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Case Report **OPEN ACCESS**

A case of femoral-facial syndrome in a newborn of a Malay diabetic mother: Lessons for primary care

Nik Khadijah Nik Mustaffa Shapri [1], Juliawati Binti Muhammad [1*], Azlina Ishak [1], Shimilaaida Mohamad Ali Janaf [2], Mohd Haikal Bin Abdullah Zawavi [3], Jusoh Awang Senik [2]

[1] Department of Family Medicine, School of Medical Sciences, Universiti Sains Malaysia, Kelantan, MALAYSIA
[2] Pasir Mas Health Clinic, Kelantan, MALAYSIA
[3] Hospital Raja Perempuan Zainab II, Kelantan, MALAYSIA
***Corresponding Author:** juliawati@usm.my

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ABSTRACT

Femoral-facial syndrome (FFS) is a rare congenital disorder characterized by femoral hypoplasia and distinctive craniofacial anomalies, and it is frequently associated with maternal diabetes. We report the first documented Malaysian case of FFS in a neonate born to a 36-year-old Malay woman with gestational diabetes and obesity. Early antenatal ultrasound examinations did not detect any abnormalities. Fetal anomalies only became evident later in pregnancy when breech presentation and anhydramnios were noted, highlighting the difficulty of early prenatal diagnosis, particularly among high-risk mothers. A female infant was delivered via caesarean section at 35 weeks' gestation. She exhibited multiple dysmorphic features, including micrognathia, cleft palate, low-set ears, absence of the femur, hemivertebrae, and polydactyly. Despite multidisciplinary neonatal management, the infant died at two months of age due to severe nosocomial infection.

Keywords: femoral-facial syndrome, femoral hypoplasia-unusual facies syndrome, maternal diabetes, prenatal care

INTRODUCTION

Femoral-facial syndrome (FFS), previously termed femoral hypoplasia-unusual facies syndrome, was first described in the 1970s and remains a rare congenital disorder characterized by a distinctive combination of femoral hypoplasia and craniofacial dysmorphism. The disorder is rare globally, with estimated point prevalence of 1 in 1,000,000 and about 90 reported cases so far [1, 2].

There are no published statistics or case series describing the incidence or prevalence of FFS in Malaysia. Although the precise etiology is not fully established, nearly half of all reported cases are associated with maternal diabetes, suggesting a possible teratogenic influence [3].

FFS is a clinical syndrome with a variety of clinical features categorized as very frequent, relatively frequent and less frequent. Very frequent signs are present in 80-90% of patients. Examples are cleft palate, absent or abnormal femur, micrognathia/retrognathia, short limbs (micromelia, femur especially) and abnormal vertebral size or shape. Other characteristics that are relatively frequent (20-30%) are upwardly slanting eyelids, elongated philtrum, low set and poorly formed ears, fused bones of the spine and preaxial foot polydactyly [4]. Meanwhile the less frequent signs are strabismus, rib fusion, radioulnar synostosis, scoliosis, renal hypoplasia, ventriculomegaly and cerebral structural abnormalities that can be seen in imaging tests. Diagnosis is largely clinical and one of exclusion, requiring recognition of the typical phenotype and careful differentiation from other skeletal dysplasia. The diagnosis is made with the presence of femoral hypoplasia and two or more facial anomalies [5].

The increasing prevalence of obesity and diabetes in pregnancy worldwide, particularly in Malaysia [6], heightens the relevance of FFS. Poor glycemic control during early organogenesis has been strongly linked with congenital malformations, underscoring the importance of preconception care and optimized antenatal monitoring. Reporting new cases is therefore vital to enrich the literature, raise awareness among primary care providers, and strengthen the evidence base for prevention and early detection.

CASE DESCRIPTION

A 36-years-old woman, gravida 5 para 3 + 1 at 35 weeks was referred to tertiary hospital by the primary health clinic for a routine scar assessment and decision on mode of delivery. Antenatally, she had underlying obesity with body weight of 102.2 kg. She was diagnosed with gestational diabetes mellitus

This case has been sent for e-poster presentation at the 3rd Kelantan International Health Conference 2025.

Figure 1. Features of dysmorphism including low set and underdeveloped ears, elongated philtrum, thin upper lips, small bifrontal region, microphthalmia, micrognathia, wide spaced nipple, short and malformed lower limbs, extra toes at the right lower limb with structural CTEV (Source: Field study, reprinted with permission of the parents)

at early second trimester. Her biweekly blood sugar monitoring was under control. However, it was noted that she had poor weight gain for about 1.7 kg for the total duration of her pregnancy. Her ultrasonography scans throughout the pregnancy were reported as normal. She had one previous caesarean section. There is no family history of congenital birth defect, and she denied taking over-the-counter medications or supplements during early pregnancy. She was not in a consanguineous marriage.

During obstetric review at 35 weeks, it was noted that the fetus was in breech position with anhydramnios. However, it was difficult to visualize any congenital anomaly via ultrasound scan due to her thick abdomen. They proceeded with emergency LSCS for anhydramnios with breech presentation and a female baby was delivered vigorous, with good apgar score and birth weight of 2 kg.

Further examination of the baby noted multiple facial and body anomalies. The baby has low-set and underdeveloped ears, small bifrontal region, microphthalmia, micrognathia, cleft palate, short neck, widely spaced nipple, spina bifida, short bilateral lower limbs with extra toes on the right foot. Skeletal radiographs were performed showing evidence of hemivertebra in the lumbar region, absence of bilateral femurs and few other abnormalities (**Figure 1** and **Figure 2**). Chromosomal study sent reported as 46XX with no extra chromosome.

At 10 minutes of life, the baby developed respiratory distress requiring nasal prong 2 L/min and was admitted to neonatal ward. She was able to be discharged home on day 26 of life with comprehensive multidisciplinary team plan including growth and development assessment at nearest primary health clinic with orthopedic and plastic surgery outpatient review. MRI brain, vision and hearing test appointment was also arranged. Unfortunately, she was readmitted on day 30 of life due to acute respiratory distress syndrome. Despite intensive respiratory support and feeding assistance at neonatal intensive care unit, the baby finally succumbed to death at day 67 of life due to severe nosocomial infections with underlying multiple congenital anomalies.

DISCUSSION

FFS is a sporadic congenital disorder with no clearly established genetic cause, though maternal diabetes remains the strongest and most consistent association [1]. Hyperglycemia during early pregnancy has been shown to disrupt embryogenesis through mechanisms such as oxidative stress, excess production of free radicals, abnormal apoptosis, and impaired migration of neural crest cells [7]. These effects are particularly detrimental during the critical period of organogenesis in the first eight weeks of gestation, which explains why strict glycemic control prior to conception and during early pregnancy is crucial in preventing congenital malformations.

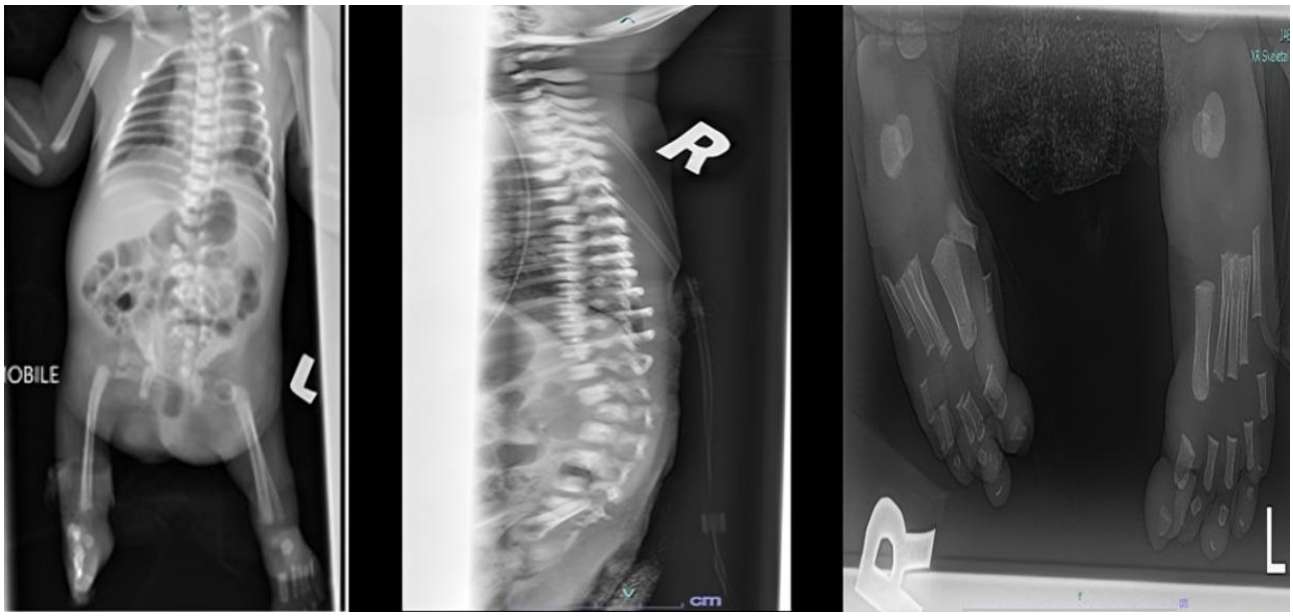

Figure 2. Skeletal survey showing absent right femur with overlapped right tibia and fibula, absent left femur with presence of left tibia and fibula, absence of 12th rib, hourglass pelvis shape, hemivertebrae of L1 and L5, extra metacarpal bone and phalanx of right foot (Source: Field study, reprinted with permission of the parents)

In our case, the patient came for booking at 14 weeks gestation which is quite later than what is recommended before 12 weeks of gestation. She has no known diabetes previously and has no pre pregnancy care done prior to her latest pregnancy. Besides that, maternal obesity might also be a risk factor for the congenital anomalies. It is found that maternal overweight/obesity in addition to gestational diabetes is associated with higher risk of birth defects [8]. Pre pregnancy care including early detection of diabetes or impaired fasting and post prandial blood sugar level, optimization of blood sugar level in diabetes and achieving optimal weight before embarking pregnancy is very important.

Gestational weight gain during pregnancy serves as an important indicator for identifying potential maternal and neonatal health issues. In our case highlighted above, we noted that the mother had poor weight gain throughout her pregnancy. According to National academy, in women with obesity, the recommended total and rate of weight gain are 5-9 kg and 0.17-0.27 kg per week, respectively [9]. Compared to pregnant mother with normal BMI, maternal obesity is usually associated with excessive weight gain. However, inadequate gestational weight gain was also associated with maternal-fetal complications such as higher risk of stillbirth, small for gestational age and preterm birth. Therefore, any inappropriate weight gain according to the gestational age should prompt a further scrutiny for possible causes such as foetal growth restrictions.

In the literature, although antenatal detection is possible, it accounts for fewer than 30% of cases, with the majority being identified postnatally [2]. On ultrasound, suspicion of FFS arises when there is shortened or absent femur(s) (often bowed, “telephone-receiver” shaped) together with facial anomalies such as micrognathia/retrognathia, sometimes with cleft lip/palate and growth restriction [5, 10]. In our case, the maternal follow-up was mostly handled at the primary health clinics and the condition was likely overlooked during prenatal care visits as there was no specialized detail ultrasound scan performed. This is because not all mothers with gestational diabetes routinely referred to anomaly scans. The Malaysian obstetric handbook states that mid-trimester anomaly scans are recommended for mothers who have high HbA1c or pre-existing diabetes [11]. Therefore, it is critical that primary care doctors possess strong prenatal ultrasound abilities to identify severe congenital abnormalities in diabetic mothers who are not candidates for detailed scans. This will require standardized training and continuous competency assessment.

The prognosis of infants with FFS is generally variable, though predominantly determined by functional limitations, as intellectual and psychomotor development is typically preserved. Although some children survive into childhood and undergo repeated orthopedic and reconstructive procedures, a significant proportion die during the neonatal period from severe prematurity, respiratory compromise, or sepsis [1, 2, 5, 12]. In this case, our patient succumbed to a severe nosocomial infection at two months of age. Survivors typically require multidisciplinary care involving neonatologists, orthopedic surgeons, plastic surgeons, audiologists, speech and language therapists, and developmental specialist [4]. The neonatal period may be complicated with respiratory distress due to upper airway obstructions, nutrition and feeding difficulties. Recurrent urinary tract infections have also been observed [10]. Coordination of this care is often best achieved through a family physician or primary care team who can provide continuity, anticipate complications, and support the family holistically.

Finally, ethical and psychosocial aspects must not be overlooked. Early identification of severe anomalies raises complex issues regarding parental counselling, decision-making, and the option of pregnancy termination in jurisdictions where this is legally permitted. Families may also require psychological support to cope with bereavement or the challenges of raising a child with complex needs [13] . Although genetic testing was normal in our case, genetic counselling should still be offered to help parents understand recurrence risks and provide reassurance for future pregnancies.

This case adds to the limited global literature on FFS and represents, to our knowledge, the first reported case in Malaysia. It underscores the need for vigilant preconception and antenatal care for women with diabetes and obesity, greater emphasis on early anomaly detection in primary care, and the importance of multidisciplinary and family-centered approaches in managing affected infants.

CONCLUSION

This case, the first reported from Malaysia, highlights the importance of meticulous preconception optimization of glycemic control and maternal weight to reduce the risk of malformations during critical periods of organogenesis. Inadequate gestational weight gain, as observed here, should also prompt clinicians to consider possible maternal and foetal complications.

The challenges in prenatal detection of FFS, particularly in obese mothers or those with limited access to detailed anomaly scans, underscore the need to strengthen ultrasound competencies among primary care doctors and ensure equitable access to advanced imaging. Survivors of FFS face complex, multisystem challenges that require coordinated multidisciplinary input, with family physicians playing a pivotal role in providing continuity of care and holistic support for affected families.

Author contributions: NKNMS: conceptualization; **JBM:** supervision; **AI:** writing–review & editing; & **SMAJ, MHBAZ, & JAS:** methodology. All authors agreed with the results and conclusions.

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Ethical statement: The authors stated that the study does not require any ethical approval since it is a single anonymized case report with no experimental intervention, in accordance with institutional and national guidelines. Written informed consent was obtained from the patient’s parents for publication of clinical details and images. All identifying information has been removed, and patient confidentiality was strictly maintained. Access to clinical data was restricted to the treating team and stored in secure, password-protected institutional systems.

AI statement: The authors stated that generative AI tools were used only for language refinement and grammar correction during manuscript preparation. The authors reviewed, edited, and take full responsibility for the scientific content, interpretation, and accuracy of the manuscript. No AI tool was used for data analysis, clinical decision-making, or image generation.

Declaration of interest: No conflict of interest is declared by the authors.

Data sharing statement: Data supporting the findings and conclusions are available upon request from the corresponding author.

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