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Radio-Diagnosis

CHONDRODYSPLASIA PUNCTATA – AN ANTENATAL CASE REPORT

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ABSTRACT Chondrodysplasia punctata (CDP) represents a heterogeneous group of rare skeletal dysplasias characterized by punctate calcifications in the epiphyseal cartilage, a flat or depressed nasal bridge (Binder's facies or midfacial hypoplasia), and can be associated with other abnormal skeletal findings. The condition may be associated with various genetic mutations and can present in both rhizomelic and non-rhizomelic forms, each with distinct clinical implications or acquired form of CDP that can occur secondary to maternal autoimmune diseases. We report a case of chondrodysplasia punctata diagnosed by ultrasound imaging at 20 weeks' gestation. Imaging features are described and differential diagnosis with other forms of chondrodysplasia punctata is discussed.

KEYWORDS :

INTRODUCTION:

Chondrodysplasia punctata (CDP) is a group of skeletal dysplasias characterized by stippled (punctate) calcifications within the epiphyseal cartilage. While most forms of CDP are genetic—such as rhizomelic CDP due to peroxisomal disorders—there is also a less common, acquired form of CDP that can occur secondary to maternal autoimmune diseases, particularly Systemic Lupus Erythematosus (SLE).

Systemic Lupus Erythematosus (SLE) is a chronic autoimmune disease that can affect multiple organ systems and is characterized by the production of various autoantibodies, including anti-Ro (SSA) and anti-La (SSB). In pregnant women with SLE, these autoantibodies can cross the placenta and interfere with fetal development. One recognized fetal manifestation is lupus-associated chondrodysplasia punctata (LACP)-a non-genetic, teratogenic form of CDP.

Infants born to mothers with SLE may exhibit transient stippled calcifications in the vertebral bodies, long bones, and other cartilage-rich regions. Unlike inherited forms of CDP, lupus-associated CDP is typically self-limiting and may not be associated with long-term skeletal deformities.

Case Details:

We present the case of a 29-year-old woman, gravida 2, abortion 1 (G2A1), who presented to the antenatal outpatient department on 22nd July 2024 for a Targeted Imaging for Fetal Anomalies (TIFFA) scan at 20 weeks of gestation, calculated based on her last menstrual period (LMP) of 19th February 2024 and an expected date of delivery (EDD) of 26th November 2024.

The patient had a previous obstetric history of missed abortion at 12 weeks of gestation in 2022, managed by dilation and evacuation (D&E). Following the miscarriage, she developed constitutional symptoms, including weight loss, anemia, and malar rash. Further evaluation revealed autoimmune hemolytic anemia secondary to systemic lupus erythematosus (SLE), with positive antinuclear antibody (ANA) and anti-ribonucleoprotein (anti-RNP) serologies, confirming the diagnosis of lupus.

She was managed with the following immunomodulatory and supportive medications:

- Prednisolone 2.5 mg orally once daily (OD)
- Hydroxychloroquine (HCQ) 200 mg orally at bedtime (HS)
- Azathioprine 50 mg OD (later discontinued)
- Ecosprin (aspirin) 150 mg HS (initiated on 20th July 2024)

In the current pregnancy, a viability scan performed on 12th April 2024 showed a uterine didelphys with a single live intrauterine gestation located in the left uterine horn, corresponding to a gestational age of 6 weeks and 4 days.

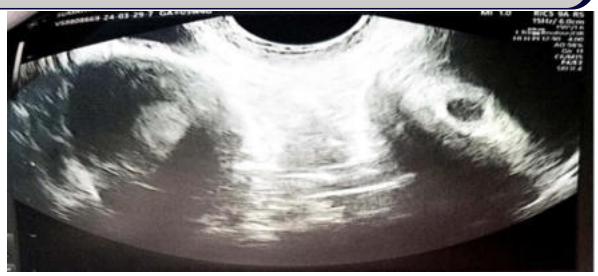

Fig:1 Transvaginal sonography scan showing two uterine cavities with g-sac in left uterine cavity.

A nuchal translucency (NT) scan performed on 21st May 2024 demonstrated a single live intrauterine gestation of13 weeks + 1 day with nuchal translucency measuring 2.3 mm and presence of nasal bone.

Due to her high-risk status, including SLE with hemolytic anemia and previous miscarriage, serial fetal monitoring was performed. A growth scan at 17 weeks revealed biometric parameters consistent with gestational age.

On 22nd July 2024, a TIFFA scan was performed, which revealed a single live intrauterine gestation with a gestational age of 21 weeks and 5 days, and biometric growth parameters appropriate for gestational age. The scan showed notable craniofacial abnormalities, including a flat facial profile and an obtuse frontonasal angle. In addition, skeletal abnormalities were observed in the form of punctate echogenic foci within the epiphyses of the long bones.

Based on the radiological findings, a diagnosis of Chondrodysplasia Punctata Syndrome (CDP) was made. Given the maternal history of SLE with autoimmune hemolytic anemia, a diagnosis of lupus-associated Chondrodysplasia Punctata, a rare non-genetic form of CDP, was considered.

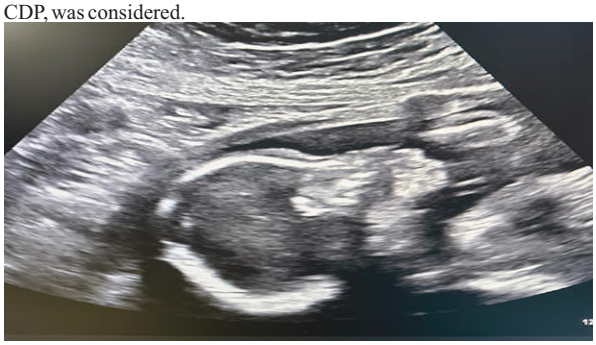

Fig 2: Mid sagittal face shows:Flat face and obtuse naso-frontal angle.

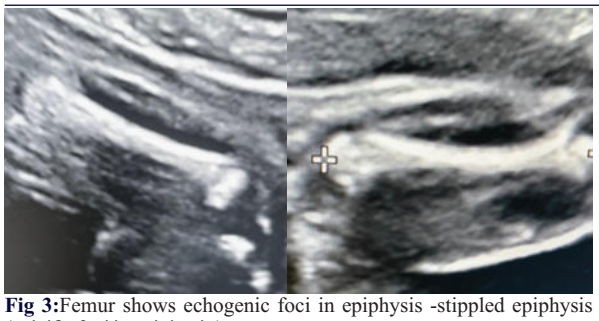

Fig 3: Femur shows echogenic foci in epiphysis -stippled epiphysis (calcific foci in epiphysis)

Termination of pregnancy was carried out on 26th July 2024. The induction protocol began with oral administration of Tablet Mifepristone 600 mg on the same day. This was followed by Tablet Misoprostol 200 mcg, administered per vaginally every 4 hours, for a total of five doses. Subsequently, Injection Oxytocin was initiated on 30th July 2024 at 10:54 AM to augment uterine contractions and facilitate completion of the termination process.

Following termination of pregnancy, physical examination of the fetus revealed dysmorphic features, notably low-set ears and a flattened facial profile, consistent with the craniofacial abnormalities observed on prenatal ultrasound. A skeletal radiograph was subsequently performed, which demonstrated punctate calcifications (stippling) in the epiphyses of the long bones, along with flattening of the midface and nasal bridge. These postnatal findings confirmed the prenatal diagnosis of Chondrodysplasia Punctata, supporting a likely teratogenic or autoimmune etiology related to maternal systemic lupus erythematosus.

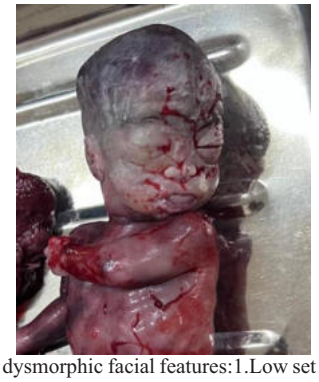

Fig 4: showing dysmorphic facial features:1.Low set ears,2.Flat nasal bridge,3.Large philtrum, 5.Macroglossia .

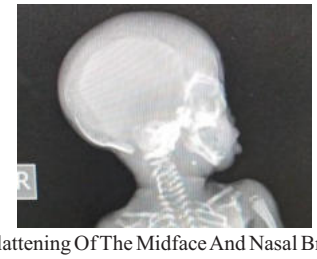

Fig 5: Shows Flattening Of The Midface And Nasal Bridge.

Fig 6: punctate calcifications (stippling) in the epiphyses of the long ChatGPT

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