Case Report:

Thrombocytopenia with absent radii (TAR) Syndrome in a male child: report of a rare case.

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Abstract:

Introduction: Thrombocytopenia with absent radii (TAR) Syndrome is a rare congenital defect presents with hypomegakaryocytic thrombocytopenia and bilateral radial aplasia and preserved thumb. Case presentation: A three and half year’s old male baby born out of non-consanguineous marriage with thrombocytopenia and bilateral absence of radius. Such type of anomaly has been previously reported in the children of a non-consanguineous marriage was few. Conclusions: Though rare incidence, all thrombocytopenia with any skeletal deformity cases in newborn or infancy to toddler age group must be thoroughly investigated to exclude TAR syndrome. Case is presenting here because of rarity.

Keywords: Thrombocytopenia, Absent of radius, TAR syndrome.

Introduction:

Thrombocytopenia with absent radii (TAR) syndrome is an autosomal recessive genetic rare disorder. Hypo megakaryocytic thrombocytopenia and bilateral absent of radius that may have an additional anomalies1. It was first described in 1929 but designated as a syndrome in 1969 by Hall et al2. It is not a variant of Fanconi’s anemia3. The two consistently essential features are hypomegakaryocytic thrombocytopenia and bilateral radial aplasia. The other associated features are limb abnormalities, intermittent leukocytosis, eosinophilia, cardiac defects, renal anomalies, mental retardation, and milk protein allergy4. The most important cause of mortality among children with TAR syndrome is bleeding. The rate of mortality in this syndrome primarily depends on the platelet count and other accompanied anomalies. The most frequent cause of death was due to the intracranial bleeding, especially during the first two years of life5,6.

Case Report:

A three and half years old male child was born out of non-consanguineous marriage. Pregnancy was uneventful. No history of significant maternal illness during pregnancy. The primi gravida mother without any, history of abortion was healthy throughout the gestational period and had normal vaginal delivery. The baby had normal weight at birth and no evidence of prematurity at birth. Family history was not significant. Since birth till six month baby had no significant complain and reported to orthopaedic department for orthopaedic problem.

Investigation reports:

Face-not dysmorphic, normal intelligence (fig-1). Complete blood count showed only thrombocytopenia (11,000/µl). Bone marrow aspiration study showed markedly reduced megakaryocyte. Orthopedic evaluation showed bilateral absence of radius with preserved thumb and functional fingers on each hand. No other skeletal deformity noted (Fig-II). USG-whole abdomen showed mild hepatomegaly and absence of right kidney but the patient had no kidney related problem. 2D Echocardiography

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showed patent foramen ovale and small sub-aortic ventricular septal defect (VSD)

Discussion:
The thrombocytopenia-absent radius (TAR) syndrome is a congenital malformation syndrome characterized by bilateral absence of the radii and a thrombocytopenia. The lower limbs, gastrointestinal, cardiovascular, and other systems may also be involved. Shaw and Oliver in 1959 were the first to describe this condition, but it was Hall et al in 1969 who reported the first major series of patients. In this reported case isolated thrombocytopenia and bilateral absence of radius with preserved thumb and functional fingers on each hand, with absence of right kidney and cardiac problem patient was diagnosed as TAR syndrome. Varying degrees of thrombocytopenia, present in 100% of diagnosed cases with TAR syndrome. In earlier studies researchers found that patients with TAR syndrome were usually diagnosed at birth, due to thrombocytopenia as they present with petechial rash or bleeding manifestation like bloody diarrhea in the first week of life or later during the next four months. Platelet counts at birth are usually 15,000 to 30,000/uL (4, 7) but in present case it was not found at neonatal life. Present case presented at 6 month with few petechial rashes with platelets count of 11000/μL. PBS and bone marrow examination shows isolated hypo megakaryocytic thrombocytopenia. The presenting case also ruled out other possible cause of thrombocytopenia.

Though the exact cause of thrombocytopenia in TAR syndrome is not clearly known but the possibilities are a) the absence of humoral or cellular stimulators of megakaryocytopoiesis (b) the absence of megakaryocytic progenitor cells (c), cellular defects in megakaryocytic precursors (for example, receptor defects) or (d) the presence of humoral or cellular inhibitors of megakaryocytopoiesis (2). Bone marrow examination showed marked reduction of megakaryocytes in this presenting case (1).

The other most important sign of TAR syndrome is bilateral absence of radii. The upper limb abnormalities range from isolated absent radii with preserved thumbs like this case, to phocomelia. The etiology of radial aplasia in TAR syndrome is a primary failure of chondrogenesis. Lower limb abnormality occurs in 46% cases, but less severe than upper limb defects (8). The presenting case showed bilateral absence of radius with preserved thumb and functional fingers on each hand. No other skeletal deformity noted.

Congenital heart anomalies (Tetralogy of Fallot, atrial and ventricular septum anomalies) appeared in 22-33 % of such cases (2,3). Patent foramen ovale and small sub-aortic ventricular septal defect (VSD) were observed in presenting case. In 2000 there was first report of horseshoe kidney in association with TAR syndrome followed by a clinical study of 34 cases with TAR syndrome in 2002 where horseshoe kidney was noted in two cases (9). But our case presented with absence of right kidney comparatively in high age was rarely reported.

Conclusion:
TAR syndrome though rare but in new born babies and infants with thrombocytopenia and bloody diarrhoea even, all thrombocytopenia with any skeletal deformity cases in newborn or infancy to toddler age group must be thoroughly investigated to exclude TAR syndrome.
References:-


