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Original Research Paper

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CASE REPORT: ARTHROGRYPOSIS MULTIPLEX CONGENITA

Kanika Chauhan	Department of Pediatrics, Civil Hospital Arki, Himachal Pradesh, India.
Sanket Vashist*	Department of Dermatology, Zonal Hospital Dharashala, Himachal Pradesh, India. *Corresponding Author
Aashutosh Sharma	Department of Dermatology, Zonal Hospital Dharashala, Himachal Pradesh, India.

KEYWORDS :

INTRODUCTION

Arthrogryposis multiplex congenita is a descriptive term used for congenital contractures in more than one joint. It may or may not be part of a syndrome. Any etiology that leads to decreased fetal movement may lead to congenital contractures. This condition is rather rare with an incidence of 1 in 2-3000 live births with both genders affected almost equally.

Arthrogryposis multiplex congenita is a rare sporadic non progressive congenital disorder that is characterised by multiple joint contractures and can incorporate muscle weakness and fibrosis.[1] It consists of several conditions of different etiology and varied clinical features, including multiple congenital contractures in multiple body joints. As proper fetal growth is dependent on fetal movement so any etiology decreasing fetal movement may lead to this condition. We present a case of a newborn who succumbed to this condition at birth.

CASE REPORT

A 23 yr old primigravida presented to the obstetrics and gynaecology department as a case referred in view of decreased fetal movements and fetal bradycardia. The mother was a booked case with uneventful antenatal period. No significant medical history. All the investigations were normal. Mother had been complaining of decreased fetal movements over the last fifteen days for which multiple biophysical profiles had been done, which were normal. On examination there was fetal bradycardia therefore the patient was taken for emergency caesarean section. Amniotic fluid was thickly stained with meconium and the newborn had no respiratory efforts, was centrally cyanosed and had a heart rate of less than 60 beats per min but to our surprise the baby was not limp, instead the baby had developed flexion contractures at neck, elbow, hip, knee joints symmetrically. Micrognathia was presemt. No other external congenital anomalies were noted. Resuscitation was started as per protocol but the baby could not be resuscitated.

DISCUSSION

Arthrogryposis multiplex congenita involves contractures of multiple joints present at birth where fat or fibrous tissue replace muscle tissue partially or completely. Arthrogryposis is usually symmetrical, but, sometimes joints may be involved to varied extents. All four extremeties are involved in 50-60% cases, lower limbs in 30-40% cases, and upper limbs in 10-15% of cases.[2] There are multiple etiologies which include neurological diseases (brain, spine or peripheral nerve), connective tissue defects (diastrophic dysplasia), muscle abnormalities (muscular dystrophies or mitochondrial diseases), space limitations (oligohydraminos, fibroids, uterine malformations or multiple pregnancy), intrauterine or fetal vascular compromise (impaired normal development of nerves, or anterior horn cell death), and maternal diseases (diabetes mellitus, multiple sclerosis, myasthenia gravis, infection, drugs, or trauma).[3,4]It could also be a part of a syndrome. Various syndromes associated with it are Mobius, Pierre-Robin, prune belly and Zellweger syndrome.

Arthrogryposis multiplex congenital is further divisible into two groups each of which has different causes, signs and symptoms. First group consists of conditions with a normal neurological function. These include amyoplasia, distal arthrogryposis syndrome, Multiple pterygium disorders, any systemic connective tissue disorder, fetal crowding. The second group consists of conditions where central or peripheral nervous system, motor end plate or muscle is abnormal.

This condition is diagnosed mostly in the second or third trimester with an ultrasound scan. An abnormal position and lack of mobility is noted in routine ultrasound. Mother may complain of reduced in utero fetal movements. The most common findings on ultrasound are fixed flexion deformities, micrognathia, altered amniotic fluid volume, limb deformities, cerebral ventriculomegaly, dysmorphic features and growth retardation. Other congenital anomalies may be associated, namely scoliosis, lung hypoplasia, growth retardation, mid-facial hemangioma, abdominal hernias, eye abnormalities, tracheoesophageal fistula and congenital heart defects[5]. The early the contractures occur, the harder early prenatal diagnosis will be.[6] Treatment is multidisciplinary. It may include splints or serial casting, passive stretching and physiotherapy. Surgical release of contractures is another option.

CONCLUSION

Once the diagnosis is suspected the family must be counselled regarding the condition, complications at birth and during labour. Also they should be informed about the future complications if the baby survives. Genetic counselling should be offered to parents to identify a genetic correlation so that recurrence can be anticipated. Arthrogryposis multiplex congenita can be a lethal condition, if associated with a fatal malformation, as in our case. Most of the times prognosis at birth is good.

REFERENCES

1. Kalampokas E, Kalampokas T, Sofoudis C, Deligeoroglou E, Botsis D. Diagnosing arythrogryposis multiplex congenita: a review. ISRN Obstet Gynecol.2012;264:9-18.
2. Margarita VF, Avila LC, Zamora EH, Kofman S, Bravo AH. Characterization of a group unrelated patients with arthrogryposis multiplex congenita. Jornal de Pediatria 2016;92(1):58-64.
3. Hall JG. Arthrogryposis multiplex congenita: etiology, genetics, classification, diagnostic approach, and general aspects. J Pediatr Orthop B. 1997;6(3):159-66.
4. Polizzi A, Huson SM, Vincent A. Teratogen update: maternal myasthenia gravis as a cause of arthrogryposis. Teratology,62: 332-41.
5. Brooks JG, Coster DJ. Arthrogryposis multiplex congenita : a report of two cases. Aust N Z J Ophthalmol. 1994;22(2):127-32.
6. Scott H, Hunter A, Bedard B. Non lethal arthrogryposis multiplex congenita presenting with cystic hygroma at 13 weeks gestational age. Prenat Diagn. 1999;19(10):966-71.