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Beyond the Classic TAR Phenotype: Unilateral Absent Radius with Tracheoesophageal Fistula

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Abstract

Background: Thrombocytopenia with absent radius (TAR) syndrome is a rare congenital disorder characterized by thrombocytopenia and bilateral radial aplasia with preserved thumbs. We report an unusual case of a 2-day-old term male neonate presenting with respiratory distress, excessive frothing, unilateral absence of the right radius, and thrombocytopenia (30,000/mm^3). Chest radiography confirmed Type C tracheoesophageal fistula (TEF). Following platelet transfusion, the infant underwent successful fistula ligation and primary esophageal anastomosis. Postoperative management included serial platelet monitoring and transfusions as required, with an uneventful recovery. The infant was discharged with orthopedic follow-up for limb reconstruction. This rare association of unilateral radial aplasia with TEF expands the phenotypic spectrum of TAR syndrome and underscores the importance of early recognition and meticulous perioperative platelet management to optimize surgical outcomes.

Keywords: Thrombocytopenia with absent radius syndrome; TAR syndrome; Tracheoesophageal fistula; unilateral absent radius; thrombocytopenia; neonate.

INTRODUCTION

Thrombocytopenia-Absent Radius (TAR) syndrome is a rare congenital disordercharacterized by thrombocytopenia due to hypomegakaryocytic thrombocytopenia and radial ray defects, classically involving bilateral absence of the radii with preservation ofboth thumbs [1]. The syndrome has an estimated incidence of approximately 0.42 per 100,000 live births and is associated with variable hematological manifestations and congenital anomalies affecting the cardiovascular, renal, and gastrointestinal systems[2].

While bilateral radial aplasia is considered the hallmark skeletal abnormality, atypical phenotypes, including unilateral radial absence, have been infrequently reported, broadening the clinical spectrum of the disorder. Tracheoesophageal fistula (TEF), commonly associated with esophageal atresia (EA), is a congenital foregut anomaly requiring early surgical correction[3]. The coexistence of TAR syndrome and TEF is exceedingly uncommon, and reports describing this association are scarce. The presence of thrombocytopenia, upper limb skeletal anomalies,and neonatal airway pathology significant perioperative challenges, including difficult vascular access, optimization of platelet counts, prevention of perioperative hemorrhage, airway management, and meticulous anesthetic planning.

We report the successful perioperative anesthetic management of a neonate with an unusual presentation of TAR syndrome characterized by unilateral absence of the radius associated with tracheoesophageal fistula. This case highlights the importance of recognizing atypical phenotypic variants of TAR syndrome and underscores the need for multidisciplinary approach and individualized anesthetic management to achieve favorable surgical outcomes. To the best of our knowledge, the coexistence of TAR syndrome with unilateral radial aplasia and TEF has been reported only once previously in the literature, making this an exceptionally rare clinical presentation.

CASE PRESENTATION

A two-day-old term male neonate was born at our institution by normal spontaneous vaginal delivery. He had swallowing problem and increased oral secretion. The antenatal period was uneventful, with no history of maternal illness, fever, rash, drug exposure, or other significant complications during pregnancy. There was no family history of congenital anomalies or inherited disorders. On admission, the neonate was hemodynamically stable, alert, pink, and active. Anthropometric measurements were appropriate for gestational age, with a birth weight of 2.7 kg, a length of 50 cm and a head circumference of 34 cm, all corresponding to approximately the 50th percentile. There were no hematological signs like petechiae, ecchymosis, hepatomegaly, and splenomegaly. He had right absent radii and radial deviation of right upper limb with no thumb deformity (Figure 1). There were no deformities in the lower limbs. There was no clinical evidence of congenital heart disease and the other systems were normal. Initial laboratory investigations gave the following results: complete blood count: hemoglobin, 10g/dL; mean corpuscular volume, 105 fl; mean corpuscular hemoglobin, 34 pg; reticulocytic count, 6%; TLC: 49,000/uL, 60% s. neutrophils, 5% myelocytes, 5% metamyelocyte, 10% normoblasts; platelet count, 30,000/uL; prothrombin time, 12.8 seconds; partial thromboplastin time, 28.0 seconds (cont 28). Liver function tests had the following results: bilirubin (total), 6.14 mg/dL; (direct), 0.24 mg/dL; glutamic pyruvic transaminase, 28 U/L; and glutamic oxaloacetic transaminase, 34 U/L. Electrolytes and kidney function were: Na,136 mmol/L; K, 4.8 mmol/L; serum creatinine, 0.9 mg/dL.

Radiography of the forearm showed right arm absence of radii with normal thumbs and radial deviation of the hands. Diagnosis of EA with TEF was made by barium-contrast study of the upper pouch of the esophagus (Figure 2). Based on these physical findings, thrombocytopenia and X-ray results the diagnosis of TAR Syndrome with single absent radius and trachesoesophageal fistula was made. The differential diagnosis of TAR includes mainly three congenital anomalies: Patients with Fanconi' anemia,Thalidomide embryopathy causes phocomelia and Holt-Oram syndrome but differ from TAR in various aspects.

Figure 1: Baby with TAR syndrome showing shortened forearms with radial deviation of the hands.
Figure 2 : Chest X-Ray of the patient showing nasogastric tube in the esophagus and coiling of the nasogastric tube within the esophagus.

DISCUSSION

TAR syndrome is a rare autosomal recessive disorder characterized by thrombocytopenia and radial ray defects with preservation of the thumbs[4]. In addition to hematological and skeletal abnormalities, patients may have associated cardiac, renal and gastrointestinal anomalies. Differential diagnoses include Fanconi anemia, Holt-Oram syndrome and trisomy 18; however, the presence of preserved thumbs, neonatal thrombocytopenia and the absence of pancytopenia help distinguish TAR syndrome from these conditions. The characteristic hematological finding is hypomegakaryocytic thrombocytopenia, which is most severe during the first year of life and predisposes affected infants to life-threatening hemorrhage and increased susceptibility to infections. Platelet counts generally improve with age.

Esophageal atresia with tracheoesophageal fistula (TEF) is a common congenital foregut anomaly frequently associated with other congenital malformations, including those seen in the VACTERL spectrum[5]. Esophageal atresia (EA) occurs in approximately 1 in 3000 to 4500 births globally. However, its association with TAR syndrome is exceptionally rare, making the coexistence of these conditions an unusual clinical entity requiring careful multidisciplinary perioperative management. Esophageal abnormalities have been described in only one case with TAR syndrome till date. This case describes a rare association between TAR syndrome and esophageal atresia. While TAR syndrome was unlikely to have directly affected the patient's prognosis, the associated hematological and immune abnormalities may have increased susceptibility to sepsis, thereby influencing the clinical course.

CONCLUSION

TAR syndrome is a rare genetic disorder classically characterized by thrombocytopenia and bilateral radial aplasia with preserved thumbs. We describe a male neonate presenting with thrombocytopenia, bleeding manifestations, unilateral radial dysplasia, and a rudimentary thumb—features that deviate from the typical phenotype of TAR syndrome.

While the clinical findings are most consistent with an atypical variant of TAR syndrome, the unusual combination of anomalies raises the possibility that this presentation may represent a distinct phenotypic entity rather than a true variant of TAR syndrome. Further reports and molecular genetic studies are needed to clarify its nosological classification.
These variants of TAR syndrome need to be diagnosed as serial monitoring of platelet count is essential in the management of patients.

CONSENT

Written informed consent was obtained from the patient's next-of-kin for publication of

this case report and any accompanying images.

Acknowledgement

We thank the parents of our case for providing written informed consent and permission to publish the photos and x-ray of their baby.

Financial or Other Competing Interests

None.

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