



### Case Report

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## Femoral hypoplasia- unusual facies syndrome: A case report and literature review

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### Abstract

The femoral hypoplasia- unusual facies syndrome (FH/ UFS); also known as femoral facial syndrome (FFS), is a rare condition characterized by a variable degree of unilateral or bilateral femoral hypoplasia associated with facial anomalies. The two principal characteristics of this syndrome, micrognathia and shortened femur, can be demonstrated by sonographic (US) imaging in the early stages of pregnancy. This disorder was first discovered by Daentt *et al* in 1975 and is more common in females. The femoral hypoplasia- unusual facies syndrome is a rarely considered clinical entity that has a strong association with maternal diabetes. This report describes a case of FH/ UFS followed in our Physical and Rehabilitation Medicine (PRM) department for proximal bilateral femoral agenesis. He had facial dysmorphism, bilateral testicular agenesis with a rare finding like bilateral second toe supraductus.

**Keywords:** Femoral hypoplasia- unusual facies syndrome, Femoral-facial syndrome, Micrognathia, Physical and Rehabilitation Medicine.

### INTRODUCTION

Femoral-facial syndrome (FFS) is an extremely rare fetal malformation of unknown etiology. It encompasses a spectrum of anomalies that include bilateral femoral hypoplasia and craniofacial dysmorphism with cleft palate, thin upper lip, micrognathia, flat philtrum, short nose with broad tip and upslanting palpebral fissure<sup>[1,2]</sup>. Other frequent findings include hypoplasia of fibulae, club foot, lumbar spine and pelvic anomalies. Systemic anomalies like cardiovascular and genitourinary anomalies may be associated with limb defects. The two principal characteristics of this syndrome, micrognathia and shortened femur, can be demonstrated by sonographic (US) imaging in the early stages of pregnancy. Despite this, it has rarely been reported in the first trimester<sup>[3,4]</sup>. This disorder was first discovered by Daentt *et al*<sup>[5]</sup> in 1975 and is more common in females<sup>[1]</sup>. In the majority of cases, it is sporadic, but an association has been established between this condition and insulin-dependent diabetes mellitus. The femoral hypoplasia- unusual facies syndrome is a rarely considered clinical entity that has a strong association with maternal diabetes<sup>[6]</sup>.

Etiopathogenesis of the syndrome remains unknown<sup>[7]</sup>. Majority of the cases described have been sporadic.

We describe here a case with FH/UFS followed in our Physical Medicine Department for proximal bilateral femoral agenesis.

### CASE REPORT

We present the case of M.S, a 5 year-old-child, referred for a waddling gait and a trunk deformation. He was issued from a consanguineous marriage. His mother was 45 years old. She has type 2 diabetes mellitus and she took oral antidiabetic agents. At the beginning of pregnancy, switch to insulin treatment was not performed. There was no history of taking any offending drug during pregnancy.

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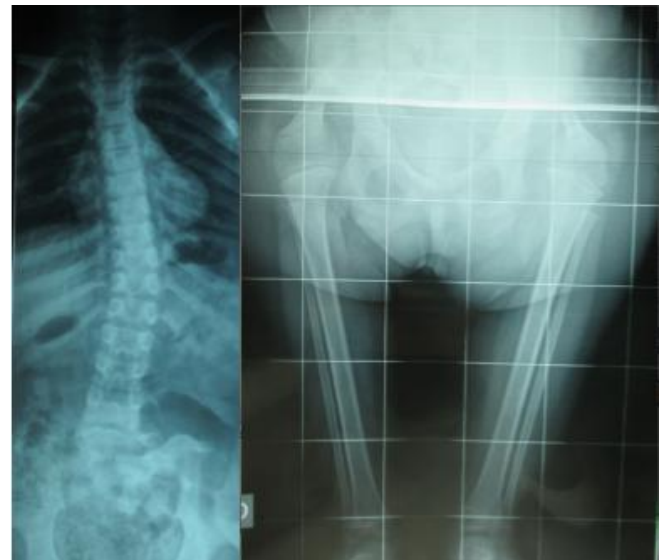
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Fifth-month ultrasound revealed bilateral femoral agenesis. Otherwise her pregnancy was uneventful up to term. Then she delivered a male baby by normal vaginal delivery. Baby cried immediately after birth.

The Psychomotor development was normal apart from a delay in walking around the age of 2 years. On physical examination, the height was 88 cm and the weight was 23 kg. Appearance was dysmorphic. Micrognathia, flat philtrum, small pinched nose and thin upper lip were present (figures 1,2,3). On spine examination, the occipital axis was diverted to the right. Left millimetric dorsal gibbosity and lumbar lordosis were found (figures 4,5). He also had a 1 cm leg length discrepancy in the right side. Moreover, the child had bilateral testicular agenesis confirmed by ultrasound and bilateral second toe supraductus (figures 6,7).

The radiological assessment concluded to an agenesis of the two femurs and the absence of scoliosis (figures 8,9).

The patient benefited from functional rehabilitation sessions while the surgery was not indicated.



Figures 8, 9: Spine and legs x-rays: scoliotic attitude and agenesis of the femurs

### DISCUSSION

The literature review has enumerated fifty cases previously described. This syndrome includes bilateral femoral hypoplasia, facial dysmorphism with cleft palate, micrognathia, long philtrum, thin upper lip with short broad tipped nose and dysplasia of hips. In addition to craniofacial and skeletal abnormalities other visceral abnormalities in cardiovascular and genitourinary systems may be found. Absence of the femur is a rare finding in FFS<sup>[7]</sup>. Maternal diabetes is known to have teratogenic effects. Malformation including neural tube defects, caudal dysgenesis, vertebral defects, congenital heart defects, femoral hypoplasia and renal anomalies are described in infants of diabetic mothers. However craniofacial anomalies have been rarely reported in such infants<sup>[8]</sup>. The caudal dysplasia syndrome and FH-UFS have been reported to be more frequent among infants of diabetic mothers<sup>[9]</sup>. Sironomelia or caudal dysgenesis is a syndrome in which there is insufficient mesoderm in the caudal most region of the embryo resulting in abnormalities of lower limbs and urogenital system but craniofacial anomalies are always absent. The presence of characteristic facial defect with cleft palate together with cardiovascular and genitourinary anomalies has sharply contrasted this case of FH- UFS with sironomelia. This syndrome may be attributed to fetal constraint, secondary to oligohydromnios<sup>[10]</sup>. Our case had bilateral femoral hypoplasia, micrognathia, upper thin lip, leg length discrepancy and bilateral testicular agenesis. With this framework the case fits into classical clinical spectrum of FH-UFS. The particular features are male sex, bilateral testicular agenesis and bilateral second toe supraductus.

Regarding the outcome there may be problems in speech development but many patients have been of normal intelligence and most of them have been ambulatory.

### CONCLUSION

Maternal diabetes is known to have teratogenic effects such as heart defects, kidney abnormalities but also femoral agenesis. The survival and intelligence of these patients is normal. Their main disability is physical, since the femoral hypoplasia can affect the gait of these patients. There are few cases reported in the literature leading to the originality of our case.

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Figures 1, 2, 3: Facial dysmorphism with micrognathia, flat philtrum, small pinched nose and thin upper lip



Figures 4, 5: Spine examination: Left millimetric dorsal gibbosity and lumbar lordosis



Figures 6, 7: Bilateral second toe supraductus.

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