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SEVERE FETAL AKINESIA AT 19 WEEKS: A CASE REPORT

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INTRODUCTION

The term "fetal akinesia" is defined as a complete or partial absence of fetal movements. It can occur in isolation or be associated with other malformations, some of which can be lethal. The first observations of this syndrome were made by Péna and Shokeir in 1974 [1]. Since then, several cases have been reported, though the syndrome remains rare, with an estimated frequency between 1 in 3,000 and 1 in 15,000 births [2]. In some cases, the risk of recurrence can be as high as 25% [3]. In practice, the detection of fetal akinesia during an ultrasound examination is always a source of anxiety, accompanied by diagnostic, etiological, and therapeutic challenges.

OBSERVATION

We present the case of Mrs. X, a 23-year-old woman, G2P1, who was 19 weeks pregnant. She had no pathological history, including no infectious episodes or neurological disorders. The pregnancy was poorly monitored, with only an early ultrasound at 7 weeks confirming the pregnancy.

The clinical examination was normal, with a blood pressure of 11/7 and a uterine height consistent with the gestational age. The obstetric ultrasound revealed an evolving pregnancy appropriate for the gestational age but with significant diffuse subcutaneous edema, fetal immobility, claw-like hands, congenital heart disease (ventricular septal defect), and craniofacial dysmorphism. There was no ascites or pleural effusion. Fetal movements were absent at all times, even during long and repeated ultrasound examinations.

A termination of pregnancy was performed due to the severity of the fetal condition and the early onset of akinesia (at 19 weeks of gestation), with the family's consent. The expulsion resulted in a 400 g fetus presenting generalized edema, retrognathia, low-set ears, thin limbs, multiple joint stiffness, and camptodactyly. Unfortunately, an autopsy was not performed (Fig. 1).

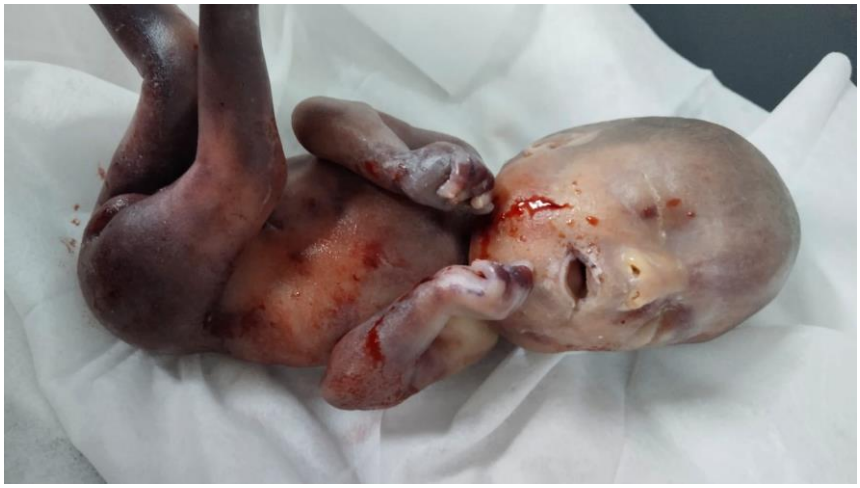

Figure 1 *(Photo of the newborn with foetal akinesia.)*

DISCUSSION

The phenotypic expression of fetal akinesia is not uniform; various clinical presentations can be observed. Major forms, often presenting early, are commonly referred to as multiple pterygium syndromes. This severe major form includes diffuse severe pterygia of the neck, axillary, and crural regions, a large cystic hygroma, facial dysmorphism with micrognathia, and pulmonary hypoplasia [4]. This form is subdivided into 20 clinical subtypes currently linked to at least 37 genetically defined entities [5]. This condition can be inherited in an autosomal recessive, autosomal dominant, or X-linked manner [6][7][8].

Genetic analysis of mutations in neuromuscular junction genes such as CHRNG and ECEL1 can reveal the pathogenic cause of the fetal akinesia deformation sequence and multiple pterygium syndrome. The information obtained is useful for genetic counseling and clinical management [9] [10].

Fetal akinesia can occur later, around 15-16 weeks, called the complete fetal akinesia sequence, associated with arthrogryposis with or without pterygium, facial dysmorphism with micrognathia, and even cleft palate, and pulmonary hypoplasia [11]. Minor forms may also be observed at a later stage and are characterized by hypotonia, limb malposition without arthrogryposis, and are often specific to primary muscular pathologies [12].

The etiologies of fetal akinesia are multiple, including maternal or fetal, neurogenic or myogenic causes. In some cases, a central neurological cause is identified, such as an anomaly of the posterior fossa (e.g., Arnold-Chiari malformation) or an ischemic, infectious, or metabolic origin affecting the sustentorial level.

In other cases, like congenital Steinert's myotonia, the condition may involve muscle. Antenatal diagnosis is possible through molecular biology, while it remains almost impossible for other myopathies like rod myopathy and myotubular myopathies [13].

Additionally, fetal akinesia can be of maternal origin, such as transmitted maternal myasthenia. In such cases, the prognosis is good, with spontaneous improvement occurring in a few days to several weeks postnatally [13].

Although a specific cause (such as an infection or prior neurological disorder) was not identified in this case, the observed anomalies could indicate a genetic disorder or systemic syndrome not detected before pregnancy. Severe fetal anomalies, such as craniofacial dysmorphism and camptodactyly, are often associated with rare genetic syndromes, although an autopsy was not performed to confirm this hypothesis.

Diagnosis of fetal akinesia can be made by prenatal ultrasound as early as the 12th week of gestation. Fetal magnetic resonance imaging can be a useful adjunct to evaluate findings related to the central nervous system [14]. It can reveal agenesis of the corpus callosum, lissencephaly, hydrocephalus, and spinal cord anomalies.

The prognosis for this syndrome depends on several factors, including the gestational age at which immobility is detected, its etiology, and its progression over time. It is better when maternal myasthenia is the cause and in the absence of cerebral involvement.

The continuation of the pregnancy is discussed in the case of minor forms or maternal conditions with a good prognosis (e.g., maternal myasthenia), and delivery is carried out under the best conditions. When immobility is detected early or of serious etiology, medical termination of pregnancy is proposed, and an autopsy along with placental examination should be performed.

The rarity of cases of fetal akinesia and the variability of associated anomalies underscore the need for further research to better understand underlying causes and improve diagnostic and therapeutic options. Genetic studies such as exome sequencing and in-depth analysis of genetic variants to identify the genetic causes of complex syndromic diseases offer opportunities for better genetic understanding, genetic counseling, and prenatal diagnosis [15].

CONCLUSION

Fetal akinesia is a rare symptom. It presents in various forms depending on the timing and severity of the condition. The etiologies are multiple, and the prognosis depends on the gestational age of onset and its progression. The study of this case highlights the importance of attentive prenatal monitoring and early assessment of fetal anomalies to allow for informed and appropriate management decisions regarding pregnancy.

Competing interests

The authors declare no competing interest.

Authors' contributions

Conception and design of study: Nadia Marouen ,Ben Farhat Imen, Aidi Hadhami ,Tarhouni KHouloud ,Maryam Dhouibi

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