International Journal of Academic Health and Medical Research (IJAHMR)
ISSN: 2643-9824
Vol. 9 Issue 4 April - 2025, Pages: 68-69

KLIPPEL FEIL syndrom : case report and literature review

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Abstract: *Klippel-Feil Syndrome (KFS) is a rare congenital disorder characterized by the fusion of cervical vertebrae, leading to a classic triad of a short neck, low posterior hairline, and limited neck mobility. This case report details a prenatal diagnosis of KFS in a 38-year-old woman presenting with severe hydramnios and fetal anomalies. A literature review is included to contextualize the clinical findings and discuss associated anomalies, diagnostic approaches, and management strategies.*

Keywords— Klippel-Feil Syndrom, hydramnios, congenital disorder, fusion of cervical vertebrae

1. INTRODUCTION

Klippel-Feil Syndrome (KFS) is a rare congenital disorder characterized by the abnormal fusion of two or more cervical vertebrae. First described in 1912 by Maurice Klippel and André Feil, this condition often presents with a classic triad of symptoms: a short neck, limited neck mobility, and a low hairline at the back of the head. However, the clinical presentation can vary widely, and KFS is frequently associated with other skeletal, cardiac, or renal anomalies. Early diagnosis and multidisciplinary management are crucial for improving quality of life and preventing complications.

2. CASE REPORT

A 38-year-old gravida with a history of one previous cesarean section presented to our obstetrics department with significant abdominal distension and respiratory discomfort. The patient reported an estimated gestational age of approximately 32 weeks; however, the pregnancy was unmonitored, with no prenatal ultrasounds or routine screenings conducted. There was no history of consanguinity or known genetic disorders.

2.1 Clinical Examination:

- **Vital Signs:** Stable; afebrile.
- **Abdominal Examination:** Marked distension with difficulty in assessing uterine size; fetal movements were faintly perceived.
- **Pulmonary Examination:** Fine crackles at the right lung base, suggestive of diaphragmatic compression due to uterine enlargement.

2.2 Investigations:

- **Obstetric Ultrasound:** Revealed massive polyhydramnios (amniotic fluid index >30 cm) and a singleton fetus in cephalic presentation. Fetal biometry corresponded to approximately 33–34 weeks of gestation. Notable findings included:

a) Shortened cervical spine with apparent fusion of multiple cervical vertebrae.

b) Abnormal spinal alignment with localized cervical kyphosis.

c) Low implantation of the head onto the trunk, indicating a short neck.

d) Reduced cervical mobility and fixed hyperextended head posture.

e) No major cerebral anomalies detected; however, postnatal MRI was recommended for detailed assessment.

f) No other visceral anomalies were clearly visualized, though the examination was limited by fetal position and excess amniotic fluid.

These findings were highly suggestive of Klippel-Feil Syndrome.

3. DISCUSSION

Klippel-Feil Syndrome is characterized by congenital fusion of cervical vertebrae, resulting from faulty segmentation during embryonic development (1). The classic triad—short neck, low posterior hairline, and limited neck mobility—is present in approximately 50% of cases

3.1 Associated Anomalies:

KFS is often associated with other congenital anomalies, including:

- **Skeletal:** Scoliosis, Sprengel deformity
- **Neurological:** Cervical myelopathy, as reported in cases presenting with tetraplegia following minor trauma
- **Cardiovascular:** Congenital heart defects
- **Renal:** Renal anomalies, necessitating thorough urogenital evaluation.
- **Craniofacial:** Cleft palate, facial asymmetry (2)

3.2 Prenatal Diagnosis:

Prenatal identification of KFS is challenging due to the subtlety of sonographic findings. However, key indicators include:

- Shortened cervical spine with vertebral fusion.
- Abnormal head-to-trunk positioning.
- Reduced neck mobility.
- Associated anomalies detectable via detailed ultrasound.

In our case, the presence of severe hydramnios and the aforementioned fetal anomalies prompted the suspicion of KFS.

3.3 Management and Prognosis:

Management of KFS requires a multidisciplinary approach, involving obstetricians, neonatologists, geneticists, and pediatric surgeons (3). Prenatal counseling is essential to discuss potential outcomes and plan for delivery in a tertiary care center equipped for high-risk neonates. Postnatal imaging, including MRI, is crucial for assessing the extent of vertebral fusion and associated anomalies (4).

The prognosis of KFS varies depending on the severity of vertebral fusion and the presence of associated anomalies. Early diagnosis and intervention can improve outcomes, particularly in preventing neurological complications.

4. CONCLUSION

This case underscores the importance of considering Klippel-Feil Syndrome in the differential diagnosis when prenatal ultrasound reveals cervical spine anomalies and severe hydramnios. Early detection facilitates appropriate counseling, delivery planning, and postnatal management to address potential complications associated with this rare congenital disorder.

5. ACKNOWLEDGMENT

The authors declare no interest conflit

6. REFERENCES

1. Sivri A, Mentes C, Hascelik Z, Ozker R. Klippel-Feil Syndrome: A Case Report and Review of the Literature. *Int Arch Surg*. 2005;3(2):163-166.
2. Kumar A, Sinha S, Singh A. Klippel Feil Syndrome: A Rare Case Report. *J Clin Diagn Res*. 2016;10(1):RD01-RD02.
3. Kumar S, et al. A Case Report of Klippel-Feil Syndrome Presenting as Tetraplegia. *Cureus*. 2023;15(7):e10387824.
4. Paradowska A, Szeląg B, Sławecki K. Klippel−Feil Syndrome - Review of the