Early Prenatal Sonographic Diagnosis of Lethal Arthrogryposis Multiplex Congenita: A Case Presentation

Kazım GEZGİNÇ, Rengin KARATAYLI , Fatma YAZICI , Tuba TURSUN, Ali ACAR

Konya, Turkey

In this report, a 32 years old woman with increased nuchal translucency at 11th weeks of gestation and the diagnosis of Arthrogryposis Multiplex Congenita is demonstrated. After informing the family, chorion villus sampling (CVS) was performed. Fetal chromosomal analysis was reported as normal (XX). On control, lower and upper extremities had flexion contractures in 2D obstetrical ultrasonographic examination, besides bilateral club foot and absence of movement was detected. The family was informed and pregnancy was terminated. Lethal arthrogryposis, which is usually diagnosed by the demonstration of multiple joint contractures during the second or third trimester of pregnancy, may present as increased nuchal translucency thickness at early weeks of gestation.

Key Words: Arthrogryposis multiplex congenita, Prenatal diagnosis, Nuchal translucency

Gynecol Obstet Reprod Med 2012;18:86-88

Introduction

Arthrogryposis Multiplex Congenita (AMC) is a syndrome characterized by multiple joint contractures, decreased fetal movement, and absence of movement in one or more joint, flexion or extension contractures. It is thought to have myopathic or neurogenic origin or abnormality of connective tissue.[1,2]

The incidence of AMC is 1 in 3.000-10.000 births.[3,4] Fetal myopathy, neuropathy, connective tissue and skeletal abnormalities thought to play role in its etiology.[4,5] Less frequently, viral infections, autoantibodies against fetal acetylcholine receptors and maternal diseases like multiple sclerosis and myotonic dystrophy were defined as causes in the literature. The pathogenesis is not known exactly.[4]

Prenatal ultrasonographic diagnosis is frequently put by seeing restricted or absent fetal movement.[3] The ultrasonographic findings of AMC are usually seen in the 2nd and third trimester. Most of the cases are associated with pes equinovarous, elbow and wrist flexion deformities, flexion or extension deformities of the knee.[2] Rarely, malposition of extremities, increase of nuchal thickness and polyhydramnios can be seen as ultrasonographic findings.[3]

Selçuk University Meram Faculty of Medicine Department of Obstetrics and Gynecology, Konya

Address of Correspondence: *Rengin Karataylı*
Selçuk University Meram Medical School, Department of Obstetrics and Gynecology, Konya
renginkaratayli@hotmail.com

Submitted for Publication: *17. 08. 2011*
Accepted for Publication: *12. 12. 2011*

In this report, we present a case that had increased nuchal translucency in the 11th week and had the diagnosis of AMC ultrasonographically on the 18th week of gestation. For which the pregnancy was terminated.

Case Report

32 years old nulliparous woman was found to have increased nuchal translucency in the 11th. Week gestation, No other abnormality was seen in the obstetrical ultrasonographic examination during her routine control visits. After informing the family (Chorion Villus Sampling, CVS) was done for the patient. The patient came to our clinic for control. Lower and upper extremities had flexion contractures in 2D obstetrical ultrasonographic examination bilateral club foot (Figure 1) and absence of movement was seen.

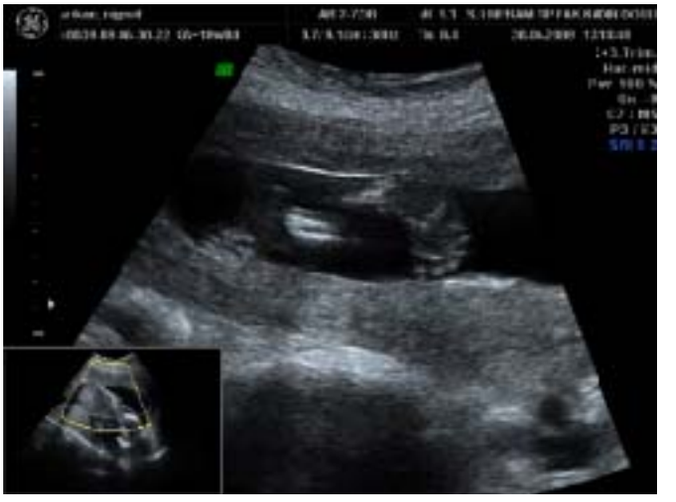

Figure 1: Bilateral club foot

According to her last menstrual period she was at 18 weeks of gestation. BPD (Biparietal Diamater) was consistent with 18 weeks and 5 days, AC (Abdominal Circumference) was 18 weeks and 1 day and FL (femur Lenght) was 19 weeks and 6 days. A 300 gram fetus was seen. Nuchal fold was measured as 6.8 mm (Figure 2)

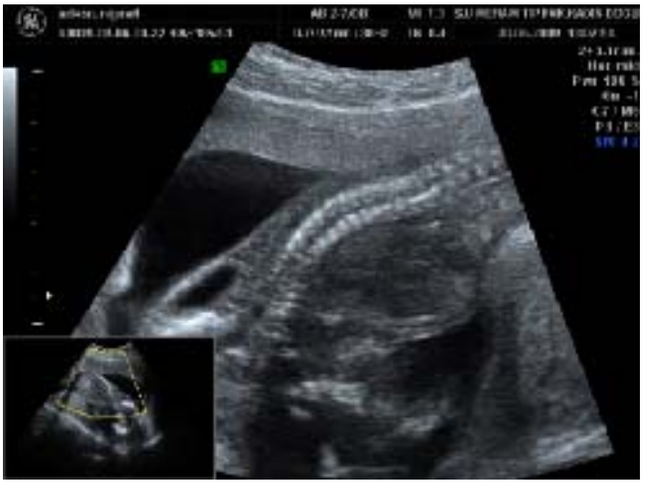

Figure 2: Nuchal fold was measured as 6.8 mm

There was no history of drug use or systemic disease in the mother. First degree of consanguinity was present between spouses. The family was informed and termination of the pregnancy was requested. The pregnancy was terminated by induction with oxytocin. The postmortem examination of fetus revealed 18-19 weeks female fetus, a significant limitation of movement of all joints, ulnar deviation of hands, bilateral pes equinovarus, increased nuchal translucency, oval ears, flattened nose and nose root were observed. In post-natal radiographic examinations revealed an evident extremity contractures (Figure 3) was noticed. Fetal chromosomal analysis was normal (XX). Maternal and paternal karyotype was normal. TORCH (toxoplasma gondii, rubella, cytomegalovirus, herpes simplex virus) screening revealed no acute infections.

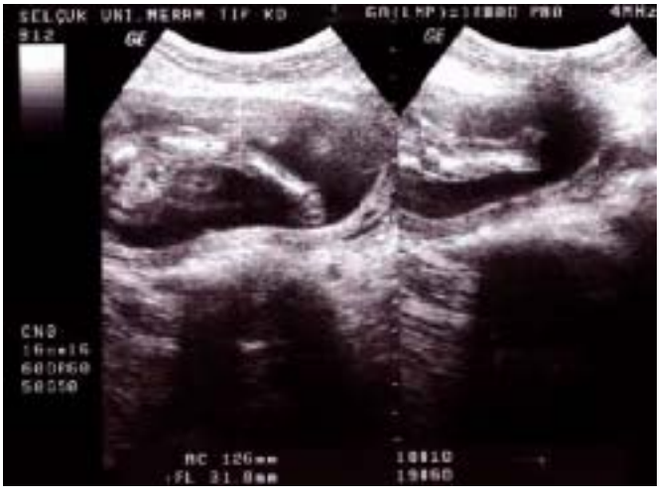

Figure 3: Extremity contractures

Discussion

Arthrogryposis Multiplex Congenita is defined as congenital non-progressive limitation of movement in two or more joints due to soft tissue contractures.[6] The etiology is multifactorial. The brain, spinal cord or peripheral nerve abnormalities involving neuropathic, myopathies, connective tissue diseases, narrowing of uterine cavity, maternal disease, intrauterine Vascular insufficiency are potential causes:[4,6] Many theories were introduced to explain the etiology. Last year Arthrogryposis was thought to develop as a result of joint movement limitations. Whatever the reason, lack of movement in uterus (fetal akinesia) may be associated with Arthrogryposis.[6]

It may be associated with complex of multisystemic congenital abnormalities or genetic diseases and chromosomal abnormalities. It may be seen related to single gene defects (autosomal dominant, autosomal recessive and X-linked recessive), chromosomal abnormalities and mitochondrial defects. Many genetic mutations have been identified to be related to Arthrogryposis Multiplex Congenita.[6] Hyett et al.[2] identified increased nuchal translucency in fetuses with chromosomal abnormalities.

In our case, no systemic diseases were present in parents, maternal, paternal and fetal chromosome analysis was normal. Although viral infections are rarely considered in etiology, TORCH panel was sent for the mother and it was normal. First-degree consanguinity of parents may indicate that genetic factors may be affecting factors.

Ultrasonographic findings of AMC are often seen in second and third trimester. The majority of cases may be associated with pes equinovarus, flexion deformities at the elbow and wrist, flexion or extension deformity of the knee.[2] Contractures due to immobilization of the joint are often explained by muscle weakness.[3] In our case, flexion contractures of the lower and upper extremities, limbs anomalies and bilateral club foot and absence of movement in extremities were noticed.

In addition to these, ultrasound findings of some cases may include: a short umbilical cord, limb malposition, increased nuchal thickness, polyhydramnios, pulmonary hypoplasia and micrognathia.[2,5] Often these findings can not be diagnosed until 16-18 weeks.[3,4]

Hyett et al.[3] emphasized the presence of limb contractures in all cases and that micrognathia is often associated with lethal AMC, and that diagnosis by these findings might not be confirmed till the 27th week of pregnancy. In our case, increase in nuchal translucency was detected in 11th week. In second trimester, when patients come for control, increase nuchal

translucency and contractures of the extremities and absence of movement were observed, and initial diagnosis of AMC was thought.

Cystic hygroma, subcutaneous edema, increased nuchal translucency are especially observed in lethal cases, they can be associated with decreased fetal movement and joint contractures, which may provide diagnosis in the first trimester.[2,3,4]

The diagnosis was delayed because when increased nuchal translucency was detected early in the patient, limb and joint abnormalities could not be detected and the patient was late in coming for her follow-up. Because nuchal translucency is associated with lethal cases, pregnancy was terminated on family request. Prenatal diagnosis of AMC with multiple joint contractures may be difficult to detect until the end of the second trimester, so increased nuchal translucency in 10-14th weeks should be kept in mind for AMC diagnosis.[2] Because of high risk of recurrence, patients should be screened in her subsequent pregnancy for fetal movements and limb position.[5]

Letal Artrogripozis Multipleks Kongenitanın Erken Prenatal Sonografik Teşhisi: Vaka Sunumu

Bu vaka sunumunda, gebeliğinin 11. Haftasında artmış fetal ense kalınlığı tespit edilen ve sonrasında intrauterin Artrogripozis Multipleks Kongenita tanısı alan 32 yaşında bayan hasta sunuldu. Aile bilgilendirildikten sonra, koryon villus örneklemesi yapıldı. Fetal kromozomal inceleme sonucu normal (XX) olarak tespit edildi. Kontrol 2D ultrasonografik incelemede, fetal alt ve üst ekstremitelerde fleksiyon kontraktürü, bilateral club foot ve hareketsizlik tespit edildi. Aileye bilgi verilerek, istekleri doğrultusunda gebelik sonlandırıldı. Genellikle ikinci veya üçüncü trimesterde multipl eklem kontraktürlerinin tespiti ile teşhis edilen letal artrogripozis, erken gebelik haftalarında artmış ense kalınlığı ile de belirti verebilir.

Anahtar Kelimeler: Artrogripozis multipleks kongenita, Prenatal tanı, Ense kalınlığı

References

1. Tıraş Ü, Çelikel B, Dallar Y, Tanyer G. Arthrogryposis Multiplex Kongenita. T Klin Jinekol Obst 1998;8:140-2.
2. Hyett J, Noble P, Sebire NJ, Snijders R, Nicolaides KH. Lethal congenital arthrogryposis presents with increased nuchal translucency at 10-14 weeks of gestation. Ultrasound Obstet Gynecol 1997;9:310-3.
3. Lin IW, Chueh HY, Chang SD, Cheng PJ. The application of three-dimensional ultrasonography in the prenatal diagnosis of arthrogryposis. Taiwan J Obstet Gynecol 2008; 47:75-8.
4. Madazli R, Tüysüz B, Aksoy F, Barbaros M, Uludağ S, Ocak V. Prenatal diagnosis of arthrogryposis multiplex congenita with increased nuchal translucency but without any underlying fetal neurogenic or myogenic pathology. Fetal Diagn Ther 2002;17:29-33.
5. Dane B, Dane C, Aksoy F, Cetin A, Yayla M. Arthrogryposis multiplex congenita: analysis of twelve cases. Clin Exp Obstet Gyneco. 2009;36:259-62.
6. Güneş F, Ekmekçioğlu Y, Özgüner A , Karadayı N, Baran A. Arthrogryposıs Multıplex Congenita. Kartal Eğitim ve Araştırma Klinikleri 1995;1:4-7.