

## Case Report

# Rare Case of Fetal Akinesia Deformation Sequence (FADS): Antenatal Diagnosis at 13<sup>th</sup> Weeks of Gestation

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## Abstract

**Background:** Fetal Akinesia Deformation Sequence (FADS) is a rare condition characterized with abnormal multiple joint contractures. The limitation of fetal movements which start in early pregnancy weeks especially at the 7<sup>th</sup> or 8<sup>th</sup> gestational weeks is the most important factor in etiology and diagnosis of the case. Such deformation causes fetal morbidity, mortality and economic burden in postnatal period and late period of child life occupied with various multiple surgeries. It may be lethal even in neonatal stage, few survive with rehabilitative support. It should be diagnosed antenatally with ultrasound by noticing reduced fetal movements and abnormal positioning.

**Case:** A 23 year old, 3<sup>rd</sup> gravida patient (G3P1A1L1) at 13 weeks of gestation, with history of consanguineous marriage, showed abnormal limb positioning in USG. Duty time given detailed USG showed fixed extension at both hip joints, fixed extension at left knee joint and fixed flexion at right knee joint with club feet. FADS were suspected and pregnancy was terminated in view of poor prognosis.

**Conclusion:** Detailed history taking and duty time given 11-14 weeks USG shall be done in all antenatal patients. FADS could be diagnosed even in early (13weeks) gestational age.

**Keywords:** Fetal Akinesia Deformation Sequence (FADS); Congenital muscular dystrophy; Amyoplasia; Otto syndrome; Arthrogryposis; Ultrasonographic antenatal screening

## Abbreviations

FADS: Fetal Akinesia Deformation Sequence; USG: Ultrasonographic Antenatal Screening

## Introduction

The etiology of Fetal Akinesia Deformation Sequence (FADS) is variable and not entirely understood but it is presumed to be multifactorial. In most cases, Fetal Akinesia Deformation Sequence (FADS) is not a genetic condition. However, in approximately 30% of cases, a genetic cause can be identified. Arthrogryposis or Fetal Akinesia Deformation Sequence (FADS) is a generic term used in connection with a very heterogeneous group of disorders with a common feature which includes multiple congenital joint contractures 1 in 3000 ratio [1]. FADS is a term used to describe multiple joint contractures affecting two or more (multiplex) joints which are present at or before birth [2,3]. Neonate may die of respiratory dysfunction, few faces mental retardation and limited limb movements with structural abnormality. Although many survive till adulthood but they need to go through multiple orthopaedic interventions and rehabilitative procedures. If such cases are diagnosed earlier in antenatal life, then sudden stress to the parents and future suffering of the fetus may be prevented and fetus may be treated or terminated according to the severity of the condition, or at least further future treatments plans could be made well in advance [2-4]. Fetal akinesia can also lead to polyhydramnios, pulmonary hyperplasia, micrognathia, ocular hypertelorism, and short umbilical cord. At the early stage of

embryogenesis, joint development is almost always normal and motion is essential for the normal development of joints and their contiguous structures. Absence or reduced fetal movement causes development of the extra connective tissue around the joint. This results in fixation of the joint, limited range of motion and development of the joint contractures. Prenatal ultrasound diagnosis is usually based on the detection of abnormal limb position, restrictive fetal movement with reduced or absent response to acoustic stimulation, growth restriction, polyhydramnios, and pulmonary hyperplasia. Low-set malformed ears, hypertelorism, short neck, cleft palate, scalp edema, thoracic deformities, camptodactyly, and micrognathia may also be found.

## Case Report

A 23 years old 3<sup>rd</sup> gravida patient (G3P1A1L1) with previous 1 normal delivery of male child and 1 spontaneous abortion at 8 weeks of pregnancy came to outpatient department for routine antenatal check-up for the first time at 13 weeks of gestation. Vitals were stable. On Ultrasonographic examination persistent bending and crossing of left lower limb was noticed. On further examination right club foot was noticed and no limb movements in any of the lower or upper limbs were seen over a period of time. Expert USG by Fetal Medicine Expert was carried out which showed right lower limb was in fixed extension position at both hip and knee joint while left lower limb was in fixed extension position at hip joints and fixed flexed position at knee joint (Figures 1,2 and 3). Upper limb movements were restricted. On detailed history taking, patient gave history of



**Figure 1:** Ultrasonography showing fixed flexion at elbow joint, fixed extension at hip and knee joint (Left), fixed extension at hip 7 fixed flexion at knee joint (right).



**Figure 3:** 4D USG with no fetal movement of joint contractures.



**Figure 2:** Gross examination picture of just delivered abortus.



**Figure 4:** X-ray of the affected abortus showing curving of bones.

consanguineous marriage. Possibility of Fetal Akinesia Deformation Sequence (FADS) was made. Termination of pregnancy in view of poor prognosis was carried out. 13 weeks old fetus was delivered. On gross examination fetus clearly showed stiffness of limbs and even on moving it externally limbs were very rigid (Figure 3). Placental bits were sent for DNA preservation. FISH test was done to rule out association of Trisomy 18 and associated chromosomal anomalies which turned out to be negative. X-rays were taken of the fetus on the day of delivery which showed curving of bones (Figure 4).

History: Consanguineous Marriage

USG: Restricted limb movements, fixed joints, club foot

Gross examination: Rigidity of limbs and stiffness of joints

X-ray: Curving of bones

FISH: Trisomy 18 ruled out

DNA preservation: Form placenta bits for any further study in future

## Discussion

In present case we found, FADS which are a developmental deformation of a fetus that is characterized with several joint contractures with unknown cause [2]. Although, the joint contractures are considered as the common USG findings for FADS/Arthrogryposis

cases. In our case these findings were determined as clinical findings rather than particular diagnosis [5]. Diagnosis of Fetal Akinesia is most important and crucial as  $\frac{3}{4}$  cases of Fetal Akinesia are missed or undiagnosed in early pregnancy resulting in fetal anomalies being diagnosed in later stage of pregnancy or at birth. So that care must be taken while examining in early ultrasound [5]. In most of the cases, while 3D/4D ultrasonographic findings are examined, one should must carefully observe immobilization of the fetus, vertical or changed abnormal position, cerebral anomaly, morphological changes in neurological evaluation, abnormal nuchal translucency, cystic hygroma, small chest, thin ribs, scoliosis, abnormal bone or plicomelia, curved bone contractures, cycloencephalopathy can be observed [3,5,6]. These kinds of findings obtained from the USG should and must be carefully evaluated. Comparison of FADS cases with other genetical disorders such like trisomy cases especially with trisomy 18 (Edwards' syndrome) is present in most of the cases which should be ruled out at first [3,7,8]. Fetal birth defect or Edwards' syndrome is the most abundant trisomy (trisomy 18) syndrome after Down syndrome that shows major abnormalities in such cases [7]. Hence, prenatal diagnosis must be done carefully to these patients. In our case the trisomy 18 and Fish was done which was negative. Trisomy is the first seen parameters in diagnostic point of view. Fetal movements can be seen ultrasonographically as early as 8 weeks of gestation [7,9,10]. Antenatal ultrasonography should be

given some time for evaluation of fetal movements especially in high risk cases, i.e., oligohydromnios, increased nuchal translucency or consanguineous marriages. When suspected a detailed scan should be done and movements should be observed over half an hour to an hour. This may help to diagnose FADS as early as possible. Early diagnosis of such cases can save the future morbidity of the child from the series of surgical events by future. Genetically birth defects are associated with most of such cases, hence must be evaluated with patience, diagnosed and ruled out at early stages of pregnancy.

## Conclusion

We present this case of its rare incidence and also to highlight the fact that detailed early anomaly scan is an integral part of antenatal follow up around 11-14 weeks of gestation, so that if any anomaly is detected, it is possible to terminate the pregnancy in due time. Ultrasound technique is such a medical gift in this kind of conditions where proper diagnosis can be done at early stage to save the quality of life of fetus from future serial surgical events. Due to associated conditions in Fetal Akinesia Deformation Sequence (FADS) with several neurological conditions, poor prognosis, termination should be considered when diagnosis is known before viability.

## Case report message

1. Thorough duly time given 11- 14 weeks ultrasonography may detect rare congenital anomalies early, which can help in medical management. That's why ultrasonography should be done very carefully with patience.
2. Every fetus should be observed for its range and frequency of movements, just the structural study is not justified all the time.
3. Genetical profile should be done to avoid such conditions.

## Disclosure for Informed Consent

Proper informed and written consent in local understandable language was taken from the patient and her husband (parent of the affected fetus) regarding publication of the same case.

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