



New Syndrome or Unreported Feature? Ulnar Ray Deformity with Thrombocytopenia and Overlapping Features of Rubinstein-Taybi Syndrome TAR Syndrome, Cornelia De Lange Syndrome

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Abstract

Upper hand deformities and various skeletal and systemic anomalies are parts of various syndromes. Ulnar bone deformities are extremely rare. We report a case with absent ulna on left side and deformed ulna on right side, absent carpal and metacarpal bones both side except digital bones of thumb were present on both side with various other feature of Rubinstein-Taybi syndrome, TAR Syndrome, Cornelia De Lange syndrome. Till date no such case reported worldwide.

Introduction

Upper hand deformities are commonly associated with various other skeletal and systemic anomalies and parts of various syndromes like Holt Oram, Rubinstein-Taybi syndrome, TAR Syndrome, Cornelia De Lange syndrome. Ulnar bone deformities are very-very rare. We report a case with absent ulna on left side and deformed ulna on right side, absent carpal and metacarpal bones both side except digital bones of thumb were present on both side with various other feature of Rubinstein-Taybi syndrome, TAR Syndrome, Cornelia De Lange syndrome. Whether this is a new syndrome or a new unreported feature of Rubinstein-Taybi syndrome, TAR Syndrome, Cornelia De Lange syndrome. Till date no such case reported worldwide.

Case Presentation

A newborn admitted at neonatal nursery Government Mahila Chikitsalaya Ajmer just after birth with poor cry, sluggishness and malformation of both upper limbs with normal apgar score at birth. Age of mother was 21 year and age of father was 25 year. They had 2nd degree consanguineous marriage. No family history of any congenital malformation of other congenital diseases. No history of any antenatal risk factor except USG shows mild oligohydramnios and intrauterine growth retardation. No history of any drug intake or radiation exposure during pregnancy period. This child is 1st para of 1st gravida mother.

Gestational age assessment by new ballard score was 34 ± 2 weeks. On examination weight, length and occipitofrontal circumference were 1500 gms, 40 cm, 27 cm. Child had both side short deformed forearm with absent of palms and fingers except thumb present in both side with web and contracture at elbow joint both side. Child also had hirsutism, long and distorted eyelashes, sparse eyebrows, beaked nose, long columela, periorbital odema, slight proptosis of eyes, low set ear, low posterior hair lines, retrognathia, high arched palate. Baby also had broad base of great toe both side, syndactyly of 2nd and 3rd toes on right foot, clinodactyly of 5th toe both sides. Child had recurrent nasal block, tearing from eyes, vomiting, and feeding difficulty in nursery.

X-ray chest was normal. X-ray spine normal. X-ray long bones of upper limb showed absent ulna on left side while deformed ulna on right side, absent carpal and metacarpal bones both side except digital bones of thumb were present on both side. Spine and lower limb bones were normal. Ultra-sonography of cranium and abdomen were normal. ECG was normal. Complete blood count showed thrombocytopenia (platelets count of $1.44 \mu\text{l}$, $1.00/\mu\text{l}$, and $80000/\mu\text{l}$) during three different CBC, otherwise CBC was normal. All other blood investigation were normal including CRP, urea, creatinine, SGPT, sugar, Blood culture.

Child was kept on mother feed/suthi feed and IV cefotaxime and amikacin. 10th day onward

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Received Date: 22 May 2017

Accepted Date: 05 Jun 2017

Published Date: 12 Jun 2017

Citation:

Narayan JP, Sharma D, Kumawat R, Jain A. New Syndrome or Unreported Feature? Ulnar Ray Deformity with Thrombocytopenia and Overlapping Features of Rubinstein-Taybi Syndrome TAR Syndrome, Cornelia De Lange Syndrome. *Ann Clin Case Rep.* 2017; 2: 1375.

ISSN: 2474-1655

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child had repeated episodes of vomiting. So again USG abdomen was performed but again that is normal. Stomach was performed, child kept nil by mouth by 2 days. Thorough examination and investigation performed. Injection meropenam, linezolid were started. All investigations were normal. Domperidone drops started and sithi feed tried now child took feed well and on 20 days of life successfully discharged. Further investigations were not performed because parents were not affording cost of advanced investigation.

Discussion

RSTS is characterized by slow development of height and weight, microcephaly, dysmorphic facial features, broad thumbs, and big toes [1]. The prenatal development is normal, with average or near-normal growth parameters at birth. The growth charts typically approach the lower limits of normality in the first postnatal period, primarily reflecting hypo-feeding exacerbated by gastroesophageal reflux. Subsequently, the tendency of overweight or obesity (earlier in males than females) can be observed during adolescence. Specific and recently reviewed growth charts are essential for appropriate assessment of the growth of affected individuals [1]. Facial features are primarily characterized by low frontal hairline, arched/thick eyebrows, downslanting of palpebral fissures, a protruding beaked nose with columella below alae nasi, dysplastic and low-set ears, an arched palate, mild micrognathia, dental anomalies (altered conformation, malocclusion, and overcrowding of teeth), and atypical smile (“grimacing”) with nearly completely closed eyes. The feet and hands typically present an enlarged first finger and clinodactyly of the fifth finger, whereas polydactyly with bifid thumbs and first toes is rarely observed. Other skeletal anomalies include abducted thumbs, vertebral anomalies, ligamentous laxity, severe and prolonged aseptic inflammation of the femur head, anomalies similar with Perthes disease (3%), and occasionally slipped capital femoral epiphysis [1]. Particularly, high risk of cervical vertebral abnormalities (instability of C1–C2, os odontoideum, hypoplasia of the dens, fusion of the cervical vertebrae) has been reported [1-4], with possible stenosis at the craniovertebral junction, which may cause cervical myelopathy. Complex neuroradiological issues including corpus callosum dysgenesis (17%) [3,4], Chiari type I malformation with or without syringomyelia [5], Dandy Walker malformation and hydrocephalus, and tethered cord have been reported and are still under investigation [1]. Cerebrovascular abnormalities such as spontaneous dissection of the supra aortic arteries and cerebral infarction due to dissecting aneurysm of the anterior cerebral artery have also been reported [6].

Ulnar clubhand is secondary to ulnar ray deficiency. This is a rare anomaly and is usually isolated, although it can be in association with Larsen syndrome or TAU syndrome (thrombocytopenia and absent ulna with mental retardation and facial dysmorphism) [7]. The condition may be associated with skeletal dysplasia and arthrogyposis. Prenatal differentiation between ulnar club hand and radial club hand is difficult, and in many cases ulnar club hand is associated with a radial ray defect also. Stoll C et al. [8] reported a case of TAU syndrome with pancytopenia (hemoglobin 9 g, 2,000 PMN, 75,000 platelets) microcephaly, facial dysmorphism, skeletal deformities (kypho-scoliosis, club feet, club hands) and mental retardation. Roentgenograms showed bilateral agenesis of the distal part of the ulna with dislocation of the head of the radius. No other skeletal parts were absent. They reported that condition is probably due to an autosomal recessive gene [8].

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