

# Fetal Akinesia / Hypokinesia Deformation Sequence (FADS): Two and Three Dimensional Ultrasound Presentation

## Fetal Akinezi / Hipokinezi Deformasyon Sekansı (FADS): İki ve Üç Boyutlu Ultrason Bulguları

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**ABSTRACT** Fetal movements are essential for the normal fetal development. Any factor limiting fetal movements result in fetal akinesia/hypokinesia deformation sequence (FADS). Recent data suggest that the condition known as Pena- Shokeir syndrome in the past is actually a phenotype developed as a result of fetal akinesia/hypokinesia rather than a true syndrome, itself. We report a fetus with fetal akinesia deformation sequence diagnosed with second trimester scan and three-dimensional ultrasound. The condition being lethal, early recognition and appropriate counseling is important. Most of these cases can be recognized at early second trimester ultrasound. Three dimensional ultrasound is very helpful especially in diagnosis of facial and skeletal abnormalities and can be used in counseling the patients.

**Key Words:** Prenatal diagnosis; fetal movement; ultrasonography

**ÖZET** Normal fetal gelişim için fetal hareket şarttır. Fetal hareketi engelleyen herhangi bir neden fetal akinezi/hipokinezi deformasyonu sekansına (FADS) neden olabilir. Geçmişte Pena-Shokeir sendromu olarak bilinen sendromun gerçekte bir sendrom olmaktan ziyade, fetal akinezi/hipokineziye bağlı olarak gelişen bir fenotip olduğuna dair yeni bilgiler bulunmaktadır. Biz ikinci trimester taraması sırasında tespit edilen bir FADS olgusunu ve onun üç boyutlu ultrason görünümünü sunuyoruz. Bu, ölümcül bir durum olduğundan erken tanı ve danışmanlık önemlidir. Pek çok vaka ikinci trimesterin başında iki boyutlu ultrason ile teşhis edilebilir. Üç boyutlu ultrason yüz ve iskelet anomalilerinin tanısında faydalıdır ve hastalara danışmanlık verirken kullanılabilir.

**Anahtar Kelimeler:** Prenatal tanı; fetal hareket; ultrason

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Fetal in utero movements are essential for normal functional development. Fetal akinesia/hypokinesia, independent of the etiology, results in a specific phenotype, which is characterized limb contractures and dysmorphic faces. Intrauterine growth retardation, hydramnios, and pulmonary hypoplasia accompany this phenotype.<sup>1</sup> It was first described by Pena and Shokeir in 1974; as a early lethal disorder of neurogenic arthrogryposis, pulmonary hypoplasia and hypertelorism. Then, Hall suggested that Pena-Shokeir syndrome is a description of a phenotype, which is the consequence of reduced or absent fetal movements, rather than a specific syndrome, itself.<sup>2</sup> Today, a number of conditions variable in onset, presentation and severity are grouped under the name of fetal akinesia/ hypokinesia deformation sequence (FADS). Here we present a case of FADS and its

three dimensional (3D) presentation to remind and emphasize the use of 3D ultrasound in the diagnosis of fetal anomalies.

## CASE REPORT

The thirty year old, multipara woman first attended to our clinic at 20<sup>2/7</sup> gestational weeks for prenatal care. Both parents were healthy and unrelated and had no remarkable familial history. The patient had one spontaneous miscarriage and one healthy child, who was small for gestational age. In the current pregnancy she had a dating ultrasound at 10<sup>th</sup> gestational week, but no first trimester screening and serum biochemical tests. The ultrasound examination, performed for second trimester anomaly scan revealed a male fetus whose biparietal diameter, femur length, and abdominal circumference lagged two weeks beyond the expected. Amniotic fluid index was normal. The upper extremities were flexed at the elbows and wrists, hands looked like “clenched” (Figure 1). The lower extremities were also flexed at the knees and had “rocker bottom feet” and “pes equinovarus” deformity (Figure 2). The fetus had a flat profile and micrognathia. The mouth was closed. No fetal activity was observed during the examination over one hour. In the three-dimensional ultrasound examination flexion contractures of the extremities could be seen more precisely (Figure 2). Fetal echocardiography was normal.

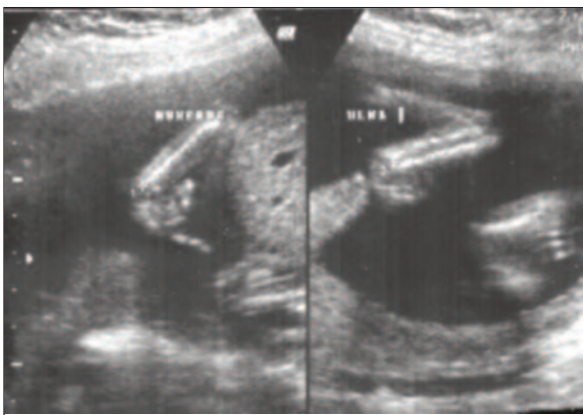


FIGURE 1: Flexion deformity of the hands.

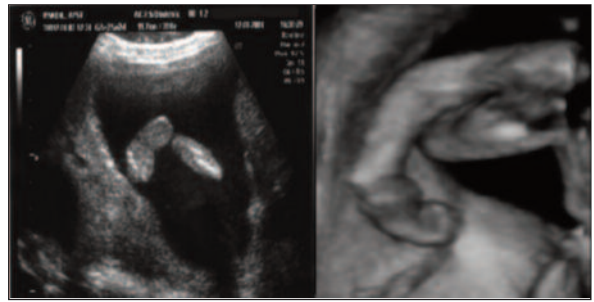


FIGURE 2: “Pes equinovarus” deformity and Flexion deformity of the knees in the 3D ultrasound.

Amniocentesis was performed for chromosomal study. The karyotype was 46,XY and the prenatal diagnosis was FADS.

This disorder considered being lethal, the pregnancy was terminated. The autopsy revealed a male fetus that had flexion contractures at the elbows, wrists, and knees (Figure 3). Both hands fifth finger was overriding the third and fourth finger. The fetus had “pes equinovarus” and “rocker bottom feet” anomaly. Horizontal palpebral fissures, epicanthal folds and hypertelorism, low set ears, micrognathia were noted during the examination of the face. In the pathologic examination; the lungs were hypoplastic and the brain revealed microcystic degeneration of the brain and microcalcification of the pituitary gland. The muscles and the skin were histologically normal.

## DISCUSSION

Normal fetal breathing, swallowing and facial muscular movements are required for the normal pulmonary development, amount of amniotic fluid, extremities and facial configuration respectively. Absence or reduction of these movements is responsible for pulmonary hypoplasia, hydramnios, limb contractures and facial abnormalities.<sup>3</sup>

Both the etiology and the phenotypic manifestations and the genetically transmission are heterogeneous. Any factor limiting fetal movement, be it of neurogenic, myogenic origin or a restrictive dermopathy may result in Pena-Shokeir phenotype, neurogenic causes can be primary cerebral malformations<sup>4-6</sup> or acquired fetal cerebral damage



**FIGURE 3:** Postabortal examination of the fetus confirmed the ultrasound findings.

secondary to hypoxic, ischemic damage.<sup>7,8</sup> Muscular dystrophies, peripheral neuropathy and rigid skin caused by a restrictive dermopathy are also reported to result in Pena- Shokeir phenotype.<sup>9</sup>

FADS is usually diagnosed in early second trimester. The onset is variable. The more severe cases have an earlier onset. The phenotype is a reflection of the severity of the disease. The disease can be full blown or regional.<sup>10</sup> The typical presentation of FADS phenotype is open mouth. In our case the fetal mouth was closed and amniotic index was normal. Depending on the region of the brain involved fetal swallowing might be spared. Trisomy 18 has many features in common with FADS. Karyotype study is required for differential diagnosis.<sup>11</sup> Multiple pterygium syndrome is also suggested lately to be a more severe form of FADS.<sup>12</sup>

The phenotype and the genetically transmission is also heterogeneous. Approximately half of

it is postulated to be sporadic, but also X- linked and autosomal recessive forms were reported.<sup>13</sup> If the etiology is a primary central nervous system malformation recurrence risk is 10-15 %. In case of hypoxic ischemic damage recurrence is 1%. The transmission varies depending on the type of muscular dystrophy.<sup>14</sup> The restrictive dermopathy being so rare actual genetical transmission was not reported.

Fetal akinesia hypokinesia sequence is almost uniformly lethal.<sup>15</sup> Both pulmonary hypoplasia and primary cerebral malformations contribute to the early lethality. Thus early prenatal diagnosis and appropriate counselling should be provided to the couples. The genetical transmission is variable. Recurrence risk should be discussed depending on the underlying specific etiology.

Diagnosis is usually by early second trimester scan.<sup>16</sup> The evaluation of the limbs and fetal face in the evaluation of dysmorphology is limited since two dimensional (2D) ultrasound is unable to provide images of the surface. The information obtained from the 2D ultrasound is dependent on the angle of insonation and imagination of the sonographer. Three dimensional ultrasound confirms the findings and has improved understanding of the relationships of the wrist, hand, fingers and thumb, and the leg, ankle and toes.<sup>17</sup> It should be emphasized that 3D ultrasonography is a complementary, but not necessarily an alternative to the conventional 2D technique in prenatal diagnosis. Parents are provided by clear photograph like images of the fetus and the sonographer can evaluate the extent of malformation at the different angles, giving a clear impression of the shape and severity of the defect to the parents.<sup>18</sup>

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