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Case Report

Thrombocytopenia absent radii syndrome in a twenty five day old female neonate: a case report

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ABSTRACT

Thrombocytopenia absent radii (TAR) is a rare genetic disorder associated with multiple additional anomalies. We report a case of a 25 day old female neonate with thrombocytopenia and bilateral absent radii with flexion of the elbow joint and radial deviation of the wrist joint. TAR, although rare are not uncommon. Thus, we have presented a 25 day old neonate with radiological features of absent radii and fluctuating platelet counts that subsequently showed thrombocytopenia that warranted the diagnosis of TAR syndrome in this case.

Keywords: Thrombocytopenia, Radii, Syndrome, Neonate

INTRODUCTION

Thrombocytopenia absent radii (TAR) syndrome is a rare congenital defect that is characterized by reduction in the number of platelet counts and absent of the radius.[1] It was initially thought to be a variant of Fanconi's anemia but now known to be a separate entity.[2] This syndrome was first described in 1951 with some families having more than one member affected.[3] In the united stated, this syndrome rarely occur, so also in Nigeria, however no definite incidence are obtainable. Internationally the incidence rate of 0.42 cases per 100,000 lives birth is reported in in Spain. With a male to female ration being 1:1, there is no ethnic or racial predilection reported for TAR so far.[4]

The rarity of this congenital malformation in our environment prompts the report of this case.

CASE REPORT

ZA is a twenty five day old female neonate. She is the first child of a twenty two year old primi para in a monogamous setting. She attended her antenatal care in our hospital, Usmanu Danfodiyu University Teaching Hospital (UDUTH), Sokoto. The pregnancy was carried to term and delivery was vertex and spontaneous and uneventful. There was no history of maternal illness during the pregnancy, was only placed on the routine anti-malaria prophylactic dose (pyrimethamine), and haematinic (folic acid and fasolate).

Physical examination shows a healthy stable child with normal vital sign. The musculoskeletal examination revealed bilaterally shortened forearm with fixed elbow flexion and radial deviation. No sign of baby rashes or active bleeding site. Thumb and the nail eminences were normal. Echocardiography and abdominal examinations were essentially normal.

The laboratory examination show normal platelet counts at birth as well as the pack cell volume (PCV), neutrophils, eosinophils, electrolyte and creatinine and liver function test. On follow up laboratory investigation show the platelet counts to be slightly deranged from normal value without clinical symptom.

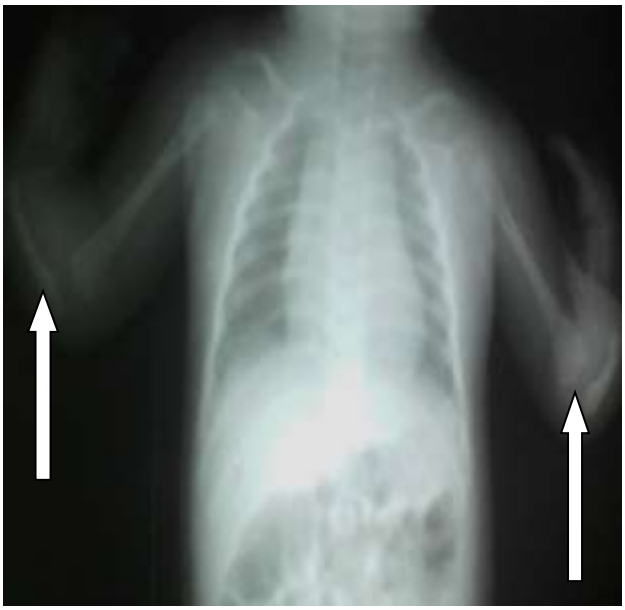

Figure 1: A plain radiograph showed congenital absent radii (arrows) with hypoplastic of both ulna bones and flexions of both elbow joints.

Echocardiography and abdominal ultrasound were essentially within normal limits. Skeletal survey X-ray shows bilateral absent radii with flexion of the elbow joint and radial deviated wrist joint. The demonstrated carpal, metacarpal and phalanges are within normal limits.

On the bases of the follow up deranged platelet counts and the radiograph that show absent radii and normal thumb lead to the diagnosis of TAR syndrome. Subsequent follow up we discovered that the mother developed depression and was receiving treatment in the psychiatric department and the child developed malnutrition and died of its complication from electrolyte derangement and aneamia.

DISCUSSION

Thrombocytopenia absent radii (TAR) is a rare genetic disorder that may be associated with multiple additional anomalies.[1] Thrombocytopenia which may be transient is seen in 100% of cases diagnosed with TAR syndrome.[5] Patients are usually diagnosed at birth due to thrombocytopenia as they will present with patechial rash or bloody diarrhea in the first week of life or late during the next four month of life[6] this was not found in our case. TAR is said to present with symptomatic thrombocytopenia in the first week of life but this was not the presentation in our case.[3] Fifty percent of patients are symptomatic in the first week of life and others at age of four.[7] The frequency of thrombocytopenia episodes decreases with age. By school age near normal platelet counts are expected. If patients survive the initial two years of life, life expectancy is normal.[8]

The two features that are essential for the diagnosis of the syndrome are thrombocytopenia and bilateral absent radial aplasia as in case here presented.[1] The association of radial aplasia with thalidomide poisoning has been reported.[9] In our case there was no history of thalidomide poisoning.

All TAR syndromes should have documented thrombocytopenia because the platelet counts fluctuate over time. Therefore, if there is a strong clinical suspicious of TAR syndrome with normal platelet counts, it should be repeated as seen in our case. A single platelet counts does not exclude TAR.[10]

In distinguishing TAR from other syndrome 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. Thumb is typically present in Holt Oram syndrome but blood counts are normal.[3]

Other numerous anomalies described in TAR include; asymmetric first rib, cervical ribs, spinal bifida, fused cervical spine, nuchal fold, Meckels diverticulum, uterine anomalies, dorsal pedal edema, short stature, large horseshoe shaped kidney.[3] All of these were not found in our patient.

The main stay of hospital treatment is supportive care and by far the most significant is platelet transfusion in order to prevent bleeding without adverse effect.[3,9]

CONCLUSION

Thrombocytopenia absent radii (TAR) is a rare genetic disorder but not uncommon. Thus, we have presented a 25 day old neonate with radiological features of absent radii and fluctuating platelet counts that subsequently showed thrombocytopenia that warranted the diagnosis of TAR syndrome in this case.

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