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Case Report

Systemic lupus erythematosus in pregnancy with rare anomaly of rhizomelic chondrodysplasia punctata in baby

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ABSTRACT

Rhizomelic means shortening of the bones closest to the body's trunk. Chondrodysplasia refers to malformation (the dysplasia part of the word) of the cartilage (the chondro part of the word). Punctata refers to an unusual stippling on the end of the bones that join the shoulders and elbows (the humerus) and the top of the leg and the hip (femur). On X-ray an infant's bones look spotty at the ends. Here, we report a case of rhizomelic chondrodysplasia punctate (RCDP) in newborn of a known systemic lupus erythematosus (SLE) patient. Consent for publication of this rare case for academic benefit has been taken from the patient.

Keywords: RCDP, SLE, Autoimmune disease

INTRODUCTION

Rhizomelic chondrodysplasia punctata (RCDP) is a rare disorder of peroxisomal metabolism, with an estimated incidence 1 : 100,000. This is more commonly found in the females. The main features of the disease are shortening of the proximal long bones, punctate calcifications in the metaphysis and epiphysis of long bones and the thoracic and lumbar vertebrae, dysmorphic face, and severe growth retardation.[1]

Systemic lupus erythematosus (SLE) is a chronic autoimmune disease in which the immune system attacks cells and tissue in the body, resulting in inflammation and tissue damage. SLE can affect any part of the body, but most often harms the heart, joints, skin, lungs, blood vessels, liver, kidneys and nervous system. The number of SLE cases varies around the world. The incidence of new cases has been reported to range between 1 and 10 per 100,000 person-years, while overall prevalence varies between 20 and 70 per 100,000. It is more common in women than in men. In particular, women of childbearing age (15-40 years), are 9 times more likely than men of the same age to develop the disease. SLE is also more prevalent in certain ethnic groups. African-American are three times more susceptible to the disease than Caucasians. Asians are at higher risk of developing the disease.[2]

Although patients with SLE are as fertile as women in the general population, their pregnancies may be associated with complications. The prognosis for both mother and child is best when SLE has been quiescent. Here, we report a case of rhizomelic chondrodysplasia punctata in newborn of a known SLE patient

CASE REPORT

A primigravida 33 years female patient, with 35 weeks of gestation presented to emergency of hospital with complaint of leaking per vaginum. She was a booked case. She was a known case of systemic lupus

erythematosis since 7 years and was taking tablet cortisure 6mg, tablet chloroquine 200 mg daily along with calcium, zinc and folic acid which were continued throughout her pregnancy. Her pregnancy was uneventful and SLE remained quiescent during this pregnancy. Her anomaly scan showed no anomalies but last trimester ultrasound showed shortening of proximal long bones and absence of nasal bone with flat nose, intrauterine growth restriction. Patient had an average height and no family history of dwarfism or long bone disorders was present.

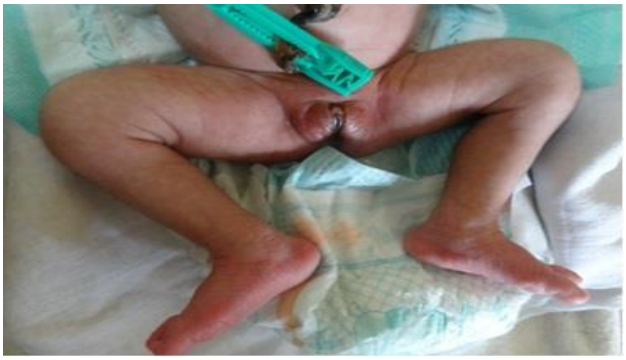

Figure 1: Bilateral short femur.

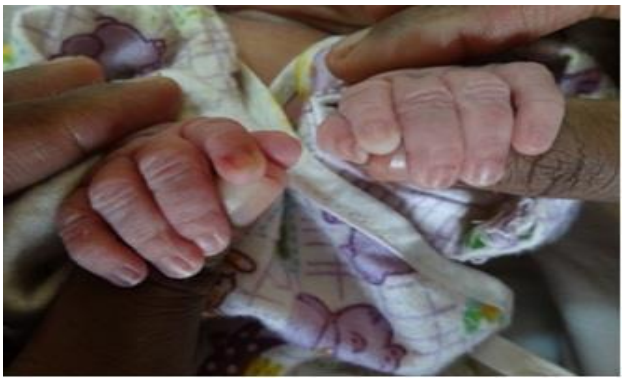

Figure 2: Bilateral short index finger.

Figure 3: Absent nasal bridge.

After admission, Patient was started with oxytocin for induction of labour, finally delivered by caesarean section for indication of failure of induction.

Baby was attended by paediatrician. It was a female child of weight 1.78 kg, detected to have bilateral short femur, bilateral short humerus, and bilateral short index finger, absent nasal bridge. Eyes showed no evident abnormality. The baby was shifted to neonatal intensive care unit for observation and further investigations.

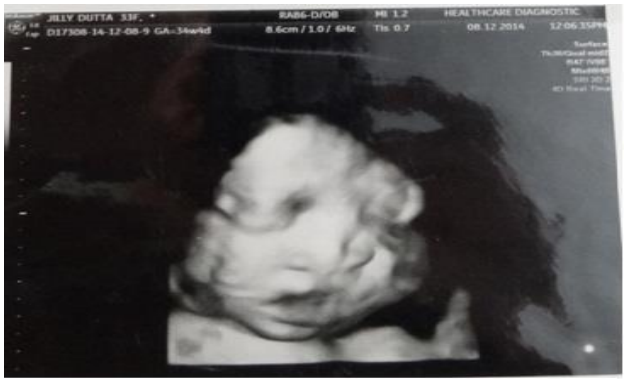

Figure 4: 3D ultrasound picture showing absent nasal bridge.

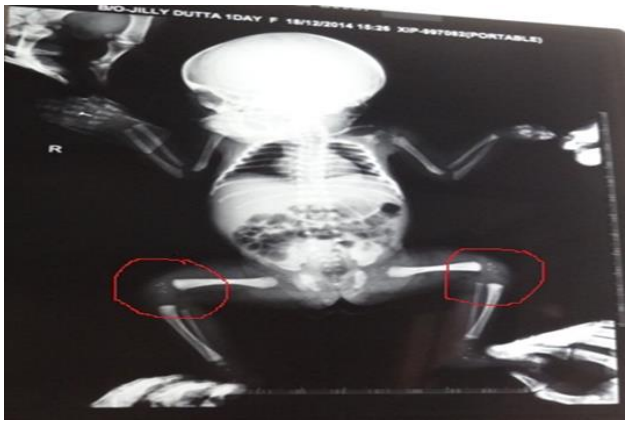

Figure 5: Infantogram suggesting chondrodysplasia punctata and bilateral short proximal long bones (humerus and femur).

Intra operative and post-operative period of mother was uneventful. She was continued with tablet cortisure and chloroquine and breast feeding was allowed.

Investigations of the baby showed cardiac anomaly-acyanotic congenital heart disease with ventricular septal defect and tricuspid regurgition. Infantogram suggested stippled epiphyses, bilateral short proximal long bones (humerus and femur). Karyotype of baby was normal. Mother and baby were discharged after 8 days of observation. Till date both mother and baby doing well.

DISCUSSION

Chondrodysplasia punctata congenita is an entity of genetic heterogeneity characterized by the presence of stippled epiphyseal and extra-epiphyseal calcifications in X-rays. There are at least three distinct types which differ

in their mode of inheritance: the autosomal recessive, the X-linked dominant and the autosomal dominant type. Recently a mesomelic chondrodysplasia punctate has been recognized. It is associated with de novo translocation (13;18) (q14:q23): Mesomelic dysplasias are characterized by limb shortening which is most prominent in the middle segment of the extremities (forearm and lower leg) in contrast to rhizomelic chon drodysplasia punctata where there is shortening of proximal long bones. In addition to several syndromic forms a few patients with sporadic or familial forms and without precise nosological classification have been reported so far.[3]

The diagnosis chondrodysplasia punctata congenita is mainly based on radiological examinations. Prenatal diagnosis of RCDP is possible from first trimester onwards by demonstration of peroxisomal dysfunction in cultured chorionic villous or amniotic fluid cells, but this was not offered in our case.[1]

Ultrasonographic antenatal diagnosis has been reported in severe cases in late 2nd and 3rd trimester by detecting severe growth restriction, skeletal asymmetry and polyhydramnios.[4]

CONCLUSION

Chondrodysplasia punctata is a rare anomaly. To our knowledge, this is the first case reported so far in literature where SLE in mother is co existent with RCDP in baby along with multiple other malformations

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