Annals of Clinical Case Reports

Case Study
Published: 16 Aug, 2019

Persistent Thrombocytopenia: Think about Jacobsen Syndrome

Salem Yahyaoui, Omar Yahyaoui, Dorsaf Saadouli, Sonia Mazigh, Olfa Bouyahya and Samir Boukthir*

Department of Pediatrics, Tunis El Manar University, Tunisia

Abstract

Jacobsen Syndrome is a rare genetic disorder caused by a deletion of the distal end of chromosome 11. It associates multiple congenital abnormalities including dimorphisms and mental retardation. It is secondary to a partial deletion in the long arm of chromosome 11. We report a case of Jacobsen syndrome revealed by persistent thrombocytopenia and we propose to illustrate characteristic images to contribute to the study of this condition.

Introduction

Jacobsen Syndrome is a rare genetic disorder that associates multiple congenital anomalies with psychomotor retardation. It is caused by a partial deletion of long arm of chromosome 11 [1]. We report a case of Jacobsen syndrome diagnosed in the neonatal period.

Case Study

A male 25 days aged newborn was referred for persistent thrombocytopenia. He was born at 37 completed weeks of pregnancy. The mother was immunized against toxoplasmosis and rubella. He was stunted with a birth weight of 2300 g. He had facial dysmorphism associating trigoncephaly, nasal saddle, and short nose (Figure 1). In addition, the examination showed arthrogryposis (Figure 2). Hands-X-rays showed agenesis of the third phalanx of the left middle finger (Figure 3). He had no other associated malformations. The cells blood count showed thrombocytopenia at 38000/ mm^3 which was persistent. Due to the persistence of hematological abnormalities, a myelogram was performed showing dysgranulopoiesis, and dyserythropoiesis. The blood karyotype revealed a partial deletion of long arm of chromosome 11 [del(11)(q23)] corresponding to Jacobsen syndrome. The newborn had hemorrhages every two to three months requiring platelet transfusions.

Discussion

Jacobsen syndrome is characterized by multiple congenital anomalies and intellectual retardation. Its prevalence is estimated at 1 per 100000 births with a gender ration of 2 girls/1 boy [1]. This affection results from the partial deletion of the distal end of chromosome 11. It associates stunting, mental retardation, facial dysmorphism (trigoncephaly, hypertelorism, epicanthus, short noses, retrognatism, low ear implantation, nasal tracking, palatal and dental anomalies), cardiac malformations are present in half of the cases, as well as abnormalities of the extremities [1,2]. Ocular, digestive and renal malformations are described as well as endocrine and immunological

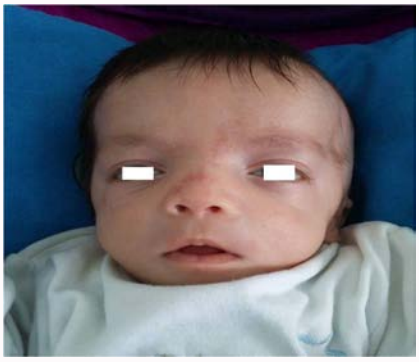

Figure 1: Facial dysmorphism associating trigoncephaly, nasal saddle, short nose.

OPEN ACCESS

Correspondence:
Salem Yahyaoui, Department of Pediatrics, Children Hospital of Tunis, Tunis El Manar University, Bab Saadoun, Tunisia,
E-mail: yahyaouisalem@yahoo.fr

Received Date: *22 Jul 2019*
Accepted Date: *12 Aug 2019*
Published Date: *16 Aug 2019*

Citation:
Yahyaoui S, Yahyaoui O, Saadouli D, Mazigh S, Bouyahya O, Boukthir S. Persistent Thrombocytopenia: Think about Jacobsen Syndrome. Ann Clin Case Rep. 2019; 4: 1702.

ISSN: 2474-1655

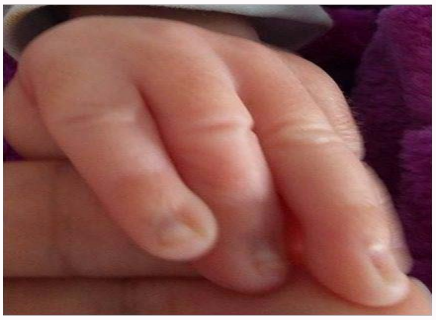

Figure 2: The examination showed arthrogryposis.

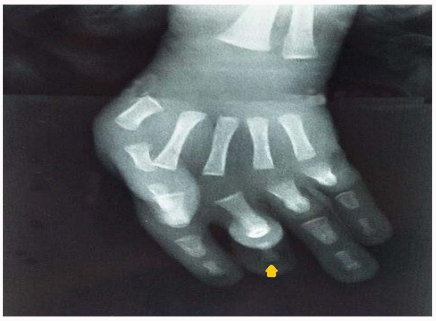

Figure 3: Hands-X-rays showed agenesis of the third phalanx of the left middle finger.

abnormalities [2]. Thrombocytopenia is the main haematological abnormality. The prognosis of these patients is conditioned by cardiac malformations. Cytopenias are often spontaneously regressive, but thrombocytopenia may persist as the presented case.

Conclusion

We reported a particular observation as Jacobsen syndrome was revealed by an early and persistent thrombocytopenia. Dimorphism was unrecognized by the primary care physician. Thus it is important to recall the clinical presentation and the characteristic dimorphism to guarantee early diagnosis and management.

Authors Contribution

Salem Yahyaoui and Dorsaf Saadouli wrote the paper. All authors provided care and follow-up for the patients

Samir Boukthir supervised the work. Guarantor: Salem Yahyaoui has full responsibility for the work.

References

1. Favier R, Akshoomoff N, Mattson S, Grossfeld P. Jacobsen syndrome: Advances in our knowledge of phenotype and genotype. Am J Med Genet C Semin Med Genet. 2015;169(3):239-50.

2. Takenouchi T, Kosaki K. Jacobsen syndrome, Braddock-Carey syndrome, and Beyond: Reflections on intellectual disability accompanied with thrombocytopenia. Am J Med Genet A. 2016;170(10):2578-9.