CASE REPORT

Successful pregnancy in a woman with arthrogryposis multiplex congenita

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SUMMARY

Arthrogryposis multiplex congenita refers to a clinical condition or syndrome characterised by multiple congenital contractures that affect two or more different areas of the body. Of the cases reported so far, an important percentage had to be terminated before pregnancy term, predominantly by caesarean section. We describe a 36 year-old woman who wanted to conceive. A multidisciplinary approach was set from the preconceptional period and special attention was given to respiratory function, potential anaesthetic difficulties and thromboembolic risks. She delivered by caesarean section at 38 weeks. This case emphasises the possibility of achieving a term delivery in these patients and points out the importance of a multidisciplinary team, specially of obstetricians and anaesthesiologists.

BACKGROUND

Arthrogryposis multiplex congenita (AMC) is a term used to describe multiple, non-progressive, congenital joint contractures that affect two or more different areas of the body.¹ The prevalence of this condition has been classically cited as 1/3000 live-births, but varies widely between case series, possibly due to different inclusion criteria.² AMC is not a diagnosis, but rather a clinical finding of a heterogeneous group of disorders, which seem to include more than 350 different conditions.³ The precise pathogenesis, although not completely understood, seems to involve decreased fetal movement (eg, fetal akinesia). The mechanism of decreased fetal movement depends on the underlying disorder, but may include neurological abnormalities, muscular abnormalities, abnormalities of the connective tissue, limitation of space within the uterus, intrauterine vascular compromise, maternal illness and exposure to specific drugs or medications.⁴ Depending on the clinical severity, patients may have highly functioning everyday life, with appropriate orthopaedic care and support and, precluding infertility from the underlying disorder, eventually become pregnant.⁵

CASE PRESENTATION

We describe a 36-year-old woman with the clinical diagnosis of AMC who was referred to our unit for her first pregnancy preconception counselling. There was no family history of AMC. She had been through 18 orthopaedic surgical procedures in order to try to correct several osteoarticular abnormalities in the lower limbs, hips and vertebral column and in spite of those maintained scoliosis and severe limitation of hip and thigh movements,

namely abduction. This woman presented specially shortened lower limbs and slightly shortened thorax and neck (she was 1.2 m tall). However, she was able to walk with crutches and had an independent life. No other conditions were present, like congenital cardiovascular anomalies or respiratory pathology (eg, asthma) and she was advised to quit smoking, which she did, by the end of the first trimester. The existence of pulmonary pathology seemed to us particularly important as at least one case report appears in literature reporting deterioration of maternal respiratory condition leading to iatrogenic preterm delivery by the 28th week of gestation.⁶ She was informed in preconception counselling about these risks and facts, but declared the wish of conceiving, though. Before planning the pregnancy she was advised to undergo respiratory function tests, results being normal. In the antenatal period this patient attended genetic counselling, careful ultrasound surveillance being indicated, as the initial diagnosis is made on ultrasound, typically in the second trimester when the fetus is noted to have fixed flexion contractures, a lack of mobility and an abnormal position.⁷ In addition, no family history of any form of AMC or anomalies like clubfoot, joint dislocation or contractures were present so fetal genetic invasive studies were deemed unnecessary; the genetics consultant's opinion was that an empiric risk of 3% of fetus being affected, although much has been discovered in recent years about the genetic basis of some forms of AMC.⁷

This woman conceived spontaneously and had her first consultation at our unit at 9 weeks and 3 days. During pregnancy she had normal results for routine laboratory tests, and special attention was given to morphological follow-up by ultrasound. Multiple scans were performed at each trimester, with special attention given to possible neural tube and skeletal abnormalities. Cervix length was regularly checked because of increased risk of preterm labour. All these parameters were found to be normal. Combined screening for aneuploidies in the first trimester demonstrated low risk for trisomy 21 and 13/18. By 18 weeks of gestation she began thromboprophylaxis with low-molecular-weight heparin once a day. She tolerated the entire pregnancy without respiratory difficulties and only by the 24th week had to stop her professional activity because of fatigue. Birth planning was postponed as much as possible to avoid the risks of prematurity, balancing against the maternal tolerance of the increasing uterus. At 23 weeks an anesthesiologist's evaluation was performed to define anaesthetic procedures for delivery. A general anaesthesia was decided in view of

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difficulties in performing locoregional techniques. Vaginal birth was considered inadvisable due to severe thigh abduction limitation and an unfavourable pelvis. She gave birth by caesarean section at 38 weeks of gestation, to a female newborn, weighing 2755 g and with an Apgar score of 7/9/10 (1', 5', 10'), under general anaesthesia. Postpartum period was uneventful.

The newborn had a normal physical examination at the time of birth, and is followed for the time being in our institution with no conditions detected so far (6 months of follow-up).

DISCUSSION

Information about obstetric care in patients with AMC is sparse. Only a few clinical cases have been described in the medical literature until now, and recommendations of obstetric management have been suggested by few, some of them ending in preterm caesarean deliveries, as the ones exposed by Duffy *et al*⁶ and Hardwick *et al*.⁸ Many women with this condition have few severe mobility limitations, have normal intellectual performance, and may expect an almost normal life, making childbearing a legitimate expectation.⁵

The main problems posed by this condition during pregnancy are limitations of respiratory function, which gives rise to an increased risk of preterm delivery for maternal intolerance, a potentially increased risk of thrombosis (depending on the patient's immobility) and anaesthesia-related technical issues. Besides, in some cases, social support may be a very important issue.⁶

Respiratory function was found to be normal in our patient. A normal spirometry was obtained before pregnancy, although literature does not point out this examination as strictly necessary. The authors found it important in order to try to access her respiratory condition, which could be severely impaired due to her short thorax and smoking habits. Nevertheless, only little discomfort was reported during gestation, which made her stop working after 24 weeks of pregnancy. She had no other respiratory symptoms until delivery. Respiratory difficulties are usually due to spinal deformities such as scoliosis, which may greatly decrease the available space within the abdomen for uterine enlargement leading to thoracic compression. Diaphragmatic function may also be affected earlier than expected in pregnancy.⁸ Cases in literature have reported the need for preterm delivery because of severe respiratory discomfort.^{6, 8} but this was not the case in our patient. In fact, the timing of delivery can be very difficult to decide when the fetal condition is normal but there is an increased risk of severe maternal conditions.⁶

Because of the higher risk of thromboembolic phenomena, a prophylactic dose of low-molecular-weight heparin was given, even though our patient had quite good mobility. The necessity of thromboprophylaxis in cases of almost normal mobility is not documented. Although one may find it unnecessary, we still chose to initiate this prophylaxis.

Anaesthetic procedures are challenging as well. Lumbar scoliosis in our *gravida* and the distorted anatomy in consequence of past surgical procedures made the use of locoregional procedures undesirable. In these patients, difficulties in intubation may also be expected, sometimes requiring alternative intubation techniques. This was not the case in our patient, though. General anaesthesia was given, and intubation was uneventful, as was the rest of the anaesthetic process.

AMC is usually diagnosed at birth, and only one in four cases may be diagnosed prenatally.⁹ When prenatal diagnosis is possible, it usually occurs in late second or early third trimesters.¹⁰ Early diagnosis, prenatal evaluation and further surveillance via

image scanning give the opportunity for family counselling concerning elevated neonatal morbidity and mortality in these cases and labour or delivery planning.¹¹ Prenatal findings contributing to diagnosis are mainly diminished fetal movements and joint contractures or skeletal deformities like *talipes*.¹⁰ These findings should guide the practitioner to a careful assessment of fetal anatomy and joints.¹¹ The most common detailed ultrasound scan findings are fixed flexion deformities, micrognathia, altered amniotic fluid volume, limb deformities, cerebral ventriculomegaly, dysmorphic features and growth retardation.⁷ In fact, clearly, the timely detection of fetal movement disorders should be improved, and AMC diagnosis should always be kept in mind. Regular scans were provided to our patient and none of these abnormal findings were detected during pregnancy. In fact, neonatal physical examination revealed no abnormalities as well.

Learning points

- ▶ A multidisciplinary team with obstetricians, anaesthesiologists and neonatologists is necessary for studying, managing and the timely planning of these pregnancies.
- ▶ Attention must be given to respiratory function, anaesthetic procedures, possibility of thrombotic events and preterm labour.
- ▶ Timely assessment of maternal pelvis is mandatory to decide if trial of labour may be attempted.
- ▶ Arthrogryposis multiplex congenita has genetic causes and attention must be given to the prenatal findings like diminished fetal movements, which may suggest further investigations.

Competing interests None.

Patient consent Obtained.

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