CLINICAL, CYTOGENETIC AND MOLECULAR FINDINGS OF A "DE NOVO" INV DUP DEL (6q)

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Introduction: Complex rearrangements resulting in inverted duplications contiguous to a terminal deletion (inv dup del) were first reported for the short arm of chromosome 8 in1976. Since then this type of structural anomaly has been described for an increasing number of chromosomes. In these rearrangements, the concomitant presence of a deletion and a duplication has important consequences in genotype-phenotype correlations. The authors describe the clinical findings and the cytogenetic characterization of a rare inv dup del involving the long arm of chromosome 6.

Material and methods: A girl aged 5 was referred for subtelomeric studies with the indication of psychomotor retardation, autistic features and stereotipies. Chromosome analysis with high resolution GTL-banding was performed on metaphases obtained from cultured peripheral blood lymphocytes. Molecular studies included MLPA (Kits P036 and P070, MRC-Holland), FISH with subtelomeric and whole chromosome painting probes specific for chromosome 6, and cCGH techniques.

Results: Initial MLPA studies detected a subtelomeric deletion in the long arm of chromosome 6; the subsequent karyotype revealed a structurally abnormal chromosome 6 with additional material in the end of the long arm. FISH analysis showed the deletion and demonstrated that the extra material was derived from chromosome 6; cCGH tecnhiques defined the extension and confirmed the breakpoints of the duplicated segment. Thus this rearrangement was interpreted as an inv dup del (6q). Since parental karyotypes were normal, this anomaly was considered "de novo".

Discussion: As far as we know this is the first description of a patient presenting with a "de novo" inv dup del (6q). We compare the clinical features in this child with the previously reported cases with either an isolated terminal deletion or a duplication of distal 6q. The authors enhance the importance of the combination of high resolution banding with molecular studies in the characterization of this rare rearrangement.