

Thrombocytopenia and Absent Radius (TAR Syndrome): A Rare Case Presenting in a Nine Day Old Neonate

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Abstract

Thrombocytopenia absent radius syndrome (TAR) is a rare genetic disorder that is apparent at birth. TAR syndrome is inherited as an autosomal recessive trait. This disorder is characterized by low levels of platelets in blood (thrombocytopenia) resulting in potential severe bleeding episodes primarily during infancy. Other characteristic findings include absence of the bone on the thumb side of the forearm and underdevelopment or absence of the bone on the radial side of the forearm. Other abnormalities may also be present such as structural malfunctions of the heart, kidneys and mental retardation that may be secondary to bleeding episodes in the brain during infancy.

Introduction

TAR SYNDROME is a rare genetic disorder. A case of TAR syndrome is hereby presented because of its rarity.

Case Report

AT is a nine-day-old female child who presented at the orthopaedic clinic with history of abnormality of both upper limbs. She was a product of a full term pregnancy. The mother was immunized against

tetanus during pregnancy and attended antenatal clinics regularly. Delivery was spontaneous vertex and the baby cried immediately after delivery, Apgar score was 10 at 5 minutes. There was no history of birth trauma and no family history of similar illness. On musculoskeletal examination, both forearms were shortened with associated bowing and fixed flexion at the elbow joints. No other abnormality was detected on the musculoskeletal systems. An impression of congenital absence of the forearm was made. Plain radiograph of the right upper limb showed absence of the radius with bowing of the ulna, and hyperextension of the wrist joint as well as flexion deformity of the elbow joint (Fig.1)

Fig.1



The left upper limb showed flexion deformity at the elbow joint otherwise the demonstrated radius and ulna were within normal limits. Radiographs of the lower limbs were also normal. Abdominal ultrasound and echocardiography showed no abnormality. The platelet count was however reduced, 50,000/mm³. A clinical diagnosis of TAR syndrome was made. The patient was admitted and transfused with platelet concentrates after which haemodynamic stability was achieved. She was subsequently referred for physiotherapy to relief the fixed flexion deformity.

Discussion

Thrombocytopenia absent radius (TAR) syndrome is a rare association of thrombocytopenia and bilateral radial aplasia first described in 1951 with some families having more than one members affected. An autosomal recessive inheritance pattern was proposed^{1,2,3}.

TAR was defined as a syndrome in 1969 and further classified as the association of hypomegakaryocyte, thrombocytopenia and gastrointestinal, haematological and cardiac systems abnormalities³. In the United States, this syndrome rarely occurs, so also in Nigeria but no definite figures are obtainable. Internationally, the frequency is 0.42 cases per 100,000 live births in Spain. No ethnic or racial predilections exist. The male to female ratio is 1:1.

TAR is congenital and the patient usually presents in the first week of life with symptomatic thrombocytopenia. This however wasn't the presentation in the present case.

Fifty percent of affected infants are symptomatic in the first week of life and 90% are symptomatic by age 4 months⁴.

Thrombocytopenic episodes are precipitated by non specific stress, infection and diet.

The upper limb abnormalities range from isolated absent

radii as was demonstrated in this patient, to phocomelia³.

Other abnormalities include bilateral radial aplasia. This present patient had unilateral radial aplasia; radial club hand, hypoplastic carpals and phalanges, hypoplastic ulnae, humeri and shoulder spindles, syndactyly, clinodactyly of fingers and toes, selective hypoplasia of middle phalanx, fifth digit and altered palmar contours.

Lower extremity anomalies occur in 46% of patients and may vary from being clinically undetectable to phocomelia which are usually less severe than those of the upper limb. Other abnormalities include hip dislocation, femoral torsion, tibial torsion, valgus and varus deformity and deformity of the knee. None of these was however seen in our patient.

Cardiac anomalies occur in 33% of patient⁴ though the cardiovascular examination and echocardiography of our patient were normal.

Facial anomalies which include micrognathia (3-30%), facial hemangioma, hypertelorism have also been reported in patients with TAR syndrome³.

Other numerous anomalies described in TAR include: asymmetric first rib, cervical rib, spinal bifida, fused cervical spine, nuchal folds, meckel's diverticulum, uterine anomalies, dorsal pedal edema, short stature, skeletal malformation and large horseshoe shaped kidneys.

In distinguishing TAR from other syndromes involving skeletal abnormalities of the upper extremities, the following may be of assistance. Thumb is always present in TAR but may be hypoplastic or absent in Fanconi anemia. Fanconi anemia also has chromosomal abnormalities, and onset of thrombocytopenia before age one year is rare though usually, pancytopenia is seen in children aged 5-10 yrs. Thumbs are also typically present in Holt-Oram syndrome, and blood cell counts are normal⁴. Characteristic skeletal involvements (i.e. absent radii) are detectable by prenatal ultrasonography as early as 16 weeks gestation, when sufficient fetal skeletal

ossification is present. Upper limb abnormalities on prenatal ultrasonography suggests numerous syndromes. Once radial aplasia has been noticed, ultrasonography of the extremities, face, and kidney is indicated.

Bone marrow sampling may reveal decreased or absent megakaryocyte; small, basophilic, vacuolated megakaryocytes; erythroid hyperplasia or normal myeloid precursor.

The management of TAR syndrome involves precaution during times of significant thrombocytopenia with platelet count less than $80 \times 10^9/L$ (usually during the first year of life) to avoid trauma, avoidance of antiplatelet medications (e.g aspirin, NSAIDS) and prolonged pressure on injection sites especially after intramuscular injections⁵.

The main stay of hospital treatment is supportive care and by far the most significant treatment is platelet transfusion. The goal of platelet transfusion is to maintain sufficient platelets to prevent bleeding without adverse effects. Prophylactic transfusion with leucocytes reduced platelet concentrates are used in patients at high risk of significant heamorrhage⁵.

Splenectomy is usually effective for the treatment of thrombocytopenia in adults.

Bone marrow transplantation (BMT) is an option for patients who continue to remain thrombocytopenic with bleeding despite platelet transfusion.

Surgical care includes splinting of the hands (and legs if indicated) during infancy to improve future function and operative correction of the arm deformities if the patient is haemodynamically stable.

Complications usually arise from hemorrhagic insults especially intracranial haemorrhage.

The clinical course is one of episodic severe

thrombocytopenia superimposed on a back ground of persistent thrombocytopenia⁶.

The frequency of thrombocytopenia episodes decrease with age. By school age, near- normal platelet counts are expected. If a patient survives the initial 2 years of life, life expectancy is normal⁷.

Summary

TAR syndrome is a rare genetic disorder characterized by low platelet level and radial aplasia. A case of a nine day old female with TAR syndrome was presented to highlight the radiological features. The patient was transfused with platelet with platelet concentrate having presented with low platelet count, and she improved there after.

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