He was the first baby born to a non-consanguineous couple. The birth weight was 2,500 g. The examination findings were: small frontal and occipital asymmetry, upslanted palpebral fissures, hypertelorism, flat nasal bridge, board neck, bilateral clinodactyly, mild mental retardation and small penis (both testes were in well-formed scrotal sacs). No cardiovascular alterations were detected.

Chromosome culture and karyotyping were performed using standard techniques and showed a karyotype of 49,XXXXY.

Although initially 49,XXXXY pentasomy was considered a variant of Klinefelter syndrome, it is currently recognized as a separate clinical entity distinguished by facial features, multiple skeletal and cardiac defects, and short stature. A 49,XXXXY karyotype is thought to arise from maternal non-disjunction which occurs during both meiosis I and meiosis II. This produces a secondary oocyte with four X chromosomes, which, when fertilized by a Y chromosome-bearing sperm, results in an embryo with 49,XXXXY syndrome.

In this case, the facial dysmorphism and small penis were the main features which led to a suspicion of sex chromosome aneuploidy that was confirmed by chromosomal analysis.

The prognosis of these children depends on the extent of severity of the condition, while the management mandates a multidisciplinary approach with pediatric endocrinology, pediatric surgery, orthopedics, psychiatry and clinical genetics evaluation.

**Keywords**: Small penis, Sex chromosome aneuploidy, Facial dysmorphism, XXXXY syndrome

**1.P30**

Large interstitial del(13)(q13q14.3): the importance of detailed clinical information in cytogenetic studies

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Interstitial deletions of chromosome 13 are known to be associated with retinoblastoma. A wider syndrome may accompany the deletion, including mental retardation and craniofacial dysmorphism. The severity of the phenotype depends on the extent of the deletion. Retinoblastoma is a malignant tumor in the retina and is the most common ocular cancer in children. The association of most cases of retinoblastoma with an interstitial del(13q) has led to the localization of the retinoblastoma gene in 13q14.

We report a case of a boy aged 8 referred for cytogenetic studies, presenting with mild mental retardation, craniofacial dysmorphia, delayed intrauterine growth (IUGR) and retinoblastoma. The karyotype was obtained from peripheral blood lymphocyte cultures using high-resolution GTG banding and standard techniques. Fluorescence in situ hybridization was performed using the LSI 13 (RB1) probe (Vysis) for region 13q14 spanning the RB1 gene.

The chromosomal analysis revealed a large interstitial deletion of the long arm of chromosome 13.

Although the exact breakpoints were difficult to establish, the deleted region did not appear to encompass the band which includes the retinoblastoma gene. Molecular cytogenetic techniques showed that the retinoblastoma gene was deleted. This confirmed the clinical indication of retinoblastoma and defined the deletion breakpoints more precisely.


Except for the presence of IUGR, the clinical description of this patient is in agreement with other reports in the literature. We would like to emphasize the importance of detailed clinical information that, together with classical and molecular cytogenetic techniques, could be useful in better defining the breakpoints, establishing correct genotype/phenotype correlation and thus providing appropriate genetic counselling. The blood samples of the parents were requested for karyotype analysis in order to clarify this chromosome deletion.

**Keywords**: del(13q), Interstitial deletion, Retinoblastoma

**1.P31**

Variable phenotypes in a group of 6 patients with distal deletions of chromosome 9p
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Keywords: Deletion of 9p, Monosomy 9p syndrome, Mental retardation, Gonadal dysgenesis

I.P32

A rare inherited case of 4q deletion detected by GTG and array analyses in a newborn

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Keywords: 4q deletion syndrome, Array analyses