ORIGINAL RESEARCH PAPER

Medical Science

Volume - 7 | Issue - 4 | April-2017 | ISSN - 2249-555X | IF : 4.894 | IC Value : 79.96

Arthrogryposis Multiplex congenital : A Case Report

KEYWORDS

E-resources, first MBBS students, learning process, Internet.

Dr. SUNITA KHANDELWAL

MD(Pediatrics) Assistant Professor, New Medical College Hospital, Govt. Medical college, KOTA

ABSTRACT Arthrogryposis multiplex congenita is a term describing the presence of various arthrogenic contractures at the moment of birth, limiting, to a certain degree, the range of joints mobility. The incidence of the disease is low--1 per 3,000 births and it is usually not determined genetically. Here we are discussing a newborn baby presented in NICU of Govt. Medical College with multiple joint contractures.

INTRODUCTION

Arthrogryposis multiplex congenita is a rare congenital disorder with multiple joint contractures involving more than one area of the body. It is just a clinical finding rather than a specific diagnosis, associated with different disorders like neurocognitive delay and malformations1 . It is a nonprogressive disease. It occurs mainly due to fetal akinesia which may be because of multiple factors like neurogenic/myopathic process, a connective tissue disorder, intrauterine compression, a vascular insult / teratogenic exposure1 . Antenatal ultrasound examination can establish the correct diagnosis.

Case Presentation:

A newborn female admitted in NICU of new medical college hospital kota with multiple joints contracture. He was born with uneventful pregnancy with small for gestational age. His birth weight was 1.7 kg and otherwise healthy child. Mother's age was 28 years with history of nonconsanguinous marriage G1P1A0.Following posture was noted :adduction and internal rotation at shoulder, flexion at elbow and wrist joint. Contractures in fingers of both hands at metacarpophalengeal and proximal interphalangeal joints. Thumb was also abnormally placed. There was limitation of abduction of hip joints and bilateral CTEV. On x-ray there was equinovarus deformity.

Sacral area there was tuft of hair. it was spina bifida occulta on MRI scan. Abdominal and cranial USG were normal.

On follow up at the age of 2 months there was poor weight gain and delayed development.

DISCUSSION:

Arthrogryposis multiplex congenita is a rare congenital syndrome manifested clinically by wide spreaded contracture and deformities of multiple joints2 . It occurs 1 in 3000 live birth3,4. It is not a specific diagnosis but rather a description of clinical findings. It is associated with morethan 300 different disorders1 .

CLASSIFICATION

- Arthrogryposis multiplex due to muscular dystrophy.
- Arthrogryposis ectodermal dysplasia other anomalies, also known as Cote Adamopoulos Pantelakis syndrome, Trichooculodermovertebral syndrome, TODV syndrome and Alves syndrome.
- Arthrogryposis epileptic seizures migrational brain disorder.
- Arthrogryposis IUGR thoracic dystrophy,also known as Van Bervliet syndrome.
- Arthrogryposis like disorder, also known as Kuskokwim disease.
- Arthrogryposis-like hand anomaly and sensorineural deafness.
- Arthrogryposis multiplex congenita CNS calcification.
- Arthrogryposis multiplex congenita distal (AMCD), with a large number of synonyms such as Arthrogryposis multiplex congenita, distal, x-linked (AMCX1) and Arthrogryposis spinal muscular atrophy. Gordon Syndrome, also known as Distal Arthrogryposis, Type 2A.
- Arthrogryposis multiplex congenita, distal type 2B, also known as Freeman-Sheldon syndrome variant.
- Arthrogryposis multiplex congenita neurogenic type (AMCN). This particular type of AMC has been linked to the AMCN gene on locus 5q35. Arthrogryposis multiplex congenita pulmonary hypoplasia, also with a large number of synonyms.
- Arthrogryposis multiplex congenita whistling face, also known as Illum syndrome.
- Arthrogryposis multiplex congenita, distal type 1 (AMCD1).
- Arthrogryposis ophthalmoplegia retinopathy, also known as Oculomelic amyoplasia.
- Arthrogryposis renal dysfunction cholestasis syndrome, also known as ARC Syndrome4 .

Causes : The causative factors are multiple and can be classified as follows: i) Extrinsic: There is insufficient room in the uterus for normal movement. For example, fetal crowding; the mother may lack a normal amount of amniotic fluid or have an abnormally shaped uterus. ii) Intrinsic: muscles, Neurological - Central nervous system and spinal cord are malformed. In these cases, a wide range of other conditions usually accompanies arthrogryposis; Connective Tissue - Tendons, bones, joints or joint linings may develop abnormally. For example, tendons may not be connected to the proper place in a joint4 . Pathogenesis: the major cause for the arthrogyrposis is fetal akinesia due to multiple causative factors generalized fetal akinesia can slso lead to polyhydromnios, pulmonary hypoplasia, micrognathia, ocular hypertelorism and short umbilical cord. During early embyrogenesis, joint development is almost always normal. Motion is essential for the normal development of joints and their contiguous structures; lack of fetal movement causes development of the extra connective tissue to develop around the joint. These results in Contractures secondary to fetal akinesia are more severe in patients in whom the diagnosis is made early in pregnancy and in those who experience akinesia for longer periods of time during gestastion3 . Normaly the fetus movements can be made out as early as 7-8 wks5 . So prenataly the diagnosis of Arthrogyrposis is made as early as possible. Absence of fetal movement with severe flexion deformities of all the 4 limbs with associated poly hydromnios sometimes, which is a poor prognostic sign. Other associated findings are cleft palate, meningocele, congenital heart disease, klippel feil syndrome6 . Arthrogryposis is seen more frequently in mothers suffering from Insulin Independent Diabetis Mellitus7 . In Arthrogryposis with genetic defect there is increased nuchal translucency8 . Differential diagnosis include Trisomy 18 where there will be only involvement of upper limbs.

Volume - 7 | Issue - 4 | April-2017 | ISSN - 2249-555X | IF : 4.894 | IC Value : 79.96

REFERENCES

1. Rink, Britton D. Arthrogryposis: A Review and Approach to Penatal Diagnosis. Obstetrical & Gynecological Survey,2011:66(6);369-377.
2. David P.Gorezyca, John P.McGuhan, Karen K.Lindfors, William G.Ellis, Arthur Grix. Arhrogryposis Multiplex Congenita: Prenatal Ultrasonographic Diagnosis.
3. Arthrogryposis: a review and approch to prenatal diagnosis/DocGuide (http://www.docguide.com/ arthrogryposis-review-and-approch-prenataldiagnosis)
4. Arthrogryposis – Wikipedia, the free encyclopedia (http://en.wiki pedia.org/wiki/ Arthrogryposis)
5. Carol M Rumack, Stephanie R Wilson, J William Charboneau, Deorah Levine. The Fetal Musculoskeletal System by Phyllis Glanc, David Chitayat, and Sheila Unger- 4th Edition – Volume II/ 2005:1413-1415
6. Peter W Callen.The Fetal Musculoskeletal System by Luis F Goncalves, Juan Pedro Kusanovic, Francesca Gotsch, Jimmy Espinoza, Roberto Romero -5th Edition/2008: 469-470
7. Ibrahim A Alorainy, Nauman B Barlas, Amer A Al-Boukai. MULTIORGAN: Pictoral essay: Infants of diabetic mothers. Indian Journal of Radiology and Imaging.2010, 20(3): 174-181.
8. R Agarwal. OBSTETRICS: Prenatal diagnosis of chromosomal anomalies: Pictoral essay. Indian Journal of Radiology and Imaging.2003,13(2): 173-188