

Arthrogryposis Multiplex Congenital: Case Report

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Abstract

Arthrogryposis is a syndrome that can be, seen in different diseases that have in common the existence of multiple joint stiffness. Once the diagnosis poses, the prognosis will depend on the severity of the clinical presentation and its precocity of installation.

Management depends on the etiology, which makes the treatment different from one case to another.

In cases of suspected Arthrogryposis, practitioners should have attention to amniotic fluid quantity, fetal motricity, swallowing movements and research an amyotrophy or abnormalities of the extremities.

We report a particular case of antenatal diagnosis of Arthrogryposis with excess amniotic fluid.

Keywords: Arthrogryposis; Fetal

Introduction

Fetal movements in the amniotic fluid are a necessary condition for adequate fetal growth.

The absence or limitation of these movements can cause severe functional or morphological abnormalities for the fetus.

Myopathy, neuropathy, restrictive dermopathy, limited movement due to malformative pathology of the uterus or teratogenic treatment are generally the etiologies of fetal akinesia.

The following is a case of arthrogryposis recently diagnosed in our department.

Case Report

A 35 year old woman; gravida 1; para 1 with no past medical history. She is referred at 28 weeks for gestational diabetes, her main complaint being very restricted fetal movement.

During pregnancy, a triple test was performed with a low risk (Trisomy 13, 18, 21) and a morphological ultrasound did not found any abnormalities, the last consultation was more than 2 months ago.

The ultrasound examination showed, a pregnancy proportional to the term; an akinetic fetus in a transverse lie, with multiple contractures, excess amniotic fluid. Further analysis confirmed arthrogryposis with club feet, clenched fists, amyotrophy, facial dysmorphia (Figures 1 and 2), and pulmonary hypoplasia with absence of respiratory movement.

Spontaneous premature labor and a cesarian section were performed. The baby died a few minutes after birth.

At autopsy, similar facial abnormalities, contracture, the neck was short (Figure 1), pulmonary hypoplasia and severe lumbar scoliosis were also present. An ankylosis of the temporomandibular joint resulted in a trismus. The brain, spinal cord and muscle were unremarkable.

A karyotype was without abnormalities. No etiology founded for this akinesia.



Figure 1: Baby with short neck.



Figure 2: Facial dysmorphism.

Discussion

The prevalence of arthrogyrosis is estimated 1/3000 to 1/10000 births. Arthrogyrosis is not a specific diagnosis but a syndrome seen in different diseases that have in common the existence of multiple congenital contractures. The etiologies are multiple and can be maternal or fetal, neurogenic or myogenic and the prognosis depends essentially on the etiology.

In this case, the challenge for obstetricians is to evaluate the fetal and maternal prognosis, in order to offer the most accurate counselling

and to be able to formulate a therapeutic action plan. Multidisciplinary care including obstetrician, geneticist and neonatologist is always recommended.

Conclusion

The antenatal ultrasound diagnosis of arthrogyrosis is often possible when the examination is done by an expert. The finding of an oligohydramnion should indicate a more detailed ultrasonographic examination in search of other anomalies.

The ultrasound finding of fetal akinesia is always source of anxiety for the obstetrician.

Identifying the etiology of congenital contractures remains uncertain until today and is an important area of research for prenatal diagnosis and pediatric care.

Recent research in molecular genetics and immunohistochemistry seems to be useful in clarifying certain etiologies.

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