Minor dysmorphic features in a newborn: an unexpected diagnosis

Jessica **Ruggiero** [1,*], Stefania **Sirianni** [2], Maria Rosa **Cutrì** [3], Ilaria **Bosio** [2], Vania **Spinoni** [2], Cristiana **Corrado** [4], Raffaele **Badolato** [1,3], Francesco Maria **Risso** [2], Carmen **Rodriguez Perez** [2]

[1] Department of Clinical and Experimental Sciences, University of Brescia, Brescia, Italy.
[2] Neonatal Intensive Care Unit, Children's Hospital, ASST Spedali Civili di Brescia, Brescia, Italy.
[3] Department of Pediatrics, Children's Hospital, ASST Spedali Civili di Brescia, Brescia, Italy.
[4] Department of Medicine, Surgery and Health Sciences, University of Trieste, Trieste, Italy.

DOI: 10.36129/jog.2022.S138

Objective. We present a clinical case of a female newborn, caucasian, of nonconsanguineous parents. The pregnancy was characterized by the findings of IUGR and bilateral renal pyelectasis at 36 weeks of gestation. Delivery was spontaneous at term. The birth weight was 2440 g (< 3rd centile), length 46 cm (4th centile), head circumference 32 cm (6th centile). The clinical evaluation revealed minor dysmorphisms, such as wide anterior and posterior fontanelles, down slanting palpebral fissures, high nasal root, bulbous nasal tip, large mouth, wide auricles, left foot talus valgus and right foot varus supinated, clinodactyly in the fourth and fifth toes.
Materials and Methods. We analysed the clinical data of the patient and researched similar cases in literature. Diagnostic investigations included medical visits, ultrasounds and genetic testing.
Results. The echocardiography showed septal atrial defect and septal ipo-dyskinesias, whereas transfontanellar and abdomen ultrasounds were normal. Array- CGH revealed the presence of a pathogenetic duplication in 9p24.3p21.2.
Conclusions. Although rare, trisomy of the short arm of chromosome 9 is one of the most common autosomal structural anomalies in newborns, also known as Rethorè syndrome. To date, different cases have been reported in literature, characterized by similar craniofacial dysmorphisms, limb anomalies, delayed mental and psychomotor development. The phenotype may be variable, depending on the size of the chromosomal abnormalities and the different genes involved, such as DOCK8 (important in immunology) and KINK1 (in oncology). Genetic studies should be extended to the child's parents to identify the source of any balanced translocation and the risk of recurrence.