ORIGINAL RESEARCH PAPER

Obstetrics & Gynaecology

KLIPPEL FEIL SYNDROME AN UNCOMMON OCCURRENCE IN PREGNANCY: A CASE REPORT

KEY WORDS:

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ABSTRACT

Klippel Feil syndrome was first described by Maurice Klipple and Andre Feil in 1912.This syndrome is characterized by skeletal, urogenital, cardiac, neurological, hindbrain, ocular, craniofacial,otolaryngeal, limb, and digital anomalies that have all been associated with Kleiple Feil Syndrome (KFS) in varying degrees. It has clinical triad of short neck, limitations of head and neck movement and low posterior hairline(1). The incidence is 1 in 42,000 births.(2) We report a case of 24 years old G3P1D1A1 patient at term with KFS(short stature with Rubella, CMV, HSV positive status with short neck and restricted neck movement, and posterior hairline managed with a complete evaluation of cardiac anomalies, neurological and oropharyngeal defects are also ruled out.) The patient has undergone elective cesarean section under the expert anesthesia team, and and neonatologist's opinions takenp for the baby to rule out KFS Patient whole genome sequencing done. Report showed significant characteristics suggestive of KFS

INTRODUCTION

CASE REPORT

A 24-year-old G3P1D1A1 AT 39 weeks of gestation presented to the emergency room on 27/12/2024 at 7.30 pm at GMCH CHH Sambhajinagar, for safe confinement. Patient was admitted with MRD NO 188417, She was G3P1D1A1 at 39 wks of gestational age not in labor.

She did not have any complaints.

No h/o consanguineous marriage

No h/o first-trimester fever with rash.

No h/o any drug intake or any history of exposure to toxins

There is no family history of any physical disability or similar illness

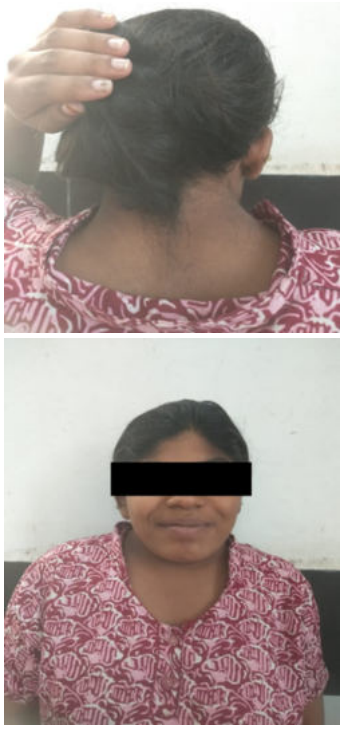

Fig 1- Patient With Posterior Hairline With Short Neck

On examination the patient was conscious ,well oriented ,moderately built HT- 138 cm WT- 52 kg BMI-27.36 KG/M2 with a short neck, restricted neck movement with low posterior hairline .She was pale, no clubbing, icterus or lymphadenopathy, no edema over the leg, periorbital region. on examination PR was 78/min, Her vitals were stable with CVS/RS within normal limits. P/A showed uterus 36 weeks, cephalic presentation,5/5 palpable, FHS146/min .uterus was relaxed.After informed consent pv examination done, os was closed, uneffaced, firm in consistency, posterior. Pelvis was contracted (both spine prominent with sacral promontory palpable) .The patient was not in labor. Keeping in mind the suspicion of Klipple Feil Syndrome, further investigations were done. The patient underwent Physician opinion, Cardiologist opinion, Otorhinolaryngologist opinion, and Neuro physician opinion.The required investigations sent along with Genome sequencing.

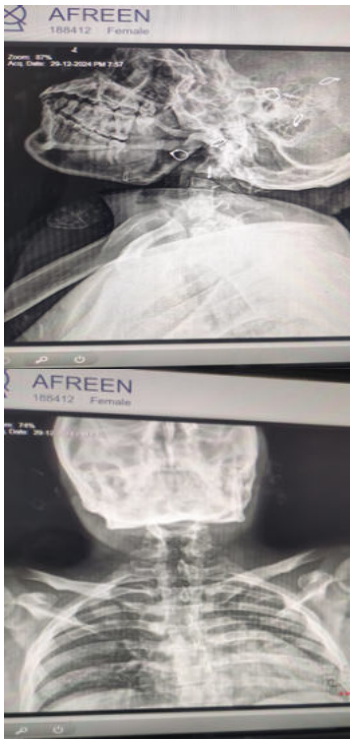

Fig 2 xray head and neck showing fused vertebrae

RUBELLA CMV HSV, IgG Antibodies positive, an X-ray skull and chest (PA view) was suggestive of fused C2-C3 vertebrae. 2D - ECHO was normal with 60% LVEF.Hearing test with tuning fork, neurological examination was within normal limit.

Whole-genome report suggestive of Variant Description:

The report describes a genetic variant in an unspecified gene. The variant is a point mutation where an adenine (A) base is replaced by a cytosine (C) base at position 575 (c.575A>C). This mutation leads to a change in the amino acid sequence, where glutamine (Gln) at position 192 is replaced by proline (Pro) (p.Gln192Pro). Highly suggestive of a variant , possibly KFS.

The final diagnosis is G3P1D1A1 at term gestation with KFS with short stature and contracted pelvis.

The patient was taken for Elective Cesarean section under the indication of CPD at term, LSCS done by a senior obstetrician under spinal anesthesia by a senior anesthetist.Operative was uneventful.There was no interop incidental finding (uterine anomaly if any) .She delivered a female of 2.6 kg, vertex presentation. Cried well immediately after birth,APGAR =8/10. No gross congenital anomaly. Neonatologist opinion advised to keep mother with the baby with immediate breastfeeding. Postoperative monitoring for five days was done and the patient was discharged home with the baby on 7th postop day with contraceptive advise and breastfeeding practices communication.

DISCUSSION

Klippel-Feil syndrome is a congenital condition characterized by the abnormal fusion of 2 or more cervical vertebrae, which results in a shortened neck. This skeletal anomaly also manifests with facial asymmetry, low hairline, and limited neck mobility, leading to chronic headaches, restricted neck mobility, and neck muscle pain. Moreover, Klippel-Feil syndrome may predispose individuals to spinal stenosis, neurological deficits, and cervical spinal deformities and instability, with some cases presenting with multiple syndromes(3). Diagnosis typically involves a comprehensive evaluation, including physical examination, imaging studies, and, in some cases, genetic testing. Management strategies aim to address symptoms and complications by using conservative approaches such as physical therapy to enhance mobility and muscle strength and pain management with medications or injections. (4)

Surgical interventions such as decompression and stabilization procedures aim to alleviate spinal cord and nerve pressure and improve spinal alignment in patients experiencing progressive symptoms. Long-term monitoring and interprofessional care are essential to effectively managing the condition's complications.

Klippel-Feil syndrome can be caused by heritable mutations in the GDF6, GDF3, and MEOX1 genes. GDF6 and GDF3 influence embryonic bone development. The MEOX1 gene encodes the homeobox protein MOX1, which regulates vertebral separation. GDF6 and GDF3 abnormalities are inherited in an autosomal dominant pattern, while MEOX1 mutations are autosomal recessive.(5)

Faulty cervical spine segmentation occurs during weeks 3 to 8 of embryonic development, resulting in persistent fusion of the involved vertebrae. The Samartzis classification scheme is frequently used to describe Klippel-Feil syndrome fusions. Type I involves a single congenitally fused segment, whereas type II involves multiple, noncontiguous, congenitally fused segments. Type III consists of multiple, contiguous, congenitally fused segments. About 25% of patients with Klippel-Feil syndrome present with type I deformities, while 50% have type II, and another 25% develop type III deformities.(6)

Klippel-Feil syndrome may be accompanied by various associated anomalies. A craniofacial examination may reveal abnormalities such as facial dysmorphism, microcephaly, or a low posterior hairline. Examination of the head and neck may reveal a short neck, limited neck mobility, and torticollis. Auscultation may detect murmurs if a cardiac disorder is present. Neurological signs can vary widely, with some patients showing no abnormalities while others may exhibit abnormal sensorimotor function and reflexes.(7)

Neonates exhibiting skeletal abnormalities during physical examination should undergo further evaluation for associated anomalies. For instance, some newborns diagnosed with Klippel-Feil syndrome may require an auditory brainstem response test to assess for sensorineural deafness.(8) Abdominal sonography may be used to investigate potential visceral anomalies, such as renal ectopia and Mullerian duplication in infants.

Approximately 50% of patients diagnosed with Klippel-Feil syndrome also exhibit concurrent scoliosis. In addition, around 50% of patients present with atlantoaxial instability, while approximately 30% experience renal disease, and another 30% suffer from deafness. (9) Laboratory and imaging tests are instrumental in identifying organ dysfunction in individuals with Klippel-Feil syndrome.

In our case, the patient presented very late at term with practically no investigations.We suspected KFS and investigated her thoroughly, finally reaching to the diagnosis.This patient ideally should undergo genetic counselling and amniocentesis in the early pregnancy to evaluate the outcome of pregnancy.This was explained to the patient.

Patients and their relatives must receive genetic counseling to guide family planning. In addition, patients and their parents should be informed about support groups such as the Genetic and Rare Diseases Information Center and Klippel-Feil Syndrome Freedom. These resources offer education, management techniques, and support to help individuals optimize their life options based on the severity of the condition.

CONCLUSION

Although kleipple feil syndrome is rare ,one should be vigilant in case a classical triad is present ,the case should be thoroughly investigated for other anamoly so that other management options are available .First trimester ultrasonography scan alongwith chorionic villi sampling lead to a early diagnosis of the fetus ,mild cases may go undiagnosed and appear at birth which may be traumatic for the partents.

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