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RESEARCH ARTICLE

A RARE CASE OF PRENATAL DIAGNOSIS OF ARTHROGRYPOSIS MULTIPLEX CONGENITA

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Abstract

It is challenging to diagnose antenatal identification of fetuses with multiple congenital contractures or arthrogryposis multiplex congenita (AMC) .The first clinical sign is often reduced fetal movement and/or contractures, as seen on prenatal ultrasounds. This can be apparent at any point, from early to late pregnancy, may range from mild to severe involvement, with or without associated other structural anomalies. Possible etiologies and their prognosis need to be interpreted with respect to developmental timing. Essentially, the severity of the disease is based on the number of joints involved. We report a case of a 17-year-old primigarvida woman who visited a tertiary care hospital in for routine antenatal scans. No movement with reduced muscle mass was seen in all the limbs. The hip joints was fixed and flexed in position whereas the knee joints were in an persistent extended position along with severe talipus deformity . All presenting features were indicative of AMC ,poor prognosis of baby and correlated with the post delivery examination of baby.

Introduction:-

Arthrogryposis multiplex congenita (AMC) refers to the development of multiple joint contractures affecting two or more areas of the body prior to birth. It affects approximately 1 in 3000 individuals, mostly reported in individuals of Asian, African and European descent with equal incidence in males and female [4] .The first clinical sign is often reduced fetal movement and/or contractures, as seen on prenatal ultrasounds. This can be apparent at any point, from early to late pregnancy, may range from mild to severe involvement, with or without associated other structural anomalies. Possible etiologies and their prognosis need to be interpreted with respect to developmental timing. The etiology of AMC is highly heterogeneous ,not inherited in most cases; however, a genetic cause can be identified in about 30% of affected people. It may be inherited in an autosomal recessive, autosomal dominant or X-linked manners. More than 400 different conditions can cause isolated or multiple contractures and the causes, genetics,

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specific symptoms and severity of these disorders vary dramatically.Making the specific diagnosis will guide prognosis, counseling and prenatal and perinatal management

Case Report:

A young primigravida woman presented for routine obstetric sonogram evaluation.Her last usg examination was performed at 16 weeks of gestation which was reported normal .From then she didn't undergo any further clinical or usg examination . At present scan a 34-week-old fetuswith cephalic presentation with good cardiac activity , was identified with multiple anomalies, including persistent fixed upper and lower limbs and absent fetal movements , bilateral clubfeet , and polyhydramnios with an amniotic fluid index (AFI) of 30 cm ,bilateral pleural effusions (R>L) ,hypoganthia. Fetal biometry revealed IUGR and Doppler examination of MCA,UA ,DV is normal Genetic counseling and an amniocentesis wererecommended. The patients is not affordable and denied for further testing .The perinatalogistcounseled the patient of the probable poor outcome for her baby based on the sonographic findings, and the patient was directed for immediate delivery. A baby girl was delivered later that evening by cesarean section, as vaginal delivery was not an option due to the severity of the limb contractures. Following delivery, a physical examination was performed, revealing the following findings: arthrogryposis of the upper and lower extremities, ulnar deviation of both hands, multiple contractures of the joints, bilateral clubfeet, cortical thumbs (thumbs in palm), large head ,respiratory distress syndrome. The baby was in NICU for a week and then succumbed to death due to respiratory failure .

Figure (A):- USG Image showing good cardiac activity.

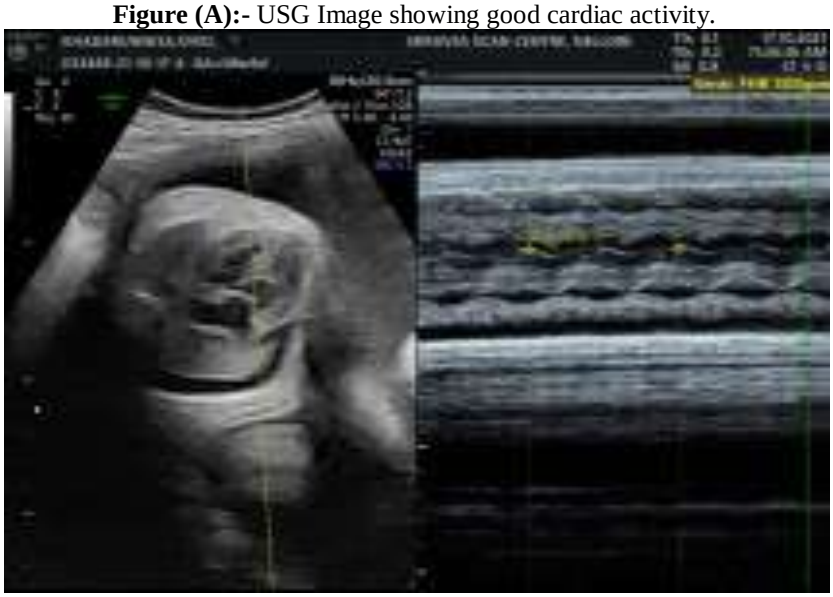

Figure (B):- Image showing hypognathia.

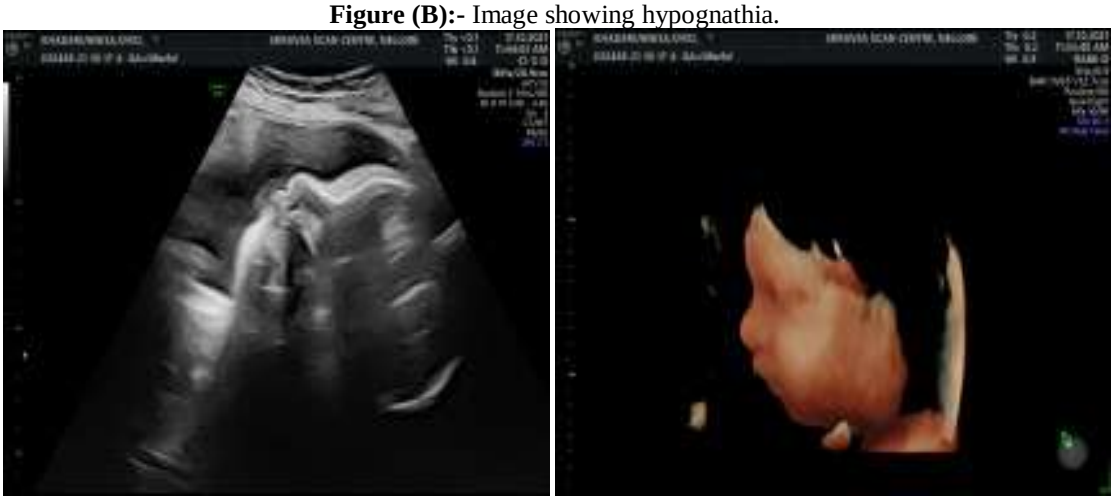

Figure (C):- Image showing pleural effusion (R>L).

Figure (D):- Image showing persistent extension at knee and flexion at hip joint inright lower limb.

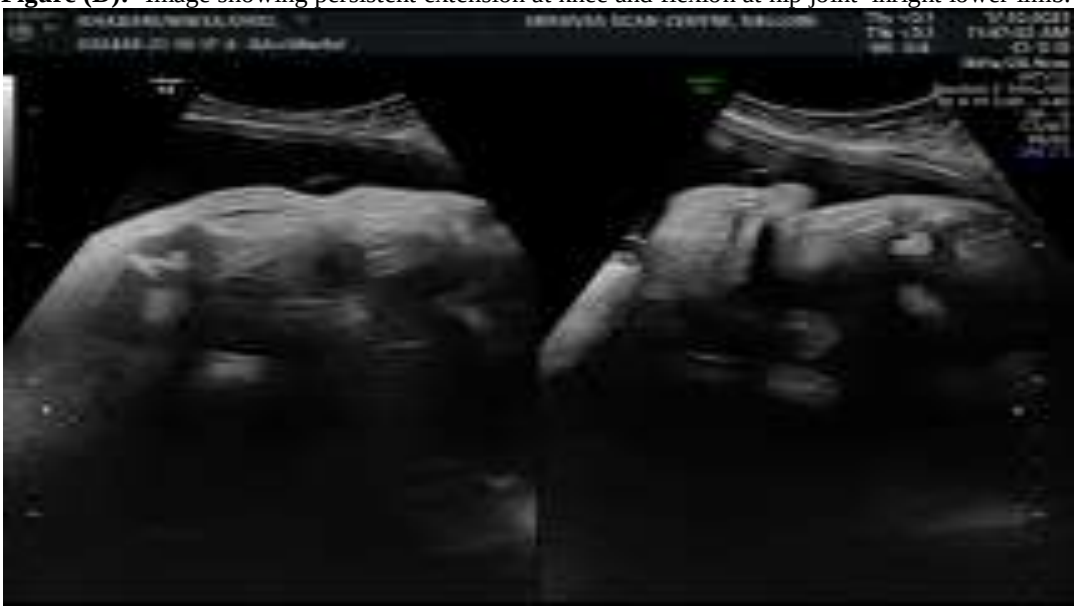

Figure (E):- Image showing bilateral clubfoot.

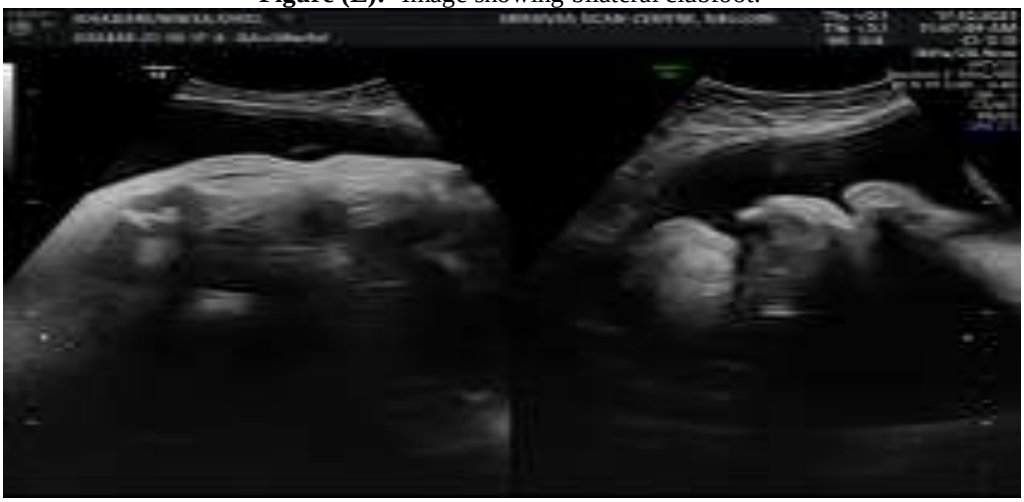

Figure F:- Image showing persistent extension at knee and flexion at hip joint in left lower limb.

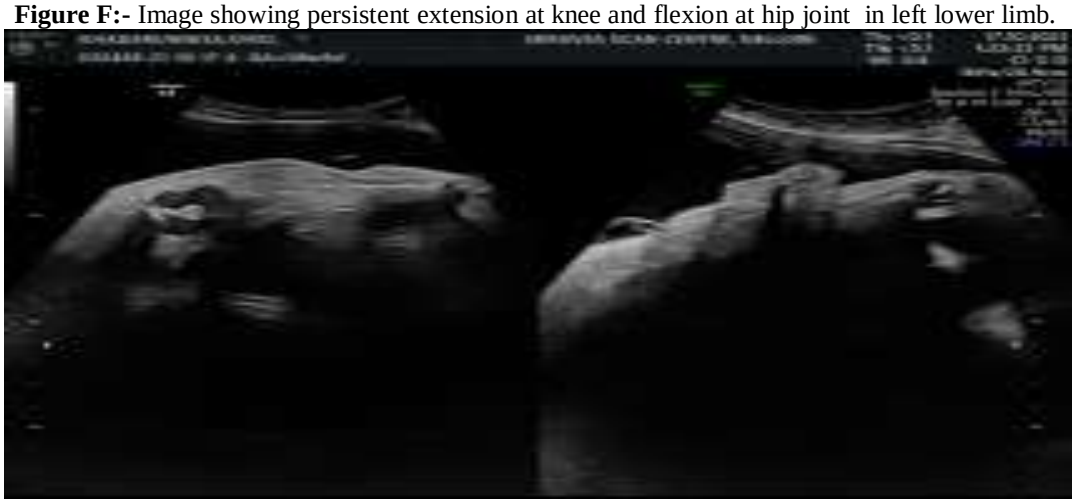

Figure G:- Image of baby after delivery.

Discussion:-

Fetal movement can be observed by ultrasound as early as 8-week gestation. The leg and arm movement and, less often, the movement of feet and hands can be examined by prenatal ultrasonography. However, arthrogryposis may not be diagnosed prior to delivery in spite of numerous prenatal ultrasound studies, primarily because fetal movement is not routinely studied. [4]

In the study by (de Vries& Fong, 2006, 2007) they suggested that the absence of limb movement during 13 min or more should prompt doubling of the observation time, since 13 min is the maximal pause between consecutive movements in a normal population at 20 weeks gestational age .Repeat assessment at 2-week intervals is required to detect changes over time. [1]

When suspicion arises, as in maternal concern about lack of fetal movement, presence of clubfoot,etc., observation for up to 45 min by an experiencedultrasound technician may be needed to examine fetal movement of each limb area. In high-risk familial situations, ultrasound studies to evaluate fetal movement should be done at 14, 16, 18, 20 and 22 weeks and again in mid-second trimester. [4]

Current ultrasound practice identifies only approximately 25% of individuals with arthrogryposis prenatally before 24 weeks of pregnancy in a general obstetrics care population . Though arthrogryposis is more often detected in routine second trimester ultrasound, there are first trimester ultrasound findings that may raise suspicion.These include increased nuchal translucency, cystichygroma, and diminished fetal movement. Othercongenital anomalies associated with arthrogryposis conditions may also be detected [1].

Image findings combined with maternal perception of decreased fetal movement can lead to more frequent diagnosis of arthrogryposis in the second and third trimesters. Club foot is the most commonly identified contracture and should prompt a thorough evaluationof other joints .[4] Other findings related to inherent dysfunction of the skeletal muscle include internally rotated and adducted shoulders, flexed or extended elbows, flexed and ulnarly deviated wrists,clenched hands, flexed and often dislocated hips, and flexed or extended knees .

AMC is not inherited in most cases; however, a genetic cause can be identified in about 30% of affected people. It may be inherited in an autosomal recessive, autosomal dominant or X-linked manners.[3] More than 400 different conditions can cause isolated or multiple contractures and the causes, genetics, specific symptoms and severity of these disorders vary dramatically. [4]

Indirect ultrasound signs of reduced fetal movement are polyhydramnios and collapsed stomach which may be a result of impairedswallowing, and facial anomalies such as micro-/retrognathia. Fetal immobility and disuse of limbs can lead to reduced bone growth, hypoechogenicity, hypomineralization and fractures of the long bones,the persistence of breech position and need for delivery by cesarean section (Hall, Aldinger& Tanaka, 2014; Nowlan, 2015; Nowlan et al.,2010). Such signs and complications usually occur in the late second or third trimester and are not specific to AMC.[2]

Classification:
According to the Munich classifcation, modifed by Hall, there are 3 types of AMC. Type I with mere joint afection as in amyoplasia, with a incidence of 1:10,000 the most common underlying cause of AMC, Type II with other system anomalies, and Type III with further neuromuscular involvement and lethal forms or intellectual disability like FADS.

Other classifications include
1. Amyoplasia,2. Distal arthrogryposis ,3. Systemic connective tissue disorders ,4 .Multiple pterygium syndromes 5. Fetal crowding
"Amyoplasia" (A=no, myo=muscle, and plasia=growth), is the most common type of arthrogryposis seen clinically. It has an incidence of 1/10,000 live births and comprises approximately one-third of arthrogryposis cases.The limb findings in amyoplasiacongenita are usually symmetric, mostly involving all four extremities. However, in some patients only the lower extremities are affected, and more rarely only the upper extremities are affected, while the trunk is spared.1 Patients with AMC affecting only the upper extremities show typical positioning, in which the shoulders are internally rotated, sloped, and rounded. The arms are extended and the wrists and hands are flexed, creating the so-called "police-man tip" position. When the lower limbs are affected, nearly all cases involve contractures around the hips, most of which are severe.Flexion, abduction and external rotation contractures are most common in the hip joints, and dislocation occurs in approximately one-third of patients. The knees may be flexed or hyperextended, and the feet are often in an equinovarus position. The muscle mass of the limbs is diminished and replaced by fibrous tissue, giving the extremities a slender appearance.

Conclusion:-

AMC describes contractures in multiple joints, in more than one area of the body, associated with severe fetal morbidity and future economic burden. Better execution and usage of ultrasound (2D,3D,4D)can give prenatal diagnosis and an opportunity for careful prenatal assessment through thorough image scanning focusing on flexion/extension, position of proximal and distal joints, jaw, and spine and other signs like IUGR , hydrops , polyhydramnios which indicates poor prognosis . When families should be counseled for potential postnatal or post term evaluation when a prenatal diagnosis of AMC is suspected

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