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Variant of Thrombocytopenia with Absent Radius Syndrome: Case Report

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Abstract

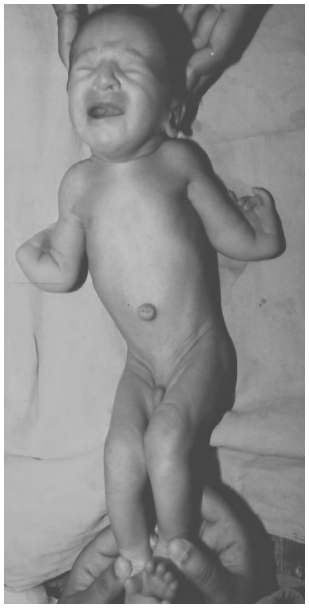
Thrombocytopenia with absent radius (TAR) is a congenital syndrome characterized by severe thrombocytopenia with bilateral absent radii and may be associated with other skeletal abnormalities. We report a case who had bilateral micromelia of upper limb, radially curved forearms and bilateral absent of thumb at birth. A severe thrombocytopenia confirmed the clinical diagnosis of thrombocytopenia with absent radius syndrome in the neonate.

Keywords: *Thrombocytopenia; Radius; Newborn.*

Introduction

Thrombocytopenia with absent radius syndrome is an autosomal disorder characterized by the neonatal onset thrombocytopenia, bilateral absence or hypoplasia of the radii with normal or poorly formed hands and other variable skeletal malformation.[1,2] It is transmitted in an autosomal recessive fashion and consanguinity is not a feature.[3] This syndrome was first noted by Greenwald and Sherman in 1929.[4] Other skeletal abnormalities like abnormal or absent humerus, dislocated hips, tibial torsion, ankylosis of knee and hypoplasia or aplasia of femur are usually present.[4,5] Here we report a case of severe thrombocytopenia with bilateral absent radius.

Figure 1: Newborn with Micromelia of Upper Limbs, Radial Curving of Forearm with Poorly Formed Hands



Case Report

The term male baby weighed 1630gm was born through normal vaginal delivery, first lived baby of second pregnancy of healthy mother and father whose ages were 22years and 26years respectively and are non-consanguineous. Mother did not have an important illness when she was pregnant and did not use any medication except multivitamins. First pregnancy of the mother

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Figure 2: X-ray Hand Show Bilateral Radial Aplasia with Radial Curving of Ulna and Absent 5th Metacarpal Bone

resulted in spontaneous abortus in 4th month because of blighted ovum. On examination, neonate had bilateral micromelia of upper limb, radially curved forearms and bilateral absent of thumb were present (Figure 1). Petechiae and ecchymotic patches were absent. Other systemic examination was normal. On hematological investigation, hemoglobin was16.8gm/dl, TLC: 12,000/cumm, DLC: 65% neutrophil, 32% lymphocyte and 3% eosinophil. Platelet count was 68,000/cumm. X-ray hand revealed bilateral radial aplasia, radial curving of ulna and absent bilateral 5th metacarpal bone (Figure 2). Echocardiography and Doppler examination was normal. Baby was discharged after 10 days.

Discussion

TAR is a congenital syndrome characterized by severe thrombocytopenia with bilateral absent radii and may be associated with other skeletal abnormalities. The most pronounced skeletal abnormality of this rare syndrome is bilateral radial aplasia. The humerus is affected in 50% of patients and may be shortened or reduced to small interconnecting bone positioned between the scapula and hand. Limbs are more involved than the trunk. Hypoplasia, aplasia or malformation of ulna seen in 78%, hand 75% and humerus 40% of cases. However, thumbs and digits are almost always present which distinguishes TAR from fanconi's anemia.[3,6] Bleeding manifestations and severe thrombocytopenia (platelet count<10-30,000/cumm) have been reported in more than 50% of cases at birth or before the age of 1 week and 90% of patient by 4 months of age.[3]

The baby had typical features of the TAR syndrome: bilateral aplasia of the radii, radially deviated hands and thrombocytopenia. But the unusual finding was bilateral absence of thumb. This malformation can be delimited from other syndromes with aplasia of the radii such as fanconi's pancytopenia syndrome, Holt-Oram syndrome, fetal thalidomide syndrome, trisomy 18 syndromes and Robert's SC-phocomelia syndrome. In fanconi's pancytopenia, facial dysmorphism and radius anomalies can be observed in 30% patients. Diepoxybutane (DEB) test is positive in most of patients.[7]. Whereas thrombocytopenia is rare in neonatal period and in later periods aplastic anemia is observed.[1,2] Robert's SC-phocomelia syndrome, an autosomal recessive syndrome of tetraphocomelia, cleft lip, cleft palate and mental retardation may be difficult to differentiate clinically from the TAR syndrome.[8] In our case, aplastic anemia, cleft palate and cleft lip was absent and DEB test couldn't be done. TAR syndrome can be diagnosed prenatally by ultrasonography and chordosynthesis.[9,10] Weinblat *et al* applied in utero thrombocyte transfusion on TAR syndrome case diagnosed prenatally.[10] Unfortunately, prenatal diagnosis could not be made in our case. The baby without severe hemorrhages, show a fairly good tolerance to thrombocytopenia after the first year of life, and a good response to steroid therapy when necessary. Prognosis depends upon the severity and duration of thrombocytopenia with overall mortality of 40%.

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