

## **Interstitial deletion 15q21 and *Prader-Willi like syndrome* phenotype: Case report.**

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**Introduction:** Chromosome 15q interstitial deletions not involving the Prader-Willi/Angelman region are uncommon and poorly characterized. Very few cases of different segmental losses involving the 15q21 region have been reported at cytogenetic level. All the described patients present with moderate to severe mental retardation and characteristic facial dysmorphic features. Some authors compare the similarity between the phenotype of these patients with some features of Prader-Willi syndrome (PWS).

**Methods:** We report the case of a girl aged 8 referred for conventional cytogenetics and fluorescence *in situ* hybridization (FISH) for the PWS region, presenting with mental retardation, almond-shaped eyes, obesity, small hands with short fingers and diminished pigmentation of the hair.

**Results:** The chromosomal analysis revealed an interstitial deletion of the long arm of chromosome 15, apparently between 15q21 and 15q22. Deletion at 15q11.2 (Prader-Willi/Angelman critical region) was excluded by FISH.

To establish the exact breakpoints molecular studies were performed using bacterial artificial chromosome (BAC) clones spanning the 15q21.3 region. The absence of signal in this region defines the proband's final karyotype as:

46,XX,del(15)(q21.3q21.3).ish del(15)(q21.3q21.3)(bA74K1-)

**Discussion:** The authors emphasize the importance of complementary FISH and molecular studies in chromosomal abnormalities and compare the proband's phenotype with similar cases described in the literature.