL banding and standard techniques. Fluorescence *in situ* hybridization (FISH) was performed using specific probes for the centromeric regions of all chromosomes (Chromoprobe Multiprobe - ISystem).

Results: Cytogenetic analysis revealed two cell lines: one with a supernumerary marker ring chromosome, 47,XY,+r (52%), and a normal cell line, 46,XY (48%). The SRC was identified by FISH with the chromosome 2 centromeric probe. Since the parents had normal karyotypes, this abnormality was “de novo”. Final karyotype of the proband was: mos 47,XY,+r[26]/46,XY[24].ish r(2)(D2Z2+)dn.

Discussion: The clinical description of this patient is in agreement with other reports of the literature. Molecular characterization by FISH analyses is an useful way of investigating the presence of euchromatin contained in a SMC and establishing new chromosomal syndromes. However, to better characterize this ring, in order to establish a more accurate genotype-phenotype correlation, more studies involving other technologies should be performed, thus allowing suitable genetic counselling.