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SKELETAL DYSPLASIA WITH JAUNDICE : AN UNCOMMON PRESENTATION

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ABSTRACT Skeletal dysplasia is known to have a wide range of presentations and can lead to marked disproportion of the long bones , spine and head in relation to the trunk. Although The incidence worldwide is estimated to be 1 in 5000 live birth, when present they contribute to significant morbidity and mortality. Skeletal dysplasia is subdivided into two types- lethal and non lethal. The most common lethal types include osteogenesis imperfecta, thanatophoric dysplasia and achondrogenesis. Non lethal skeletal dysplasia encompasses about more than 300 subtypes which can be classified according to the region of bone primarily involved (epiphyseal, metaphyseal, and diaphyseal) . Here we present a rare case of a non lethal skeletal dysplasia probably metaphyseal in origin which presented at one month age with an uncommon manifestation i.e prolongation of neonatal jaundice.

KEYWORDS : skeletal dysplasia, non lethal,, metaphyseal, neonatal jaundice

INTRODUCTION

Skeletal dysplasia is an umbrella term that includes hundreds of conditions that can affect bone and cartilage growth. It is also known as osteochondrodysplasia, a group of heritable disorders characterised by abnormalities of cartilage and bone growth. It can be accompanied by involvement of other systems including neurologic, respiratory & cardiac systems. Commonly seen skeletal dysplasia include Achondroplasia, osteogenesis imperfecta, thanatophoric dysplasia, campomelic dysplasia, and hypochondroplasia. Here we are presenting a 1 month old neonate with skeletal dysplasia and jaundice.

CASE REPORT

A 1 month old male baby , late preterm with low birth weight born out of non consanguinous marriage was brought to the hospital with complaints of yellowish discoloration of body since day 2 of life . There was also associated yellowish discoloration of urine. Since last 10 days there is increased yellowish discoloration of urine. The mother also complained of bending of lower limbs and upper limbs evident since birth. No significant antenatal history or family history of any chronic disease or skeletal abnormality could be elicited. There was no history of fever, vomiting, passing white stool or pale stool, cyanosis, lethargy, formula feed, previous fetal loss , birth asphyxia, any significant neonatal events or SNCU hospitalisation. Baby has been given birth dose of OPV, BCG, HBV vaccine.

On head to toe examination, baby has a depressed nasal bridge,micrognathia, short femur and short humerus , microcephaly,brownish hair and eyebrows, hypertelorism, long fingers & toes , bowing of limbs (both UL & LL)- rhizomelic dysplasia. Anthropometry- weight - 2.14 kg (weight/age < -3SD), length -46 cm (length/age< -3SD)

Weight/length - -2SD to -3SD . HC- 32 CM (HC <-3SD)
Upper segment/lower segment = 2:1

General examination- patient has icterus, no pallor, clubbing, cyanosis, lymphadenopathy,edema

Zone V icterus
HR-128/min, RR-42/min, SpO2-94% in room air
Chest and cardiovascular examination reveal no abnormality. As per abdomen examination it is soft, distended with liver and spleen being palpable. on central nervous system examination, anterior and posterior fontanelle is wide open.

Investigations- CBC – Hb-13.7 gm/dl, TLC-9600(N35, L15,M10,B2), TPC-114000

LFT-TOTAL BILIRUBIN-4 MG/DL , DIRECT BILIRUBIN-0.7 MG/DL, AST- 57.6IU/L, ALT-32.8IU/L, ALP-1520IU/L
SERUM CALCIUM -9.8mg/dl
TSH – within normal limits
ALP- within normal levels
TORCH panel- negative for both IgM and IgG
BLOOD GROUPING- mother and baby both B +ve
Serum CPK-138I U/L

USG ABDOMEN AND PELVIS- NORMAL STUDY
OPHTHALMOLOGY EXAMINATION – NORMAL
INFANTOGRAM- no evidence of fractures

TREATMENT – drop vitamin D3 400 IU OD, VITAMIN K ORALLY 3 mg daily for 2 days

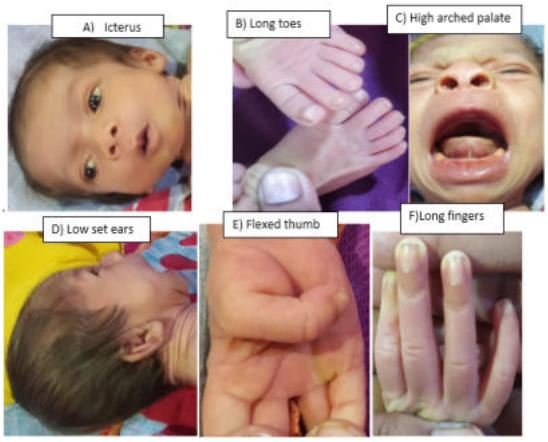
ADVISED FOR FOLLOW UP AFTER 1 MONTH AND MEANWHILE CONTINUE VITAMIN D, DROP ASTYMIN C TWICE DAILY .

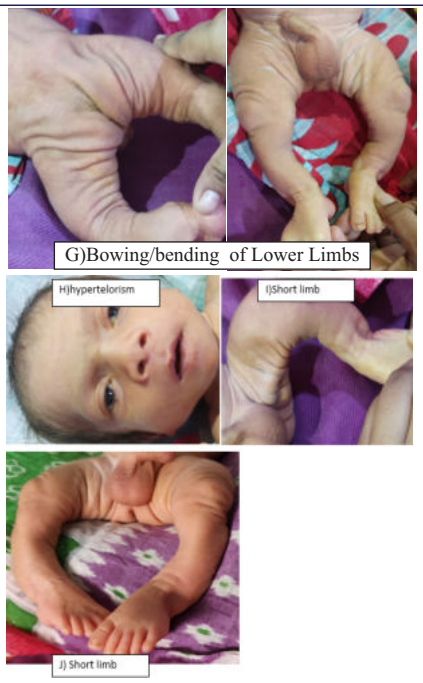
At this juncture a diagnosis of skeletal dysplasia with prolongation of neonatal jaundice was made. As TFT came to be normal and features of cholestasis was not there , by ruling out all possible causes an entity like Breast milk jaundice was made.

On follow up after 1 month , the baby recovered from jaundice . bilirubin came out to be normal which matches with the condition like breast milk jaundice only.

The infantogram has been observed to be normal now and the changes may be seen as the baby grows.. This is a non-lethal form. Counselling given to parents.

Based on clinical features and investigations a diagnosis of Schmid metaphyseal dysplasia with breast milk jaundice was made . This is an early presentation and a rare presentation of this disorder.





Figures A) icteric eyes B)Long toes C)high arched palate D)Low set ears E)Flexed thumb

F)Long fingers G)Bowing/bending of lower limbs H) Hypertelorism I,J)Short limbs

DISCUSSION

Skeletal dysplasia are heterogeneous group of conditions associated with abnormalities of skeleton in the form of bone shape ,size and density which manifest as abnormalities of limbs,chest and skull.Metaphyseal dysplasias are a group of skeletal dysplasias characterised by metaphyseal changes leading to bowing and deformity of upper and lower limb(4). Schmid metaphyseal chondrodysplasia (SCMD) is the commonest metaphyseal chondrodysplasia with an incidence of 3 to 6 cases per million population(1). It is characterised by progressive short stature which develops by age 2 years.it results from disrupted calcification of metaphyseal cartilage and restricted longitudinal growth of long bones with preservation of epiphyses(3,4). The clinical and radiographic features are not usually present at birth but manifest in early childhood with short limbs, genu varum and waddling gait. Facial features and head size are normal. Radiographs show metaphyseal irregularities of long bones (splaying,flaring, cupping), shortening of tubular bones, widened growth plates and coxa vara(3,4). Mild hand involvement with shortening of tubular bones and metaphyseal cupping of the metacarpals and phalanges and vertebral involvement seen in some cases(2,4). Motor milestones are delayed. Most neonates have normal growth parameters and progressive growth failure is evident by 2 years age. Limited mobility due to chronic joint pain may lead to obesity in later life. This case we are reporting is an early presentation of the disorder with a rare association i.e jaundice. There have been no prior reported literature regarding the same. The diagnosis is established with characteristic clinical features, radiographic changes and molecular genetic testing.Important differential diagnosis include osteogenesis imperfecta, campomelic dysplasia, thanatophoric dysplasia,hypophosphatasia and achondrogenesis. (7)

Thanatophoric dysplasia is the most common form of LSD. It is characterized by extremely short limbs, macrocephaly, small chest, platyspondyly(5,7,10). This patient had microcephaly and other features absent so excluded.

Infantile type hypophosphatasia manifests in utero. It is characterized by severe hypomineralization of all bones, micromelia, and low serum alkaline phosphatase levels(7,10) Our patient had normal alkaline phosphatase values.

Campomelic dysplasia is characterized by normal ossification without fractures, shortness and bowing of long bones, especially of the lower limbs, a bell-shaped thorax, hypoplastic scapulae, and narrow iliac wings (7,8). Campomelic dysplasia was excluded in this patient as no hypoplastic scapulae no narrow iliac wings.

Acondrogenesis is characterized by severe micromelia, unossified spine, and a short trunk with multiple rib fractures .(7,13) In our patient no unossified spine was detected.

There has been no published guidelines on treatment(4). Physiotherapy is done. There have been some trials with use of carbamazepine. Since this is inherited by autosomal dominant manner(1), genetic counselling was given to the parents alongwith supportive treatment to the baby.

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