

Arthrogyryposis : A Case Report

B. Mallikarjunappa, K. M. Shruthi

Department of Radiodiagnosis, Sri Adichunchanagiri Institute of Medical Sciences,
B.G. Nagara, Nagamangala Taluk, Mandya District, Karnataka, India.

Abstract: Arthrogyryposis multiplex congenita is a rare congenital syndrome characterized by multiple joint contractures. It is mainly due to fetal akinesia. Incidence is 1 in 3,000 live births. It is associated with 300 different disorders.

INTRODUCTION

Arthrogyryposis 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 disorder, intrauterine compression, a vascular insult / teratogenic exposure¹. Antenatal ultrasound examination can establish the correct diagnosis. We report the characteristic sonographic feature and pathological correlation of fetus with AMC.

CASE REPORT

A 24 yr old women with G₃P₂L₁D₁ was referred for the first ultrasound scan at 32 wks of gestation. Her past medical history was uneventful. Her first pregnancy ended up in IUD at 8th month of gestation (reason not known). Now she has living normal second child. Her's is 2^o consanguineous marriage. Her husband was healthy. No familial h/o any congenital disorders. There was no h/o drug use.

Ultrasound examination of the present pregnancy at 32 wks gestation demonstrated a single, live intrauterine fetus corresponding to 32 wks. There was mild scalp edema with overriding of cranial sutures, cerebellar hypoplasia and microcephaly. Neck, Chest, Abdomen and Spine was normal. No normal limb movement noted. Bilateral fixed flexed deformity noted at elbow, wrist and interphalangeal joints. Lower limb were in extended with everted foot persistently. Based on these findings arthrogyryposis Multiplex congenita with cerebellar hypoplasia was made and the patient was advised for termination of pregnancy. [Fig-1]. MRI was done to this patient, extended lower limbs, flexed upper limbs and the cerebellar hypoplasia is noted. [Fig-2]

A male live fetus of birth weight 2400 gms was delivered. Baby didn't cry after birth and died within few minutes. Gross specimen showed the following features: Microcephaly with a space which admits only a tip of finger in anterior fontanelle, Left eye cornea looks opaque, 2 congenital teeth, undescended testis and micropenia. B/L flexion deformity at elbow joint, radial decussation, flexion deformity at 3rd, 4th and 5th proximal interphalangeal joints noted. B/L genu valgum and everted foot observed. [Fig-3]

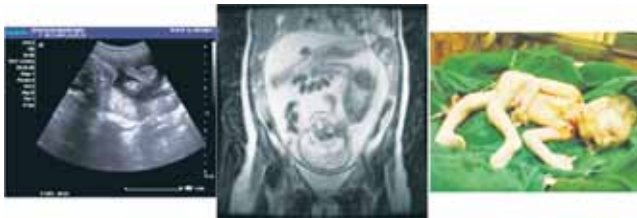


Fig - 1

Fig - 2

Fig - 3

DISCUSSION

Arthrogyryposis multiplex congenita is a rare congenital syndrome manifested clinically by wide spreaded contracture and deformities of multiple joints². It occurs 1 in 3000 live birth^{3,4}. It is not a specific diagnosis but rather a description of clinical findings. It is associated with more

than 300 different disorders¹.

CLASSIFICATION

- Arthrogyryposis multiplex due to muscular dystrophy.
- Arthrogyryposis ectodermal dysplasia other anomalies, also known as Cote Adamopoulos Pantelakis syndrome, Trichooculodermovertebral syndrome, TODV syndrome and Alves syndrome.
- Arthrogyryposis epileptic seizures migrational brain disorder.
- Arthrogyryposis IUGR thoracic dystrophy, also known as Van Bervliet syndrome.
- Arthrogyryposis like disorder, also known as Kuskokwim disease.
- Arthrogyryposis-like hand anomaly and sensorineural deafness.
- Arthrogyryposis multiplex congenita CNS calcification.
- Arthrogyryposis multiplex congenita distal (AMCD), with a large number of synonyms such as Arthrogyryposis multiplex congenita, distal, x-linked (AMCX1) and Arthrogyryposis spinal muscular atrophy.
- Gordon Syndrome, also known as Distal Arthrogyryposis, Type 2A.
- Arthrogyryposis multiplex congenita, distal type 2B, also known as Freeman-Sheldon syndrome variant.
- Arthrogyryposis multiplex congenita neurogenic type (AMCN). This particular type of AMC has been linked to the AMCN gene on locus 5q35. Arthrogyryposis multiplex congenita pulmonary hypoplasia, also with a large number of synonyms.
- Arthrogyryposis multiplex congenita whistling face, also known as Illium syndrome.
- Arthrogyryposis multiplex congenita, distal type 1 (AMCD1).
- Arthrogyryposis ophthalmoplegia retinopathy, also known as Oculomelic amyoplasia.
- Arthrogyryposis 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 arthrogyryposis; 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 arthrogyryposis is fetal akinesia due to multiple causative factors generalized fetal akinesia can also lead to polyhydramnios, pulmonary hypoplasia, micrognathia, ocular hypertelorism and short umbilical cord. During early embryogenesis, 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 gestation³.

Normally the fetus movements can be made out as early as 7-8 wks⁵. So prenatally the diagnosis of Arthrogyryposis is made as early as possible.

Correspondence: Dr. B. Mallikarjunappa, Department of Radiodiagnosis, Sri Adichunchanagiri Institute of Medical Sciences, B.G Nagara, Nagamangala taluk, Mandya district, Karnataka-571448, India. e-mail : drshruthikm@gmail.com

Absence of fetal movement with severe flexion deformities of all the 4 limbs with associated polyhydramnios sometimes, which is a poor prognostic sign. Other associated findings are cleft palate, meningocele, congenital heart disease, Klippel-Feil syndrome⁶. Arthrogyposis is seen more frequently in mothers suffering from Insulin Independent Diabetes Mellitus⁷. In Arthrogyposis with genetic defect there is increased nuchal translucency⁸.

Differential diagnosis include Trisomy 18 where there will be only involvement of upper limbs.

REFERENCES

1. Rink, Britton D. Arthrogyposis: A Review and Approach to Prenatal Diagnosis. *Obstetrical &*

Gynecological Survey, 2011;66(6):369-377.

- David P.Gorezyca, John P.McGuhan, Karen K.Lindfors, William G.Ellis, Arthur Grix. *Arthrogyposis Multiplex Congenita: Prenatal Ultrasonographic Diagnosis.*
- Arthrogyposis: a review and approach to prenatal diagnosis/DocGuide* (<http://www.docguide.com/arthrogyposis-review-and-approach-prenataldiagnosis>)
- Arthrogyposis* – Wikipedia, the free encyclopedia (<http://en.wikipedia.org/wiki/Arthrogyposis>)
- 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*
- 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*
- Ibrahim A Alorainy, Nauman B Barlas, Amer A Al-Boukai. *MULTIORGAN: Pictorial essay: Infants of diabetic mothers. Indian Journal of Radiology and Imaging*. 2010;20(3): 174-181.
- R Agarwal. *OBSTETRICS: Prenatal diagnosis of chromosomal anomalies: Pictorial essay. Indian Journal of Radiology and Imaging*. 2003;13(2): 173-188.

Sacroccocygeal Teratoma: A Case Report

B. Mallikarjunappa, B.T. Nagaraja

Department of Radiodiagnosis, Sri Adichunchanagiri Institute of Medical Sciences,
B.G. Nagara, Nagamangala Taluk, Mandya District, Karnataka, India.

Abstract: Sacroccocygeal Teratoma (SCT) is common congenital tumour that develop early in foetal life. Foetuses with this malformation are at risk for significant perinatal morbidity and mortality. This report demonstrates the role of foetal sonography in diagnosis of Sacroccocygeal Teratoma.

INTRODUCTION

Sacroccocygeal Teratoma (SCT) are relatively common congenital tumours that develop early in foetal life¹. It can be diagnosed by prenatal sonography in second or early third trimester of pregnancy². This report demonstrates the role of foetal sonography in the diagnosis of SCT.

CASE REPORT

A 28-year-old woman, gravida 1, was referred to our hospital at 30 weeks gestation. There was no family history of birth defects. The sonographic examination revealed a single intrauterine pregnancy with an estimated gestational age of 30 weeks. The study revealed a mixed echogenic mass arising from the sacroccocygeal region (figure 1). There were cystic areas within the mass. The spine appeared intact and the lower limbs appeared normal. The foetal kidneys and bladder appeared normal. There was no evidence of possible invasion of foetal pelvis and abdomen. Liquor was increased. Based on above findings, a diagnosis of external variety (type I) of SCT was made. The patient decided to continue the pregnancy and was scheduled for follow-up ultrasound 2 weeks later. She presented at 34 weeks gestation with rapid increase in the size of the uterus, premature rupture of membranes, and spontaneous labour pains. She delivered a dead foetus. At local examination, however, there was no evidence of foetal hydrops. Placental size was within normal limits.

DISCUSSION

Sacroccocygeal Teratoma is a common neoplasm with a reported incidence of 1-2 per 40,000 deliveries⁸. It contains derivatives of more than one of the three primary germ cell layers. Embryologically, SCT are thought to derive from multipotential cells in Hensen's nodes that migrate caudally and come to lie within the coccyx. SCT can be benign or malignant. Cystic lesions are more likely benign. Malignant SCT are extremely rare in foetus and uncommon in newborn infants. The likelihood of malignancy greatly increases in tumours diagnosed after the infant is 2 to 4 months old. Congenital anomalies may be present in association with SCT including genitourinary, anorectal and lower vertebral malformation and need to be ruled out during prenatal sonography^{2,3,5}. Large benign tumours are associated with significant morbidity and mortality. In such foetuses, complication results from massive intratumoral hemorrhage and dystocia^{6,7}.

Altman's classification: They are classified accordingly to the degree of exterior component or intrapelvic extension. Type I tumour (46.7%): Predominantly lie external to the foetus. Type II tumour (34.1%): Present externally but have

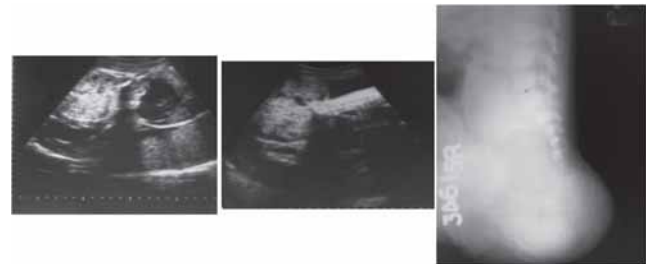


Fig 1: Longitudinal ultrasound scan of tumour showing solid and cystic components arising from caudal part of spine
Fig 2: Sagittal section scan of tumour showing solid and cystic components arising from caudal part of spine
Fig 3: X-Ray of Foetus Showing Soft Tissue Mass in Sacroccocygeal Region.

significant intrapelvic extension. Type III tumour (8.8%): They are apparent externally but predominantly lie within the pelvis and abdomen. Type IV tumour (9.8%): They are entirely presacral with no external presentation^{3,4}.

CONCLUSION

SCT are common congenital tumours that develop early in foetal life. Foetuses with this malformation are at risk for significant perinatal morbidity and mortality. The diagnostic technique of choice is ultrasonography. Early diagnosis influences clinical decision and management, which produces a better outcome.

REFERENCES

- Donnellan WA, Swenson O. *Benign and malignant sacroccocygeal teratomas. Surgery* 1968;26:316-318.
- Callen PW. *Ultrasonography in obstetrics and gynaecology. 3rd ed. Philadelphia: WB Saunders Company; 1994.*
- Izant RJ, Filston HC. *Sacroccocygeal teratomas, analysis of fortythree cases. Am J Surg* 1975;130:617-620.
- Teal LN, Anguaco TL, Jimenez JF, et al. *Fetal teratomas antenatal diagnosis and clinical management. J Clin Ultrasound* 1988;16:329-336.
- Hickey RC, Layton JM. *Sacroccocygeal teratoma. Cancer* 1954;7:1031-1043.
- Chisolm CA, Heider AL, Jeffery BS, et al. *Prenatal diagnosis and perinatal management of fetal sacroccocygeal teratoma. Am J Perinatol* 1998;15:503-505.
- Bonilia MF, Musoles F, Machado LE, et al. *Prenatal diagnosis of sacroccocygeal teratomas by two and three dimensional ultrasound. Ultrasound Obstet Gynecol* 2002;19:200-205.
- Taori KB, Khurana SD, Gyanchandani M, patil V. *Antenatal diagnosis of sacroccocygeal teratoma: Ultrasonographic diagnosis. Indian J Radiol Imaging* 2003;13:231-232

Correspondence: Dr. Mallikarjunappa B., Department of Radiodiagnosis, Adichunchanagiri Institute of Medical Sciences, B.G. Nagara, Nagamangala, Mandya District, Karnataka-571448, India.
e-mail: drnagarajbt@gmail.com