A case of Pallister-Killian Syndrome in a newborn

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Objective. Pallister Killian syndrome (PKS), a rare disorder caused by tissue-limited mosaicism tetrasomy of chromosome 12p, has a typical dysmorphic pattern: macrosomia, coarse facies, hypertelorism, small nose with long philtrum, V-shaped upper lip, low set ears, frontotemporal alopecia and pigmentary skin anomalies. Seizures and developmental delay, cardiac defects, diaphragmatic hernia, renal/anal malformations may be associated.

Materials and Methods. We report the case of a newborn with multiple congenital malformations, later diagnosed with PKS.

Results. A female baby was born from vaginal delivery at 41 weeks of gestational age. Pregnancy was unremarkable. Apgar score was 8 at 1 minute and 9 at 5 minutes. Weight at birth: 4030 g (95th percentile). On neonatal examination, hypertelorism, ogival palate, a white hair wisp, ulnar fingers deviation and anteriorly displaced anus with perineal fistula were observed. A few hours after birth, she showed severe respiratory distress, with development of persistent pulmonary hypertension, requiring intubation and ventilatory support with conventional and non-conventional systems. Antibiotics therapy was started for neonatal sepsis. A gradual improvement in the respiratory function and sepsis resolution allowed ventilator support to be interrupted. Brain MRI and encephalic ultrasound were normal. Abdominal ultrasound and MRI documented the presence of a cystic lymphangioma and multiple bilateral ovarian cysts. Unilateral hydronephrosis was also diagnosed. Genetic karyotype allowed the diagnosis of PKS: mos47,XX,i(12)(p10)[1]/46,XX[99].

Conclusions. Phenotypic and cytogenetic variability of PKS, with lack of correlation between tetrasomic cells proportion and disease severity, may be challenging for diagnosis. A detailed physical examination is mandatory for early suspicion and diagnosis.