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Case Report

Rare Occurrence of Arthrogryposis Multiplex Congenita in a 26-Year-Old Pregnant Female: A Case Report and Review of Literature

Khurram Khaliq Bhinder[1], Azza Sarfraz*[2], Shaista Riaz[3], Fizza Zubair[4], Ameena[5], Zainab Malik[1], Afaf Arif[6], Zouina Sarfraz[7]

1. Shifa International Hospital, Islamabad, Pakistan
2. Aga Khan University, Karachi, Pakistan
3. McGill University, Montreal, Canada
4. Ameerudin Medical College, Lahore, Pakistan
5. Lahore General Hospital, Lahore, Pakistan
6. Ittefaq Hospital, Lahore, Pakistan
7. Fatima Jinnah Medical University, Lahore, Pakistan

ABSTRACT

Arthrogryposis Multiplex Congenita (AMC) refers to the development of multiple joint contractures affecting two or more areas of the body, prenatally. AMC is commonly termed as congenital multiple arthrogryposis, fibrous ankylosis of multiple joints, myodystrophia fetalis deformans, otto syndrome, and rossi syndrome. Due to the permanently fixed joint position, contractures may be associated, ultimately leading to muscular atrophy. Essentially, the severity of the disease is based on the number of joints involved. We report a case of a 26-year-old woman who visited a tertiary care hospital in Pakistan for routine antenatal scans. Pulmonary hypoplasia was noted with narrowing of the chest cavity. No movement with reduced muscle mass was seen in all the limbs. The hip joint was fixed and flexed in position whereas the knee joints were in an extended position. All presenting features were suggestive of AMC. Keeping in mind the patient's history, an infectious disease etiology and the concomitant use of category C drugs in addition to genetic non-syndromic associations appear to be the causative in negative gynecological outcomes.

Keywords: Arthrogryposis Multiplex Congenita; Otto syndrome; Congenital multiple arthrogryposis; Case report; Pakistan

1. INTRODUCTION

Arthrogryposis Multiplex Congenita (AMC) refers to the development of multiple joint contractures affecting two or more areas of the body before birth. AMC is also known as congenital multiple arthrogryposis, fibrous ankylosis of multiple joints, myodystrophia fetalis deformans, otto syndrome, and rossi syndrome. The disease affects the joint prenatally. A permanent position either fixed in bent or straight leads to contractures in affected joints, which commonly leads to muscular atrophy. The severity of this disease essentially depends on the number of joints involved. We report a case of a 26-year-old-woman who visited a tertiary care hospital in Pakistan for routine antenatal scans. On diagnostic radiology intervention, AMC was found as a causative factor for uneventful gynecological outcomes.

2. CASE PRESENTATION

A 26-year-old pregnant female patient visited a tertiary care hospital for routine antenatal scans. On obtaining informed consent from the patient, the case was proceeded for publication. She was gravida 2, para 1 (G2P1+0), and married for 3 years to her cousin, confirming consanguinity.The patient had one male child born through uneventful pregnancy via spontaneous vaginal delivery. The patient was laboratory tested for typhoid, which was positive. She had a medical history of utilizing the following medications, 1) piroxicam, 2) movax, and 3) bisleri. However, the patient was unaware of conception at that time of anti-typhoid medication administration. The patient had a family history of hydrocephalus as well.

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***Corresponding Author:** Dr. Azza Sarfraz, Research Associate, Pediatrics & Child Health, Aga Khan University, Karachi, Sindh, Pakistan. Email: azza.sarfraz@aku.edu.

An antenatal scan was conducted, which revealed a single live intrauterine pregnancy with the breech-extended presentation, with a longitudinal lie. Fetal cardiac activity was present. The placenta was fundal posterior, not encroaching the lower segment. Amniotic fluid was merely adequate. Mildly dilated lateral ventricles measuring up to 12mm were noted with no dilatation of the third ventricle. Pulmonary hypoplasia was noted with narrowing of the chest cavity. No movement with reduced muscle mass was seen in all the limbs. The hip joint was fixed and flexed in position whereas the knee joints were in an extended position. The features were suggestive of arthrogryposis multiplex congenita. Pertinent gynecological and neonatal findings for biparietal diameter, head circumference, femur length, fetal abdominal circumference, an estimate of fetal weight, gestational age, placental maturity grade, and fetal heart rate are enlisted in Table 1 and illustrated in Figure 1. Fetal bradycardia, documented at 60 beats per minute was observed for 3 minutes. Cardiotocography was advised for further evaluation. Finally, the patient was referred to the gynecology department, after which aseptic abortion was induced.

Finding	Value
Biparietal diameter	5.7 cm
Head circumference	23.9 cm
Femur length	4.5 cm
Fetal abdominal circumference	15.2 cm
Estimate of fetal weight	518 gm (+/-10%)
Gestational age	24 weeks

Table 1. A snapshot of pertinent gynecological findings.

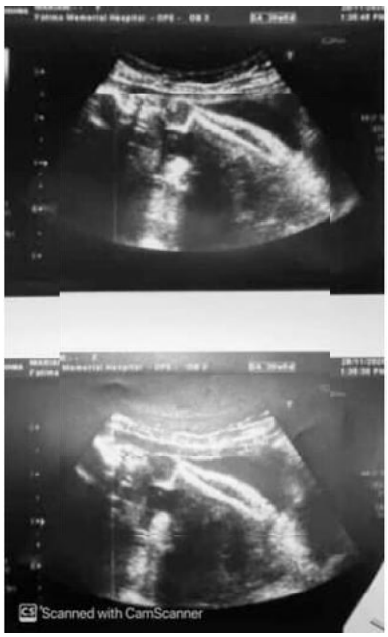

Figure 1. Antenatal scans of the patient.

3. DISCUSSION AND LITERATURE REVIEW

Arthrogryposis Multiplex Congenital (AMC) has a multifactorial etiology and pathogenesis. It is a non-progressive disorder characterized by multiple intra-articular contractures [1]. It affects about 1 out of every 3000 newborns [1]. It is reported mostly in African, European, and Asian countries, suggesting relevance in the Pakistani population [2]. About 350 genes with different phenotypic expressions are associated with AMC [2]. Besides genetic mutations, multiple environmental factors are causative. Other identifiable causes include infectious disease medications, chronic illnesses, active infection, traumas, abnormal uterine structure, and oligohydramnios [3]. Our case had an active history of typhoid fever in addition to consanguinity, further strengthening the hypothesis of infection and genetic factors as

causative. Fetal movement is imperative for the development of periarticular connective tissue and joints, particularly in the last trimester of pregnancy [4].

AMC may also be associated with multiple syndromes. However, non-syndromic associations are also common in around 10% of cases particularly those related to CNS anomalies such as Amyoplasia congenita, neuromuscular disorders, lissencephaly, corpus callosum agenesis, cerebellar vermis aplasia, fetal ventriculomegaly [4]. In our case, the patient had a family history of hydrocephalus. On the fetal scan, mildly dilated lateral ventricles measuring up to 12 mm were noted with no dilatation of the third ventricle. According to scientific literature, in many cases, abnormal nerve, muscle, and connective tissue development are involved. Hands, wrists, shoulders, hips, elbows, feet, knees, jaws, and back are typically affected [3]. In our case, documented features included pulmonary hypoplasia with narrowing of the chest cavity, no movement in all limbs, reduced muscle mass, fixed/flexed hip joint, and extended knee joints.

Medications, particularly those in Food and Drug Administration (FDA) category D and X are contraindicated in pregnant women, as they are teratogenic. However, some medications in category C are still underway, as sufficient human trials are missing. Their safety is not completely established. Both Piroxicam and Movax tablet falls in category C, wherein well-controlled studies have not been conducted in pregnant women. These medications require further conductions of human trials, with a note to recommendations in pregnancy [5]. Scientific literature establishes various teratogenic effects of both piroxicam and tizanidine [6]. It is contraindicated at 20 weeks gestation and later. It may otherwise cause prolonged labor and delivery, premature closure of fetal ductus arteriosus if used at 30 weeks of gestation or later; usage at 20 weeks or later may cause renal dysfunction of the fetus leading to oligohydramnios and even neonatal renal impairment [7].

FDA has added a new warning to the use of Nonsteroidal Anti-Inflammatory Drugs (NSAIDs) during pregnancy. The medication may lead to fetal kidney problems resulting in low amniotic fluid levels. Through 2017, the FDA received 35 reports concerning low amniotic fluid levels and kidney problems in pregnant mothers post-NSAID use. Of these, five newborns died; two had kidney failure due to confirmed low amniotic fluid while three had kidney failure without confirmed low amniotic fluid. Adverse events may commence with continued usage ranging from 48 hours to multiple weeks. There were 11 reports where amniotic fluid volume returned to normal post-NSAID discontinuation. Prolonged oligohydramnios may also cause delayed lung maturation and limb contractures [7]. As far as tizanidine is concerned, no human-based controlled trials targeting pregnant populations have been published. However, prenatal and post-natal developmental retardation was observed in rabbits and rats. At a dosage of 1mg/kg, the post implantation loss was also increased in rabbits [8].

4. CONCLUSION

While it is highly rare to document AMC during regular antenatal scans at the 24th week, it is essential to focus all available resources in developing countries like Pakistan to promote prompt diagnostic and interventional care. Keeping in mind the patient's history, an infectious disease etiology and the concomitant use of category C drugs in addition to genetic non-syndromic associations appear to be the causative in negative gynecological outcomes.

5. REFERENCES

1. Binkiewicz-Glinska A, Sobierajska-Rek A, Bakula S, Wierzba J, Drewek K, Kowalski IM, et al. Arthrogryposis in Infancy, Multidisciplinary Approach: Case Report. BMC Pediatr. 2013;13(1):184.
2. Arthrogryposis | Radiology Reference Article | Radiopaedia.org [Internet]. [Cited 2020 Dec 19]. Available from: https://radiopaedia.org/articles/arthrogryposis
3. Alves PVM, Zhao L, Patel PK, Bolognese AM. Arthrogryposis: Diagnosis and Therapeutic Planning for Patients Seeking Orthodontic Treatment or Orthognathic Surgery. J Craniofac Surg. 2007;18(4):838-43.
4. Bamshad M, Van Heest AE, Pleasure D. Arthrogryposis: A Review and Update. J Bone Joint Surg Am. 2009 Jul;91 Suppl 4:40-6.
5. Piroxicam - Side Effects, Uses, Dosage, Overdose, Pregnancy, Alcohol | RxWiki [Internet]. [Cited 2020 Dec 22].
6. Zanaflex (tizanidine) dosing, indications, interactions, adverse effects, and more [Internet]. [Cited 2020 Dec 22].
7. Piroxicam (Feldene) Use During Pregnancy [Internet]. Drugs.com. [Cited 2020 Dec 22].
8. Tizanidine (Zanaflex) Use During Pregnancy [Internet]. [Cited 2020 Dec 22].

6. CONFLICT OF INTEREST
All authors declare no conflicts of interest.

7. FUNDING ROLE
None to declare.

8. AUTHOR CONTRIBUTIONS
All authors contributed equally to the drafting of the study. Azza Sarfraz and Zouina Sarfraz are co-guarantors of the study.

9. ETHICAL STATEMENT
The patient signed an informed consent form, as per the ethical guidelines of the hospital board.