Case Report

Klippel–Feil Syndrome: A Rare Case Report

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Abstract

Background: Klippel–Feil syndrome (KFS) is defined as a congenital fusion of two or more cervical vertebrae. The most common signs are short neck, low hairline at the back of the head, and restricted mobility of neck.

Case description: A term, female child weighing 2,522 g was born to a 34-year-old, a third gravida mother with a previous history of two cesarean sections. She was delivered by an elective cesarean section and had Apgar scores of 4 and 6 at 1 and 5 minutes, respectively. Ultrasound done at 22-week gestational age showed a single, viable fetus with hydrocephalus, short broad neck, and extended limbs. After adequate obstetric, genetic counseling and given all the information about the risks involving this pregnancy, the parents opted for conservative management without any intervention. On examination, she had the typical triad of KFS, including very short neck, low occipital hairline, and reduced bilateral neck movements. The child died immediately due to respiratory distress. Her parents refused to consent for postmortem scanning.

Conclusion: Although KFS is a rare syndrome encountered less commonly and a classical triad is present in almost 50% cases, one should closely investigate for other anomalies associated with it for better, early management and rehabilitation.

Keywords: Cervical fusion, Klippel–Feil syndrome, Ultrasound.

Introduction

Klippel–Feil syndrome (KFS) was first described by Maurice Klippel and Andre Feil in 1912 in a patient with congenital fusion of cervical vertebrae.¹ Klippel–Feil syndrome is a complex syndrome of osseous and visceral anomalies that include the classical clinical triad of short neck, limitation of head and neck movements, and low posterior hairline.² This malformation is responsible for the limitation of movement of the head and a significant risk of spinal cord injury.³ The exact incidence and prevalence are unknown because of unavailability of population screening studies. The estimated incidence is 1 in 40,000–42,000 births,⁴,⁵ and the predominance of women (3:2) has been reported. Klippel–Feil syndrome can be asymptomatic. However, skeletal, urogenital, cardiac, neurological, hindbrain, ocular, craniofacial, otolaryngeal, limb, and digital anomalies have all been associated with KFS in varying degrees.⁶ This variable expression together with KFS heterogeneity has greatly complicated the understanding and diagnosis of this developmental syndrome.⁶,⁷ The authors report a case of KFS which was managed conservatively.

Case Description

A 34 years old woman, GIII PII both were by C/S presented at Ian Donald ultrasound department for first-time scan at 22-week gestational age. There was consanguineous marriage, and an antenatal period was uneventful. There was no history of any drug intake or significant infection during the antenatal period. Ultrasound showed a single, viable fetus with hydrocephalus, short broad neck, and extended limbs (Fig. 1). After adequate obstetric, genetic counseling and given all the information about the risks involving this pregnancy, the parents opted for conservative management without any intervention. On a growth scan at 33 weeks, the cervical vertebrae were found to be fused with evidence of polyhydramnios (Figs 2 and 3). The patient underwent elective cesarean section at 37 weeks due to repeated cesarean section; she gave birth to female baby weighing 2,522 g and had Apgar scores 1, 4, and 6 at 1, 5 minutes, respectively. On examination, she had typical triad of KFS, including very short neck, low occipital hairline, and reduced bilateral neck movements. The child died immediately due to respiratory distress. Her parents refused to consent for postmortem scanning.

Fig. 1: Ultrasound image showing short spine and fusion of the cervical part

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of 4 and 6 at 1 and 5 minutes, respectively (Fig. 4). On examination, she was found to have short webbed neck, low hair line, short body length (Fig. 5). There was no obvious organomegaly. She had the typical triad of KFS, including very short neck, low occipital hairline, and reduced bilateral neck movements. The child died immediately due to respiratory distress. The child’s parents denied any postmortem scanning (Fig. 6).

**Discussion**

Klippel–Feil syndrome was originally described in 1912 by Maurice Klippel and Andre Feil. Their seminal report of this condition was based on a 46-year-old male French tailor who was found to have a short neck, low posterior hairline, and limited cervical movement. Such findings have since become synonymous with the term “clinical triad.” However, many authors had described KFS existing without this triad. When we reviewed history, we found that this anomaly had been described as early as the 16th century, where union between the second and third cervical vertebrae was found in an Egyptian mummy of about 500 BC. Klippel–Feil syndrome is a complex, congenital condition with heterogeneity in both phenotypic expressions and etiological background. It has many associated deformities like Sprengel’s deformity, synkinesia, scoliosis, facial asymmetry, hearing impairment, ocular abnormalities, cleft palate, congenital heart diseases, lung defects, and genitourinary problems such as abnormal kidneys or reproductive organs. The exact cause of KFS has yet to be determined, although some have suggested a genetic aetiology, while others attribute environmental factors such as alcohol...
consumption as a cause. Recent molecular studies have suggested that mutations of the MEOX1 gene may cause a recessive subtype of the syndrome. \(^9\) Mutations in the GDF6 gene, expressed in vertebrae and within the intervertebral disc, GDF3 genes, and PAX1 have all been implicated in the pathogenesis of the disease. \(^9\) More recently, a homozygous frame shift mutation in a protein-coding gene (RIPPLY2) has been found to be responsible for a new type of autosomal recessive KFS. \(^11\) The identification of mutations in these genes may lead to new advancement of treatment and possible explanation of associated anomalies.

High-index of suspicion should be maintained when diagnosing KFS, irrespective of clinical triad findings, especially in patients who exhibit associated spinal findings such as congenital scoliosis or spina bifida.

As such, early diagnosis of KFS is necessary in identifying high-risk cases and design measures to prevent potentially fatal outcomes.

**Conclusion**

Although KFS is a rare syndrome encountered less commonly and a classical triad is present in almost 50% cases, one should closely investigate for other anomalies associated with it for better, early management and rehabilitation.

**Consent**

Verbal explanation was given to the parents and an informed written consent was obtained both for the writing up and taking the pictures. A written informed consent was obtained from the parents for publication of this case report. The authors assure that the consent form will be produced at any time upon request.

**Author Contributions**

Sally Damra: Primary doctor who diagnosed the syndrome and followed the patient, she also edited and proof read the final version. Mona Awadalla: Conceptualized the manuscript, proof read and edited the final version. Hassan Osman: Consultant obstetrician and ultrasound expert who confirmed the diagnosis.

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**References**