Arthrogryposis Multiplex Congenita: Case Report

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Arthrogryposis multiplex congenita (AMC), characterized by multiple congenital joint contractures due to decreased fetal movements, is a non-progressive rare syndrome. Prevalence is determined to be 1 in 3000 deliveries. Although it is autosomal recessively inherited, sporadic cases have also been reported. Prenatal diagnosis of AMC is difficult. Here we report a case with a diagnosis of AMC not diagnosed during antenatal follow-up.

Key Words: Arthrogryposis, Multiplex, Congenita

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Introduction

Arthrogryposis multiplex congenita (AMC) is a rare syndrome characterized by joint contractures.[1] Contractures are in the form of ankylosis affecting two or more joints[2] and these contractures usually are not progressive.[1] Although studies have determined the incidence of this syndrome as 1:3000, due to a majority of the cases ending in spontaneous miscarriage or intrauterine mortality, actual incidence is not certain.[2,3] As AMC may be isolated, it can also be seen as part of broad syndromes with the central nervous system being affected.[1] The etiology of AMC contains factors such as functional or structural disorders of central or peripheral nervous system, muscular and connective tissue diseases, pharmaceuticals, toxins.[4,5] While there are some specific ultrasonographic findings during the first and second trimesters, a prenatal diagnosis via ultrasonography is very difficult in the third trimester.[6]

Here we report a case with a diagnosis of AMC not diagnosed during antenatal follow-up.

Case Report

A 31 year-old G4P3Y3 patient was admitted to the Delivery Unit with postmaturity prediagnosis. The general condition of the patient was fine and the vital findings were stabile during admission. Medical history showed three prior vaginal deliveries, no illnesses and pharmaceutical usage during pregnancies, spouse was a third degree relative. The prior antenatal examinations were noted as normal. In the ultrasonography, the fetal measurements were compatible with 41 weeks. The fetal weight was estimated as 4431gr. Upon detection of thick meconium in the amniotic fluid during labor, the patient was admitted to caesarean section with thick meconium and macrosomia indications. A 4000 gr. 52 cm male neonate was delivered with an Apgar score of 7/9.

When bilateral lower extremity anomaly was observed, neonatal examination was requested. The examination revealed caput quadratum, contracture in both knees, flexion contracture in left elbow, bilateral club-foot, hyperextension in the footprints and the neonate was admitted to the Neonate Unit with AMC diagnosis (Figure 1, 2). After these findings, an in-depth medical history revealed that the mother had an 11 year-old daughter who was being followed-up due to arthrogryposis multiplex diagnosis. It was found out that the patient had not mentioned the child who was being followed-up and treated with AMC diagnosis during her antenatal examinations.

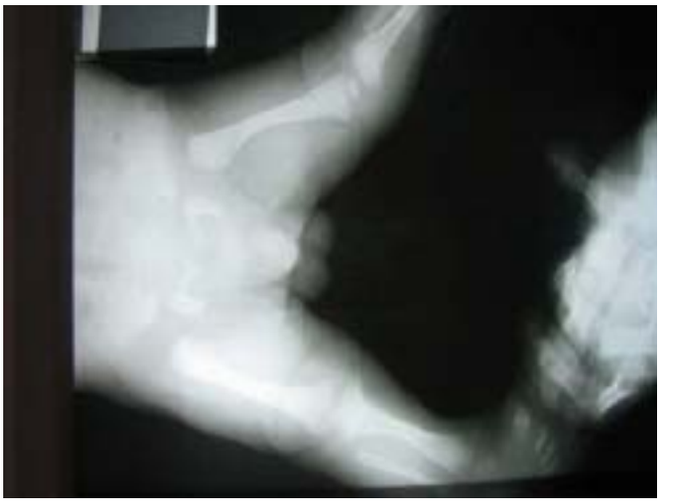

Figure 1: Normal femur view

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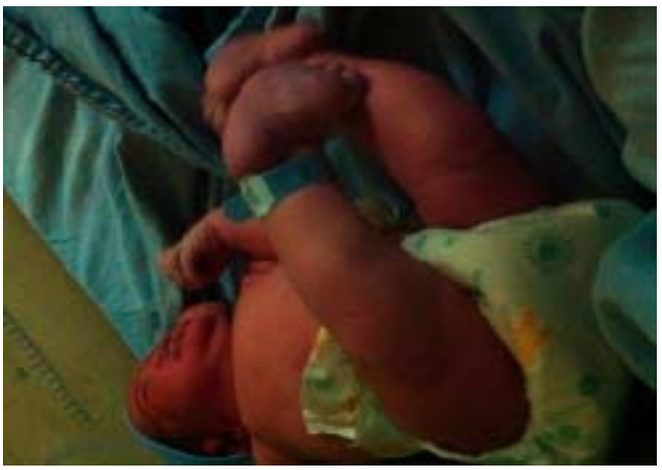

Figure 2: Bilateral club-foot, hyperextension at the knees

Discussion

AMC is a congenital disease characterized with multiple joint deformities.[7] The principle factor causing arthrogryposis is the decrease of fetal movements (fetal akinesia).[1] The fetal movements in the first trimester are very important for the development of the joints and limitation of these movements due to any reason can cause joint contractures.[8] Initially the joint development is normal, but due to reduced fetal movements connective tissue histogenesis around the joint increases and this results in joint contractures[9] While there are some specific ultrasonographic findings during the first and second trimesters, a prenatal diagnosis via ultrasonography is very difficult in the third trimester.[6] Most of the cases are diagnosed during delivery.[10]

During the antenatal examinations, patient's complaints about reduced fetal movements may provide a clue. Reduced extremity movements or posture,[10] increase in nuchal translucency and scoliosis[11] or accompanying congenital malformations, oligohydramnios can be detected with ultrasonography.[2] Better understanding of ultrasound findings and the etiology of this clinical situation provides an opportunity for careful prenatal assessment through image scanning that focuses on position and flexion/extension of proximal/distal joints, jaw and spine. The families should be informed/counseled for potential postnatal or post term evaluation in case of prenatal diagnosis.[12] In pregnancies with high risk of neuromuscular diseases, detailed ultrasonographic examination of the fetal movements and anatomy should be conducted in the early stages.

Artrogripozis Multiplex Konjenita: Olgu Sunumu

Artrogripozis multipleks konjenita (AMC) azalmış fetal hareketler nedeniyle multipl konjenital eklem kontraktürleri ile karakterize ilerleyici olmayan oldukça nadir görülen bir hastalık grubudur. Prevalansı 3.000 doğumda 1 olarak tespit edilmiştir. Otozomal resesif geçişli olabileceği gibi sporadik vakalar da gösterilmiştir. Prenatal tanısı oldukça güçtür. Bu yazıda prenatal tanı alamamış bir AMC olgusu sunulmuştur.

Anahtar Kelimeler: Artrogripozis, Multipleks, Konjenita

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