International Journal of Science and Research Archive
eISSN: 2582-8185
Cross Ref DOI: 10.30574/ijsra
Journal homepage: https://ijsra.net/

(CASE REPORT)

FEMORAL-FACIAL SYNDROME IN A 4-YEAR-7-MONTH-OLD BOY BORN TO A MOTHER WITH DIABETES MELLITUS: A CASE REPORT FROM TANZANIA

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International Journal of Science and Research Archive, 2026, 21(02), 037–045

Publication history: Received on 16 September 2026; revised on 28 October 2026; accepted on 30 October 2026

Article DOI: https://doi.org/10.30574/ijsra.2026.21.2.1943

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ABSTRACT

Background: Femoral-facial syndrome (FFS), also known as femoral hypoplasia–unusual facies syndrome, is a rare congenital multiple-anomaly syndrome characterized principally by femoral hypoplasia or agenesis and a distinctive facial phenotype. The condition has a well-described association with maternal diabetes mellitus, although its precise pathogenesis remains uncertain.

Case presentation: We report a 4-year-7-month-old boy from Tanzania who presented with a markedly shortened right lower limb that had been present since birth. His mother had a history of diabetes mellitus during pregnancy. Clinical examination demonstrated complete right femoral agenesis with marked right lower-limb shortening, facial dysmorphism characterized by micrognathia and a long philtrum, cleft palate, umbilical hernia, and left inguinal hernia. Plain radiographs demonstrated absence of the right femur, with the right tibia and fibula articulating at the acetabular region. Abdominal ultrasonography demonstrated a solitary right kidney and absence of the left kidney, while echocardiography was normal. Genetic testing was not performed because of limited resources. The child experienced significant feeding difficulty associated with the cleft palate and had poor weight gain, with body weight remaining between 6 and 7 kg from approximately 1 year to 1 year 8 months of age. Following cleft palate repair, his feeding and weight gain improved. At the most recent assessment, his body weight was 13 kg. Cleft palate repair and subsequent inguinal hernia repair were

performed at Mnazi Mmoja Hospital, Zanzibar. Orthopaedic follow-up was arranged at Tumbi Regional Referral Hospital, with plans for orthoprosthetic management of the severe limb-length discrepancy.

Conclusion: This case illustrates the characteristic phenotype and multisystem involvement of femoral-facial syndrome in a child born to a mother with diabetes mellitus. The presence of femoral agenesis, characteristic facial features, cleft palate, abdominal wall defects, and unilateral renal agenesis highlights the importance of comprehensive evaluation for associated congenital anomalies. Early multidisciplinary management involving paediatrics, orthopaedics, orthotics and prosthetics, surgery services, and rehabilitation is important for optimizing functional and developmental outcomes.

Keywords: Femoral-facial syndrome; Femoral agenesis; Maternal diabetes mellitus; Cleft palate; Renal agenesis

1 INTRODUCTION

Femoral-facial syndrome (FFS), also known as femoral hypoplasia–unusual facies syndrome (FH-UFS), is a rare congenital anomaly syndrome characterized by variable degrees of femoral hypoplasia or agenesis accompanied by a distinctive facial phenotype [1]. The facial features classically include micrognathia, a short or broad nose, a long philtrum, a thin upper lip, and upward-slanting palpebral fissures [1, 2]. Cleft palate and other craniofacial abnormalities may also occur [3]. The femoral abnormality can range from mild shortening to severe hypoplasia or complete agenesis and may be unilateral or bilateral [4].

The syndrome was initially described in the 1970s and subsequently characterized through additional case series [5]. A strong association with maternal diabetes mellitus has been repeatedly reported. Johnson et al. described three affected children born to diabetic mothers, and their review of 36 previously reported cases found that 12 of the infants were born to mothers with diabetes [3]. More recently, Lacarrubba-Flores et al. reviewed 54 typical cases of FFS and found maternal diabetes in 50.8% of the pregnancies [6]. These findings show that maternal diabetes is commonly found among mothers of children with FFS. However, FFS can also occur in children whose mothers did not have diabetes, which means that maternal diabetes is an important risk factor, but it may not be the only factor for development of the syndrome.

Although maternal diabetes is an important epidemiological association, the precise mechanism underlying FFS remains uncertain. Most cases are sporadic, but familial cases have been described, including reports suggesting dominant inheritance [7]. Rare chromosomal abnormalities have also been reported, supporting the possibility of genetic susceptibility in at least some patients [8].

The clinical spectrum may include abnormalities of the vertebral column, pelvis, upper and lower limbs, genitourinary system, cardiovascular system, and other organs [9, 10]. The severity of these abnormalities can differ from one patient to another, and some affected children may have only a few associated abnormalities while others may have more extensive congenital defects [11]. Consequently, evaluation of a child with suspected FFS should not be limited to the musculoskeletal system but should include assessment for associated congenital abnormalities [10]. A careful physical examination together with appropriate imaging and other investigations can help to identify these abnormalities and provide a better understanding of the overall clinical condition of the child.

This paper presents a 4-year-7-month-old boy with severe unilateral femoral agenesis, characteristic facial dysmorphism, cleft palate, umbilical and inguinal hernias, and left renal agenesis with a solitary right kidney. The child was born to a mother with diabetes mellitus and was managed through a multidisciplinary approach in Tanzania.

2 CASE PRESENTATION

2.1 Patient Information

A 4-year-7-month-old boy presented to Tumbi Regional Referral Hospital, Tanzania, with a markedly shortened right lower limb that had been present since birth. The child had been living with the deformity without definitive orthopaedic treatment for several years. The reported reason for delayed presentation and treatment was limited socioeconomic resources.

There was no reported family history of similar congenital limb or facial abnormalities. The mother had a history of diabetes mellitus during pregnancy. Detailed records regarding the type of diabetes, glycaemic control, and antenatal glycated haemoglobin were not available.

The child was born at Tumbi Regional Referral Hospital. Further antenatal and perinatal details were not available in the records reviewed for this report.

2.2 Feeding and Nutritional History

The child had significant difficulty feeding because of the cleft palate. During infancy and early childhood, poor weight gain was observed. His body weight remained approximately 6–7 kg between the ages of 1 year and 1 year 8 months.

Following repair of the cleft palate, feeding improved and the child subsequently demonstrated progressive weight gain. At the most recent clinical assessment, at 4 years and 7 months of age, his body weight was 13 kg. This temporal improvement in weight gain following correction of the palatal defect suggests that feeding difficulty associated with the cleft palate contributed substantially to his earlier poor nutritional status.

2.3 CLINICAL FINDINGS

Physical examination demonstrated marked asymmetry of the lower limbs, with severe shortening of the right lower limb. The clinically measured limb-length discrepancy was greater than 8 cm compared with the left lower limb and significantly affected gait and mobility.

The right femur was clinically absent, and the child had a characteristic facial appearance. Facial examination demonstrated micrognathia and a long philtrum. A cleft palate was also present at the time of the initial assessment. Other congenital abnormalities included an umbilical hernia and a left inguinal hernia.

The child was reported to have motor delay related predominantly to the severe lower-limb deformity and associated gait impairment. Communication was otherwise considered appropriate for age. There was no history of previous fracture involving the affected limb.



Figure 1: Clinical photograph demonstrating marked shortening of the right lower limb compared with the left lower limb, umbilical and inguinal hernia



Figure 2: Clinical photograph demonstrating facial dysmorphism and micrognathia.

3 DIAGNOSTIC ASSESSMENT

3.1 Radiological Assessment

Plain radiographs of the pelvis, hips, and lower limbs were performed to assess the marked shortening of the right lower limb. The radiographs showed complete absence of the right femur. The right tibia and fibula were positioned in relation to the acetabular region, resulting in a marked shortening and abnormal alignment of the right lower limb. The left femur and the other visualized bones were present.

These radiological findings were consistent with severe femoral deficiency and explained the marked limb-length discrepancy and difficulty with walking observed clinically. When considered together with the characteristic facial features, cleft palate, renal anomaly, and other congenital abnormalities, the findings supported the clinical diagnosis of FFS. Genetic testing was not performed because it was not available within the resources of the treating setting.



Figure 3: Pelvic and lower-limb radiograph showing complete absence of the right femur and abnormal positioning of the right tibia and fibula.

3.2 Renal Assessment

Abdominal ultrasonography demonstrated a solitary right kidney with congenital absence of the left kidney, consistent with left renal agenesis.

3.3 Cardiac Assessment

Echocardiography did not demonstrate an apparent structural cardiac abnormality.

3.4 Genetic Assessment

Genetic testing was not performed because of limited diagnostic resources. The family history was non-contributory, with no known relatives affected by a similar congenital disorder.

4 DIFFERENTIAL DIAGNOSIS

The principal differential diagnosis considered in a child with severe femoral deficiency is caudal regression syndrome, particularly because both conditions have a recognized association with maternal diabetes mellitus. In this case, however, the presence of characteristic facial dysmorphism and cleft palate together with severe femoral agenesis supports femoral-facial syndrome. Caudal regression generally has a different pattern of caudal spinal, sacral, pelvic, and lower-limb involvement and does not characteristically produce the distinctive facial phenotype of FFS. The distinction can nevertheless be challenging in individual patients.

Other differential diagnoses include proximal femoral focal deficiency and other syndromes associated with femoral hypoplasia. These conditions may be considered when the femoral abnormality is less severe or when the characteristic facial features of FFS are absent or not clearly expressed. In this patient, the clinical and radiological findings, together with the associated congenital anomalies and maternal history of diabetes mellitus, supported the diagnosis of FFS.

5 THERAPEUTIC INTERVENTION

Management of the child was multidisciplinary and focused on the main clinical problems, including feeding difficulty, poor weight gain, the associated hernia, and severe right lower-limb deficiency.

Because the cleft palate caused significant feeding difficulty and contributed to poor weight gain, the child was referred for surgical management. Cleft palate repair was subsequently performed at Mnazi Mmoja Hospital, Zanzibar. However, the child subsequently underwent repair of the left inguinal hernia at Mnazi Mmoja Hospital, Zanzibar.

Following recovery from the surgical procedures, orthopaedic follow-up was arranged at Tumbi Regional Referral Hospital for management of the severe right lower-limb deficiency and associated gait impairment. No definitive reconstructive femoral surgery had been performed at the time of the report.

The family was explained about the treatment options and the problems the child may have because of the short right leg. Since surgery was not planned for the absent femur, the plan was to use an orthoprosthetic limb to help the child stand and walk better. Physiotherapy and orthotic assessment were also planned to help the child improve his movement.

6 FOLLOW-UP AND OUTCOMES

The patient demonstrated improvement in feeding following cleft palate repair. His body weight subsequently increased from the previously reported 6–7 kg during the period between 1 year and 1 year 8 months to 13 kg at the most recent assessment at 4 years and 7 months.

The inguinal hernia was repaired, while the umbilical hernia remained an associated congenital finding requiring continued clinical follow-up. No cardiac abnormality was identified on echocardiography. Renal imaging demonstrated left renal agenesis with a solitary right kidney.

The child continued to have a substantial right lower-limb length discrepancy due to complete absence of the right femur. This continued to affect his gait and mobility. He therefore remained under orthopaedic follow-up, with orthoprosthetic management planned to help improve his mobility. Long-term follow-up will be needed as he grows to monitor his limb function, mobility, and rehabilitation needs, and to see if further orthopaedic treatment or surgery will be needed.

7 DISCUSSION

Femoral-facial syndrome is a rare congenital multiple-anomaly disorder characterized principally by femoral hypoplasia or agenesis and a distinctive facial phenotype. The severity of femoral involvement is variable, and unilateral as well as bilateral abnormalities have been reported. The syndrome may also involve multiple organ systems, including the genitourinary, cardiovascular, vertebral, and craniofacial systems [9, 12].

The present patient demonstrated several findings that are characteristic of FFS. The most striking musculoskeletal abnormality was complete absence of the right femur, producing a severe right lower-limb length discrepancy and significant gait impairment. This was accompanied by characteristic facial dysmorphism, including micrognathia and a long philtrum, as well as cleft palate.

The combination of severe femoral deficiency and characteristic facial abnormalities is particularly important in distinguishing FFS from isolated proximal femoral focal deficiency. The facial phenotype is a defining component of the syndrome and provides an important diagnostic clue when severe femoral deficiency is identified.

7.1 Association with maternal diabetes

One of the most consistent observations in the literature is the association between FFS and maternal diabetes mellitus. Previous literature have also identified maternal diabetes in several individuals with the syndrome. Luisin et al. reviewed 92 cases of FFS and found that maternal diabetes was present in 42.4% of the patients [14]. Burn et al., in a series of patients

with bilateral femoral hypoplasia, identified cases associated with maternal diabetes as well as idiopathic cases [4]. The literature supports the strong but not exclusive relationship between maternal diabetes and the syndrome.

The maternal history of diabetes mellitus in the present case therefore supports the diagnosis. However, the absence of detailed records regarding the type of maternal diabetes and degree of glycaemic control limits conclusions regarding the precise relationship between maternal metabolic status and the child's anomalies.

The mechanism by which maternal diabetes may result in the characteristic pattern of abnormalities remains incompletely understood. Proposed mechanisms include disruption of embryonic development related to maternal hyperglycaemia and diabetic embryopathy [15, 16]. Nevertheless, the occurrence of FFS in children of non-diabetic mothers and the rare familial and chromosomal reports indicate that maternal diabetes cannot be considered the sole cause.

7.2 Craniofacial abnormalities

The facial phenotype that are well recognized in the published cases of FFS are micrognathia, long philtrum, a thin upper lip, a short or broad nose, and other craniofacial abnormalities [16]. Cleft palate has also been reported and can have important functional consequence in children with FFS. This includes feeding difficulties that contribute to poor weight gain during early childhood [17].

In the present case, the child had cleft palate and his weight was lower than expected for his age at the early stages of development. After surgical repair of cleft palate, his feeding improved and his weight also started to increase. This clinical course emphasizes that management of FFS should address functional consequences of associated anomalies rather than focusing exclusively on the limb abnormality.

7.3 Renal and other congenital abnormalities

Renal and genitourinary abnormalities have been reported in the phenotypic spectrum of FFS; in addition to skeletal and craniofacial abnormalities. In a case report and literature review, Ghali et al. reported that genitourinary abnormalities were among the associated manifestations of FFS and accounted for approximately 53% of cases in the literature reviewed [18].

The patient in this present case had left renal agenesis with a solitary right kidney. The renal finding in this patient reinforces the importance of screening children with FFS for associated visceral abnormalities. In addition to renal imaging, cardiovascular evaluation may be appropriate because cardiac abnormalities have also been described. In the present case, echocardiography was normal. Therefore, children suspected to have FFS should undergo appropriate evaluation for renal, genitourinary, and cardiac.

The patient also had both umbilical and left inguinal hernias. Although these findings are not the defining features of FFS, their presence contributes to the broader multisystem phenotype of this child and demonstrates why thorough physical examination of abnormalities is important.

7.4 Diagnostic considerations

Diagnosis of FFS is primarily clinical and radiological. Severe femoral hypoplasia or agenesis in association with characteristic facial features should prompt consideration of the diagnosis. Prenatal diagnosis is possible when severe femoral abnormalities and characteristic facial features are recognized on ultrasound, although expression is variable. Genetic testing can also be useful in the evaluation of patients with FFS because chromosomal abnormalities have occasionally been identified in individuals with an FFS phenotype. For example, a de novo complex chromosome 2q37 rearrangement has been reported in a patient with FFS, although no single causative genetic abnormality has been established as responsible for typical FFS [19, 20].

In resource-limited settings, diagnosis may occur late because of limited access to specialized imaging, genetic services, and multidisciplinary care. In this patient, socioeconomic limitations contributed to delayed presentation and treatment. Genetic testing was not available because of limited access to specialized genetic diagnostic services in the treating setting. This was a limitation because genetic testing could have helped to identify any chromosomal or other genetic abnormalities associated with the FFS phenotype to the child in the study case.

7.5 Management

Management of FFS should be individualized according to the severity of the skeletal deformity and the associated congenital abnormalities. Because the syndrome is multisystemic, multidisciplinary care is essential. For this child, treatment

appropriately addressed the cleft palate and inguinal hernia before definitive orthopaedic rehabilitation. The improvement in feeding and subsequent weight gain after palate repair demonstrates the importance of addressing functional problems that may affect nutrition and growth.

From an orthopaedic perspective, the severe limb-length discrepancy caused by complete femoral agenesis presents a major challenge. Management options depend on the anatomy of the affected limb, hip and knee function, overall health, family preference, available resources, and expected functional benefit. Orthotic and prosthetic management can provide a practical approach when complex reconstructive surgery is not feasible or is not preferred by the family. In the present case, the family opted for non-surgical orthoprosthetic management. Continued physiotherapy and orthotic assessment are therefore important to maximize mobility and independence.

7.6 STRENGTHS AND LIMITATIONS

A major strength of this case is the detailed description of a rare congenital syndrome with severe unilateral femoral agenesis and multiple associated abnormalities, including cleft palate, renal agenesis, and abdominal wall defects. The case also demonstrates the importance of multidisciplinary management and provides a longitudinal nutritional outcome following cleft palate repair.

The principal limitations are the absence of genetic testing, incomplete antenatal maternal diabetes records, and limited documentation of some perinatal parameters. In addition, long-term orthopaedic functional outcomes after orthoprosthetic fitting were not yet available at the time of this report.

8 CONCLUSION

Femoral-facial syndrome is a rare congenital disorder characterized by femoral hypoplasia or agenesis and characteristic facial dysmorphism. This case demonstrates severe unilateral femoral agenesis associated with micrognathia, long philtrum, cleft palate, umbilical and inguinal hernias, and left renal agenesis with a solitary right kidney.

The history of maternal diabetes mellitus is consistent with the well-established association between FFS and maternal diabetes, although it does not by itself establish causation. The case also demonstrates the importance of detailed evaluation for associated congenital abnormalities.

Early recognition and multidisciplinary management are essential. In this child, correction of the cleft palate was associated with improved feeding and subsequent weight gain, while surgical repair of the inguinal hernia addressed another important associated abnormality. Continued orthopaedic rehabilitation and orthoprosthetic management are planned to improve mobility and functional independence.

The key clinical lesson is that a child presenting with severe femoral deficiency and characteristic facial dysmorphism should be evaluated for femoral-facial syndrome and systematically screened for associated craniofacial, renal, cardiac, abdominal, spinal, and other congenital abnormalities.

LEARNING POINTS

- Femoral-facial syndrome should be considered in children with severe femoral hypoplasia or agenesis accompanied by characteristic facial dysmorphism.
- A history of maternal diabetes mellitus is an important clinical clue because of the well-established association between maternal diabetes and FFS.
- Patients with FFS require assessment for associated congenital abnormalities, particularly renal, cardiac, craniofacial, vertebral, and genitourinary abnormalities.
- Management should be multidisciplinary and individualized according to the severity of the skeletal abnormalities and associated conditions.
- Associated conditions such as cleft palate may have major effects on nutrition and development and should be addressed early.
- In resource-limited settings, orthotic and prosthetic rehabilitation may provide an important functional treatment option when complex reconstructive surgery is unavailable, unsuitable, or not preferred.

STATEMENTS ON COMPLIANCE WITH ETHICAL STANDARDS

Acknowledgments

The authors would like to thank the healthcare teams at Tumbi Regional Referral Hospital and Mnazi mmoja Hospital for their support in the care and management of this case. Also, the authors acknowledged the cooperation of parents of the patient, whose willingness to share information of their child made this case report possible.

Disclosure of conflict of interest

The authors declare that they have no conflict of interest related to this case report.

Statement of informed consent

Written informed consent for publication of the patient's clinical information and accompanying images, where applicable, was obtained from the patient's mother.

Author contributions

- **Tusekile Asajenye Solile:** Conceptualization, clinical assessment, orthopaedic management, manuscript preparation, and corresponding authorship.
- **Louis Ngowi:** Clinical assessment, orthopaedic input, and review of the manuscript.
- **Janet Cassian Njau:** Paediatric assessment, multidisciplinary management, and review of the manuscript.
- **Deogratias Mpunga:** Orthotic and rehabilitation assessment and review of the manuscript.
- **Adeline Mbeyale:** Orthotic assessment, rehabilitation planning, and review of the manuscript.

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