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Arthrogryposis with single umbilical artery: A case report

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Abstract
A 24-year-old patient's second-trimester ultrasound revealed significant forearm mesomelia, arthrogryposis, and a single umbilical artery, aiding in informed pregnancy termination decision.

Keywords: Arthrogryposis, Single umbilical cord artery, Amyloplasia, Mesomelia

Introduction
The term Arthrogryposis is derived from the Greek words Arthron (joint) + Gryp (curved) + Osis (condition). Congenita is a term that is derived from the Latin word “congenitus” meaning present at birth. Arthrogryposis is a sign linked to numerous distinct diseases and syndromes. Multiple joint contractures that are present at birth are referred to by this name. It can be identified on its own or in conjunction with other congenital anomalies as a component of a syndrome involving or not the central nervous system. There is no known cause for arthrogryposis, although all cases entail foetal akinesia and subsequent joint contractures [1]. The most prevalent form of arthrogryposis, amyloplasia is characterized by quadrimellic involvement and skeletal muscle replacement with thick fibrous tissue and fat. Early physical treatment and splinting can help contractures but frequently surgery is required [2]. 350 identified genes and more than 400 medical disorders are linked to arthrogryposis, which exhibits a wide range of phenotypic expressions. Decreased foetal mobility throughout the development is the main underlying mechanism. By detecting clubfoot or joint contractures and foetal movement restrictions, prenatal imaging is essential for making an early diagnosis [3]. For the identification of different types of arthrogryposes, the presence or absence of normal neurological function in a child must be determined before making a differential diagnosis. An ordinary neurological exam shows that foetal crowding, generalized connective tissue disorders, amyloplasia, a distal arthrogryposis are the causes of arthrogryposis. However, a neurological defect suggests that the motor end plate, the central nervous system, the muscle, or the peripheral nervous system were aberrant in utero, which indicates diminished foetal mobility [4]. The prognosis of arthrogryposis is linked to the severity of the contractures, additional anomalies, and the degree of the contractures. Early physiotherapy and orthopaedic intervention are advised to help with joint mobility. In certain cases, the contractures can be treated postpartum and become less severe. While the prognosis depends on the underlying etiology, some newborns with arthrogryposis will have a positive outcome [5]. The anomaly known as a single umbilical artery (SUA) is characterized by the existence of one umbilical vein and the lack of one of the umbilical arteries, as opposed to the normal umbilical cord, which has one vein and two arteries (three-vessel cord) [6].

Case Report
A 24-year-old female was presented to the gynaecology outpatient department for a routine second-trimester checkup at 21 weeks. It was a non-consanguineous marriage and been a year since they were married. No known comorbidities, any blood transfusion history, or any allergies. Menstrual history is 4/28 days, normal flow cycle and it's been 11 years since menarche. The pregnancy history indicated that the woman was conceived spontaneously and that was confirmed by a Urinary pregnancy test (UPT) after being amenorrhoeic for 34 days. During the first trimester of the pregnancy dating scan was done which showed the SLIUF @ 8+5 weeks with an FHR of 171bpm, CL- 32mm, and the EDD was also given.

NT scan was also done where all the results were within the normal limits that is NT was 1.1mm. A double marker test was also done which showed no increased risk. The woman was on regular folic acid supplements. No history of bleeding/spotting PV, radiation exposure, fever with rash.

During the second trimester, quickening was not felt, and she was also on regular iron and calcium supplements. The first dose of TT was taken. A TIFFA scan was performed which showed a SLIUF @ 19+6 weeks, both forearms significantly were shortened with a single forearm bone, persistent hyperflexion of B/L wrist joints was seen, and finger counting was not done. Arthrogryposis, single umbilical artery, foetal heart - 4 chambered normal views LVOT/RVOT not delineated. A second TIFFA scan conducted at 20-21 weeks gestation revealed that in the foetal upper limbs, only a single bone was observed in the forearm. Fixed flexion and adduction of both hands were noted. Umbilical cord vessel with one artery and one vessel. Genetic counselling is done for the same. The patient was counselled regarding the anomalies and the patient, and her family decided on second-trimester MTP i/v/o foetal anomalies.

After the admission, all necessary investigations for MTP were performed. Abortion was induced with mechanical dilatation with intra-cervical Foley's followed by misoprostol. She expelled a dead female foetus of 300 grams. On gross examination of the foetus, anomalies like a short arm with hyperflexion of both wrist joints were noted. The placenta was not expelled within 30 minutes. The placenta was seen at the OS and hence removed manually. Under local anaesthesia dilatation and evacuation were done. On local examination umbilical cord was visible through OS, and 4×4 cm placental tissue was removed in pieces with ovum forceps.

Discussion

Aetiologically, AMC can be divided into extrinsic (extra-foetal) and intrinsic (foetal) causes. Intrauterine crowding brought on by congenital structural uterine abnormalities (such as the septate or bicornuate uterus), uterine tumours (such as fibroid), or multifetal pregnancy are examples of extra foetal causes of extrinsic aetiologies. These aetiologies are linked to favourable outcomes and deformation as opposed to disruption or malformation. Maternal illnesses like maternal immune myasthenia gravis or maternal exposure to infections, drugs, and social substances during pregnancy are examples of additional acquired causes. This acquired group of aetiologies is typically linked to a poor prognosis because they typically stem from disruptions of the central nervous system (CNS). Functional anomalies in the brain, spinal cord, peripheral nerves, neuromuscular junction, muscles, bones, tendons, joints, and restrictive dermopathies result in intrinsic or foetal aetiologies. Treatment for the foetal/intrinsic group is typically challenging, may result in developmental delays, and in rare circumstances, may be fatal [7]. Amyoplasia, a form of arthrogryposis is characterized by certain clinical features, such as internally rotated and adducted shoulders, extended elbows, flexed wrists, ulnarly deviated wrists, stiff fingers, and thumbs positioned in the palm. The lower limbs exhibit severe equinovarus contractures in the feet, knees extended, and possible dislocation of the hips. Midfacial haemangioma is common in patients. Most patients have normal intelligence. Congenital contractures of two or more separate body areas without a primary neurological or muscular disease characterize distal arthrogryposes which is another form of arthrogryposis. A consistent pattern of involvement of the hands and feet, limited involvement of the proximal joints, and variable expressivity are characteristics common to all distal arthrogryposes.[4] Current ultrasonography methods, which concentrate on reduced foetal movement and the identification of joint contractures or skeletal abnormalities on ultrasonography, are helpful in the prenatal diagnosis of arthrogryposis. Other sonographic markers, such as elevated nuchal translucency, may also be helpful; these are particularly relevant in cases where a syndrome is present and is associated with fatal Other common findings include polyhydramnios, foetal talipes, pleural effusions, micrognathia, ventriculomegaly, hydronephrosis, and collapsed stomach. AMC treatments include joint manipulation, serial casting, stretching exercises, aggressive physical therapy, and removable splints. Early aggressive management and rehabilitation with orthopaedic intervention can restore walking and daily activities [8].

Author contributions

All authors had active roles in the preparation of original manuscript and the treatment of the patient.

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Ethics approval

Institution Ethics committee approval was obtained.

Informed Consent

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List of Abbreviations

SUA-Single Umbilical Artery
UPT-Urinary Pregnancy Test
SLIUF-Single Live Intra Uterine Foetus
FHR-Foetal Heart Rate
EDD-Estimated Delivery Date
NT-Nuchal Translucency
PV-Per Vaginal
TIFFA-Targeted Imaging for Foetal Anomalies
B/L-Bilateral
LVOT-Left ventricular outflow tract
RVOT-Right ventricular outflow tract
MTP-Medical Termination of Pregnancy
AMC-Arthrogryposis Multiple Congenita
CNS-Central Nervous System
LBW-Low Birth Weight
IUGR-Intra Uterine Growth Retardation
IUFD- Intra Uterine Foetal Death

Conclusion

A single umbilical artery (SUA) is linked to a higher risk of intrauterine growth retardation (IUGR), which is also a potential cause of arthrogryposis. Detecting these anomalies prenatally is crucial for informed decision-making and effective management. Early intervention and specialist management can improve outcomes for affected individuals.

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