A VARIANT OF THROMBOCYTOPENIA- ABSENT RADIUS SYNDROME
TONY SIMON GERMANS GABRIEL GERMANS
Department of Paediatrics, CHENGALPATTU MEDICAL COLLEGE

Abstract: A newborn baby was admitted in our NICU for respiratory distress with antenatal USG revealing a congenital heart disease and an absence of the left radius. Clinical examination revealed the presence of an apical impulse on the right, with a malformed left forearm and a rudimentary thumb. Echocardiogram showed dextrocardia with patent foramen ovale. Ultrasound revealed situs inversus totalis. Hemogram showed Thrombocytopenia.

Keyword: Absent radius, dextrocardia, situs inversus totalis, thrombocytopenia.

INITIAL PRESENTATION
A 23 year old primi mother delivered a term male baby (Figure 1) via labour naturalis in our hospital. Baby did not cry at birth, but cried after stimulation. Baby was found to have deformed left upper limb with rudimentary thumb. Baby had respiratory distress soon after birth and was shifted to the neonatal intensive care unit for respiratory support. Vitals were stable. Apical impulse was present on the right. No history of consanguineous marriage, congenital anomalies or heart disease was present in the family. No history of any antenatal infection.

Figure 1: The baby at birth

On Examination of the neonate at birth
- Cry and activity was fair, baby was pink,
- Normothermic, tachypnoeic
- Apical Impulse felt on the right side
- Femoral pulse palpable bilaterally
- CVS: S1S2 heard, heart sounds heard well on the right side
- No murmur, no visible precordial pulsation seen.
- RS: BAE heard and equal, minimal SCR (+)
- P /A : soft, liver just palpable in left hypochondrium
- CNS: Tone normal, neonatal reflex present, Anterior fontanell-flat, pupils equal and reacting to light

Local Examination
Upper limbs: deformed upper limb with rudimentary thumb (Figure 2)

Figure 2. Rudimentary thumb
Lower limbs: normal on both sides.

Vitals
Heart Rate: 136/min
Respiratory Rate: 70/min
Saturation: 100 % on all 4 limbs

Hospital course
Baby was treated for respiratory distress, which improved with oxygen within 24 hours. Baby was thoroughly evaluated for other congenital anomalies. The following investigations were done when the child was in the neonatal intensive care unit

INVESTIGATIONS
NICU:
- CBC: TC: 9900, DC: P:68, L:30, E:2, Hb:12g, RBC: 4 million cells
- PCV: 36, Platelet: 50,000 cells per cumm
- Xray left upper limb: absent radius (Figure 3)
- USG Abdomen: situs inversus totalis.
- USG Cranium: normal study.
- ECHOCARDIOGRAM: Dextrocardia with patent foramen ovale.
- BLOOD GROUP: O Positive.
- PERIPHERAL SMEAR: Thrombocytopenia.

Figure 3 absent radius left forearm
TREATMENT
Warmth, Oxygen, IV Fluids, Paladay feeds and Breast feeds.

DISCHARGE
The newborn was discharged after 2 weeks of hospital stay. Parents were reassured about the newborns condition. Further course was uneventful. The child was kept on follow up.

Figure 4: Xray showing SITUS INVERSUS TOTALIS with absent radius left forearm

HROMBOCYTOPENIA- ABSENT RADIUS SYNDROME (TAR)
Gross, Groh, and Weippl described this entity in 1956, subsequently over 100 cases have been reported (1). But literature review also suggest that, it was at the same time described by Shaw and Oliver in 1959(2) Hall et al set the current diagnostic criteria for Thrombocytopenia- absent radius in 1969 (2). These include bilateral absence of radii with the presence of both thumb and thrombocytopenia

GENETICS
Autosomal Recessive pattern of inheritance (1)

PREVALENCE
The prevalence of TAR syndrome is estimated at 1,200,000-1,100,000.

CLINICAL FEATURES
Ø Skeletal features
o Arms: Bilateral absence of radius (100 %) Abnormalities of ulna include hypoplasia (100%), bilateral absence of ulna (20%) and unilateral absence of ulna (10%). Abnormal humerus(50%) with bilateral absence in 5- 10 %. The thumbs are always present (1). The classical feature is the absence of radius. The presence of the thumbs distinguishes TAR syndrome from other disorders featuring radial aplasia, which are usually associated with absent thumbs. Variants with unilateral absence of radius with esophageal atresia have been reported (3). This case is being reported as variant of TAR like the above study with single radius but with situsinversus and developmental delay.

Ø Legs: Abnormalities are present in 50% including hip dislocation, subluxation of knee, coxa valga, dislocation of patella, femoral and tibial torsion, abnormal tibiofibular joint, ankylosis of knee, small feet, abnormal toe placement. Absence of fibula

Ø Hematological: Thrombocytopenia which is present is severe in early infancy. The thrombocytopenia is associated with absence or hypoplasia of megakaryocytes ( absent in 66%, decreased in 12%,inactive in 12%). “Leukemoid” granulocytosis is seen in 62 percent of cases especially during bleeding episodes Eosinophilia is seen in 53 %. Anemia is out of proportion to apparent blood loss (1). The thrombocytopenia which may be transient is seen in all cases and will be symptomatic in over 90% of cases within the rst four months of life. Gastrointestional bleeding and occasionally intracerebral bleeding may result. The latter is an important cause of mortality in these children The advent of platelet infusions has helped to prevent the latter, which was previously the main cause of mortality.

OTHER ABNORMALITIES
Ø Congenital heart disease: Congenital heart disease are present in 33 % of cases and include primary tetralogy of Fallot and atrial septal defect (1)

Ø Gastrointestinal: Cows milk allergy or intolerance is common and can be a significant problem with introduction of cows milk precipitating thrombocytopenia, eosinophilia and /or leukemoidreactions (1). Pancreatic cyst and Meckel diverticulum are other abnormalities reported.
Ø Genitourinary anomalies include renal anomalies (both structural and functional) and in rare cases, Mayer-Rokitansky-Kuster-Hauser syndrome (agenesis of uterus, cervix, and upper part of the vagina). (4)
Ø Central nervous system involvement include mental retardation has been reported in 7 % of individuals, secondary to intracranial hemorrhage (1)(5). Delayed myelination, hypoplasia of the cerebellum, particularly the vermis and a cavum septum pelliculum on MRI has been reported.
Ø Ocular features include strabismus and ptosis.

An Initiative of The Tamil Nadu Dr. M.G.R. Medical University
University Journal of Medicine and Medical Specialities

Bibliography
(1) Smith’s Recognizable Patterns of Human Malformation: 5th edition. Saunders, an imprint of Elsevier

DIFFERENTIAL DIAGNOSIS
Ø Holt Oram syndrome
Ø Fanconi syndrome

Natural course: About 40% of patients have die usually as a result of hemorrhage during early infancy. Thrombocytopenia during that time is precipitated by viral illness. The platelet count tends to rise as the child gets older and may approach normal levels in adulthood (3). Delayed motor abnormalities due to skeletal abnormalities are seen. Bracing, splinting, or stabilization of wrist should be considered. Arthritis of wrist and knee are late complication

INVESTIGATIONS
Ø Complete blood count: Document thrombocytopenia and to rule out anemia secondary to any bleeding
Ø Skeletal survey: To document absent radius and to rule out other skeletal manifestations like abnormalities of ulna and humerus.
Ø Echocardiogram: To rule out congenital heart disease.
Ø Ultrasonogram of abdomen: To rule out genito- urinary abnormalities.
 Ø Ultrasonogram of cranium: To rule out intracranial bleed.

TREATMENT
No cure. Treatment is supportive