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Klippel-Feil Syndrome associated with ventricular septal defect

Garg V.K.*, Agrawal V., Vijay P., Agarwal M.

Department of pediatrics and Obstetrics & Gynaecology, S. M. S. Medical College & Associated group of hospitals, Jaipur, Rajasthan, India.

Correspondence Address: * Dr. Vijendra Kumar Garg, S/o Mr. P. L. Gupta, H. No. 227, Joshi colony, Shahpura road, Neem ka thana, Sikar, Rajasthan, India-332713.

Abstract

Klippel-Feil Syndrome (KFS) is a rare congenital malformation characterized by the triad of short neck, low hair line and restricted neck movements, resulting from faulty segmentation along the embryo's developing axis during early weeks of gestation. Many other abnormalities may accompany the classical triad. In the present case we have found classical triad of KFS and associated ventricular septal defect and sprengel's deformity of scapula.

Keywords: Klippel-Feil Syndrome; Sprengel's deformity; Cervical vertebra; Short neck

Introduction

Klippel-Feil Syndrome (KFS) is a rare congenital malformation characterized by the triad of short neck, low hair line and restricted neck movements due to insufficiency of segmentation of two or more cervical vertebrae[1]. This syndrome was first described by Maurice Klippel and Andre Feil in 1912 in a patient with congenital fusion of cervical vertebrae[2]. KFS occurs in approxmately 1 of every 42,000 births, and 60% of cases are females[3]. Most affected people have one or two of these three characteristic features. Less than half of all individuals with Klippel-Feil syndrome have all three classical features of this condition. Restriction of neck motion is the most common finding[4].

Skeletal system abnormalities, urinary system abnormalities, synkinesis, hearing loss, congenital heart disease and brainstem abnormalities may accompany the classical triad[5].

Treatment for Klippel–Feil syndrome is symptomatic and may include surgical intervention. The prognosis for most individuals with KFS is good if this disorder is not associated with other visceral organ defects. Herein, we present a newborn female with clinical and radiological features of KFS, associated with ventricular septal defect and sprengel's deformity of scapula. We present this rare case of Klippel Feil Syndrome with the aim that such cases should be identified and treated at an early stage to minimize cosmetic & social stigma to them and to their families.

Case report

A female child was born to a 34 year old woman and a 37 year old man with non consanguineous marriage. She was the result of their third pregnancy which lasted

approximately 30 weeks. Antenatal period was uneventful without any exposure to radiation or teratogen. She was born by preterm vaginal delivery in department of obstetrics and gynaecology of our institute. Her birth weight, length, head circumference and chest circumference was 1350 g, 39 cm, 28.5 cm and 24 cm respectively. Her gestational age was between 28 to 30 weeks by new bellard scoring. She was admitted to neonatal intensive care unit of our institute because of prematurity with very low birth weight. On general physical examination she appeared to have a short neck, with her chin resting on the sternum anteriorly and the hairline reaching the level of the upper scapulae posteriorly (figure 1). She had limitation in neck mobility, including flexion and extension as well as rotation. She was suspected to have KFS as she had classical triad of short neck, low posterior hair line and restricted neck movements. Cardio vascular examination revealed pan systolic murmur best heard at left parasternal border of third to fifth intercostal space. There was no family history of any congenital malformation and her parents and two siblings were normal phenotypically without any evidence of neck abnormality.

X-ray examination of cervical spine showed incomplete segmentation of cervical vertebra between C_2 and C_3 and fusion of C_5 and C_6. There was also sprengel's deformity of right scapula (Figure 2). Echo cardiography revealed perimembranous ventricular septal defect of 4 mm with left to right shunt. Ultra sonogram of abdomen was normal while ultra sonogram of brain shows poor development of sulci and gyri suggestive of brain parenchyma of preterm neonate.

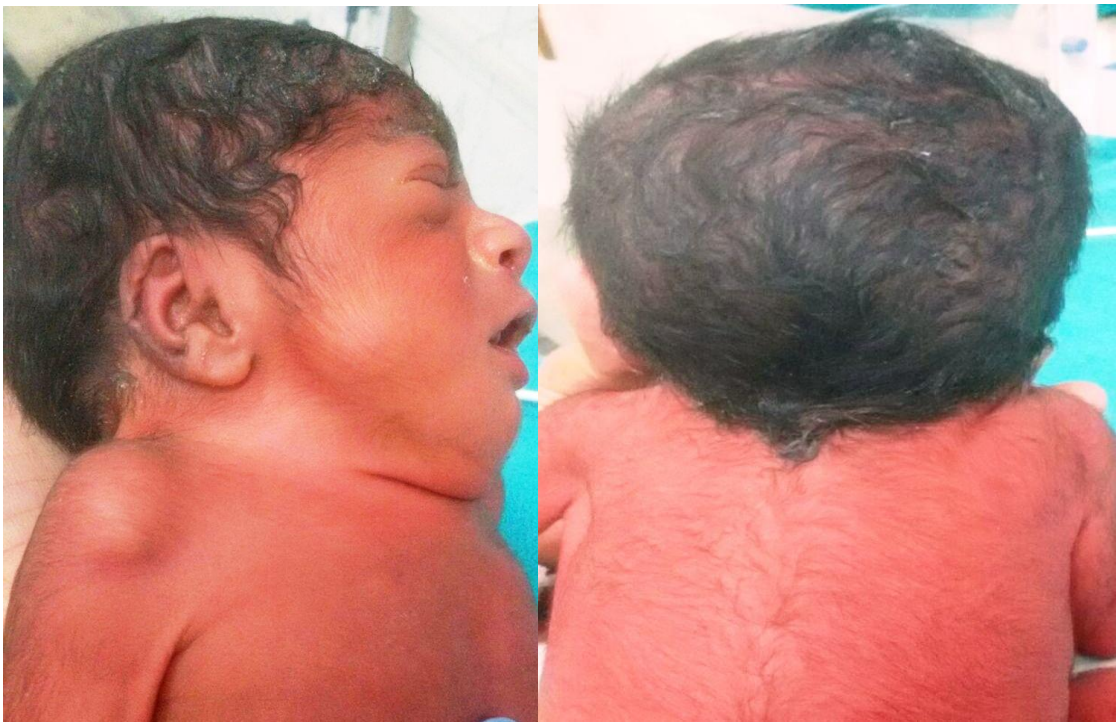

Fig. 1: Clinical photograph showing short neck and low posterior hair line

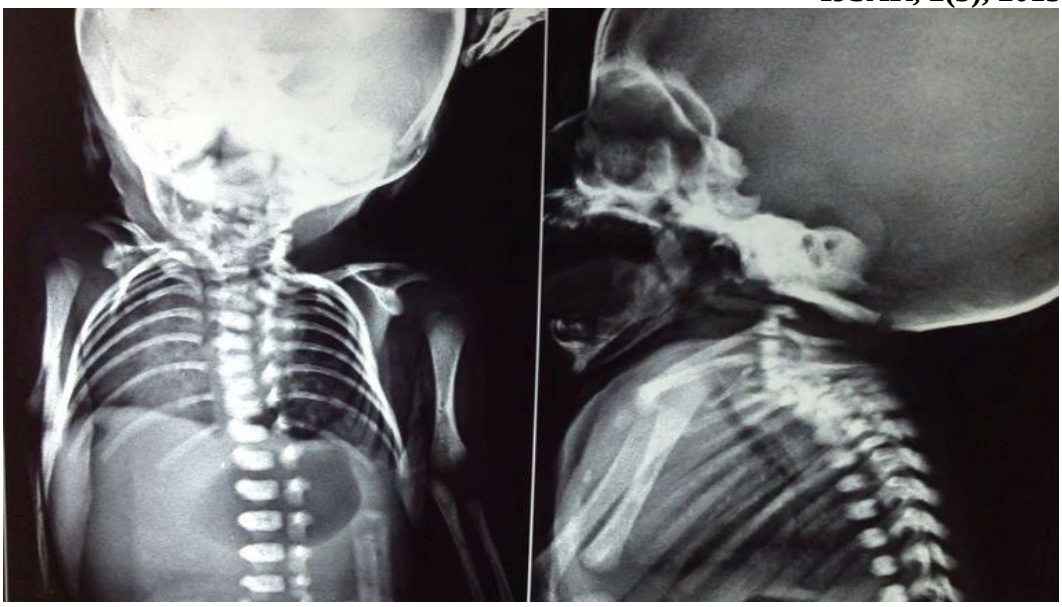

Fig. 2: Skiagram showing fusion of cervical vertebras and sprengel's deformity of scapula

Discussion

Klippel-Feil syndrome is a rare congenital disorder with fusion of cervical vertebra which may be familial or sporadic. This syndrome occurs due to failure of the normal segmentation and fusion processes of the mesodermal somites during early weeks of embryonic development[6].

KFS was first described by Maurice Klippel and Andre Feil in 1912 in a patient with congenital fusion of cervical vertebrae. There is multiple classification system for this syndrome. In 1919, Feil proposed three types of KFS based on the location and amount of fusion. Later, Clarke and co-authors suggested another classification system, where the defects were distinguished by inheritance mechanisms[7]. Recently, Samartzis et al. suggested a new classification system. According to this, Type I patients have a single-level fusion; Type II patients have multiple, noncontiguous fused segments; and Type III patients have multiple, contiguous fused segments[8]. Our case had fusion at two sites which are non-contiguous so classified as type II according to Samartzis classification.

KFS is characterized by the triad of short neck, low hair line and restricted neck movements due to insufficiency of segmentation of two or more cervical vertebrae. Other associated anomalies like scoliosis, spina bifida, sprengel's deformity of scapula, cleft palate, partial or complete conductive hearing impairment, respiratory problems, renal anomaly, underdeveloped low-set ears, congenital heart defects, anomalies of kidney and ribs may be present. Our case shows all three classical features of the KFS and was also associated with ventricular septal defect and sprengel's deformity of scapula.

Initial workup includes antero-posterior and lateral views of the cervical spine. If anomalies are found or suspected, careful assessment of the cranio-cervical junction by means of other imaging modalities is necessary to detect anomalies at that level[9]. Plain radiographs of the entire spine must be obtained to detect other spinal anomalies. Echocardiography is required to find out associated congenital heart disease. CT scan with three-dimensional reconstruction can be valuable in assessing anatomy for patients being evaluated for surgery. Ultrasonography of the whole abdomen is indicated to screen for associated renal anomalies.

Treatment for Klippel–Feil syndrome is symptomatic and may include surgery to relieve cervical or cranio-cervical instability and constriction of the spinal cord, and to correct skeletal deformity. Neurologic deficits and persistent pain are indications for surgery. Development of a compensatory curve in the thoracic spine may require surgical intervention or bracing.

The prognosis for Klippel-Feil syndrome depends on the specific anomalies present. The prognosis is good in the absence of associated abnormalities. The challenge to the clinician is to recognize the associated anomalies that can occur with Klippel-Feil syndrome and to perform the appropriate workup for diagnosis. All the cases of KFS should be thoroughly investigated in coordination with other specialists to formulate the further line of management. This disorder should be diagnosed timely, to provide close follow-up and appropriate therapy and parental counseling.

Conflicts of interest and source of funding: None

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