A case of Pallister-Killian Syndrome in a newborn

Giulia **Di Donato** [1,2,*], Valentina **Chiavaroli** [1,3], Marianna **Sebastiani** [1], Paola **Cicioni** [1], Laura **Sabatini** [1], Altea **Petrucci** [1], Silvia **Carinci** [1], Noemi **Pellegrino** [1], Eliana **Valzano** [1], Marianna **Del Torto** [1], Rita **Cognigni** [1], Simona **Di Credico** [1], Susanna **Di Valerio** [1]

[1] Neonatal Intensive Care Unit, Pescara Public Hospital, Pescara, Italy.
[2] Department of Pediatrics, G. d'Annunzio University of Chieti-Pescara, Chieti-Pescara, Italy.
[3] Liggins Institute, The University of Auckland, Auckland, New Zealand.

DOI: 10.36129/jog.2022.S128

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.