

## Morphological examination of a preterm neonate with atypical Pallister-Killian syndrome

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Pallister-Killian syndrome (PKS) is a rare genetic disorder which is cytogenetically characterized by a tissue limited mosaic distribution of isochromosome 12p (tetrasomy 12p). This chromosomal anomaly may be detected in fibroblasts and in bone marrow cells, in amniocytes, chorionic villus and other foetal tissues, whereas it is absent in peripheral lymphocytes. The percentage of abnormal cells in fibroblasts decreases with the ageing, so adult patients may be undiagnosed. We report on a pre-term male neonate of 1320 gr of weight, born at a gestational age of 29-30 weeks. He had severe respiratory distress and died after a few hours, in spite of intensive resuscitation. The external examination showed a disproportionate large face and low-set ears, limbs of normal length, with normal hands and feet, normal genitalia. At the autopsic examination all the thoracic, abdominal and pelvic organs were normal, the palate showed a schisis in the posterior side of the arch; the external appearance of the brain was normal, with hyperaemic pial membrane.

The cytogenetic analysis on skin fibroblasts showed the presence of a supernumerary metacentric chromosome, consistent with an isochromosome 12p. Subsequent interphasic FISH with a centromeric probe detected tetrasomy 12p in mosaic. The postnatal phenotype of PKS is quite severe and include coarse facies with a high forehead, sparse scalp hair, hypertelorism, broad nasal bridge, hypotonia, streaks of hypo-hyperpigmentation. Hydramnios, congenital diaphragmatic hernia and micromelia, of predominantly rhizomelic type are frequent echographic signs during the pregnancy. Hydrops fetalis, hygroma colli, increased nuchal translucency (INT), foetal overgrowth and ventriculomegaly are less frequently reported.

In the present case no foetal malformations were evident during the pregnancy, and the facial dysmorphisms and congenital malformations described in PKS were absent at the morphological and autopsic examination of the newborn. This suggests that PKS could be suspected, and then verified on skin fibroblasts, even in absence of significant congenital malformations, mainly in dysmorphic patients cytogenetically normal at standard cytogenetics on peripheral lymphocytes.

Keywords: Pallister-Killian syndrome