Rare Association of Klippel Feil Syndrome with Cleft Palate and Congenital Cardiac Deformities: A Case Report

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Klippel Feil Syndrome comprises of three characteristic deformities of short neck, a low dorsal hair line and restricted neck mobility. This is a case report of Klippel Feil Syndrome and its rare association with cleft of hard and soft palate, coarctation of aorta, dextrocardia and situs inversus. An interdisciplinary approach towards the management included cardiac surgery, cleft repair and complete oral rehabilitation of the patient. Presently the patient is undergoing speech therapy and is under regular follow up.

Keywords: *cleft lip and palate, dextrocardia, interdisciplinary approach*
J Clin Pediatr Dent 35(2): 213–216, 2010

INTRODUCTION

It is well known that there is a high likelihood of associated anomalies in patients with cleft lip and/or cleft palate. Studies have indicated that between 30% and 60% of individuals with clefts exhibit atleast one associated anomaly and this frequency is particularly high in patients with clefts of secondary palate.[1] There are over 300 syndromes associated with orofacial clefting.[2] Professionals in dentistry, medicine, or related disciplines have a responsibility to recognize the possible presence of complex syndromes in patients they see and to refer these patients to appropriate resources.

Klippel-Feil Syndrome is a condition characterized by abnormal segmentation of the vertebrae in the cervical spine (neck region). Patients with Klippel-Feil Syndrome have fused cervical vertebrae.[3] Klippel-Feil was initially described prior to x-rays, therefore the original descriptions of classic Klippel-Feil manifests with decreased range-of-motion of the neck, a short "webbed" neck and a low hairline.[4]

The frequency of Klippel-Feil syndrome is estimated to be approximately 1 in 40,000, but it is probably far more common because less severe fusions remain undetected in many patients. Most surveys report a female preponderance.[5] Although the disorder is sporadic, there are examples of familial occurrence; however, no clear mechanism of inheritance has been accepted.[5]

CASE REPORT

A female patient aged 8 years visited the Department of Pedodontics, The Oxford Dental College, Hospital and Research Centre, Bangalore, India, with a chief complaint of inability to speak properly since birth and the presence of an "opening" in her mouth. Her parents revealed that they had not noticed this "opening" in her oral cavity until when she was about 1 year old. They consulted a physician for the same but did not get any treatment/surgical closure done. This was the patient's first dental visit. The parents had two other children who had no significant medical or dental history. No history of consanguineous marriage was given.

Patient was moderately built and nourished with a communication abnormality of the articulation type and nasal phonation. Some of the characteristic features seen in this patient during the general physical examination were webbed neck, broad chest, widely spaced nipples and she was unable to flex her hands and neck completely. Oral examination revealed an incomplete midline cleft of the hard and soft palate. Other intra oral findings included root stumps with respect to 74, 75 and 85 and dental caries with respect to 84, 36 and 46.

Prior to cleft repair the patient was taken for anesthetic evaluation during which she was found to have right heart

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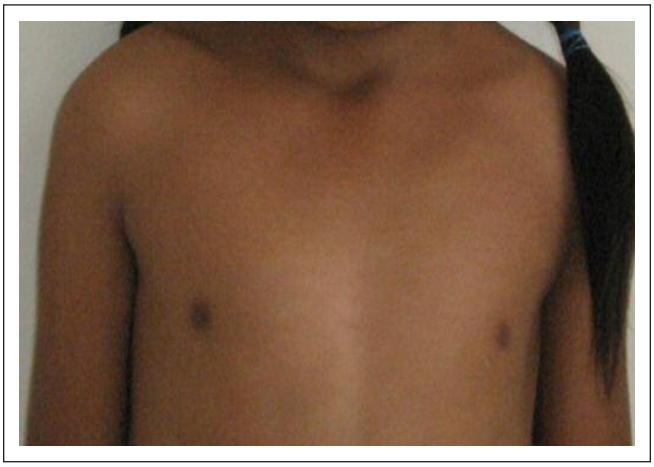

Figure 1. Photograph showing widely spaced nipples.

Figure 2. Photograph showing low hair line.

murmurs. At this stage, the patient was referred to other specialists such as a pediatrician, cardiologist and otolaryngologist for further assessment. The patient was found to have a normal blood picture. Radiograph of the chest showed an upward displacement of the scapula (Sprengel deformity), bifid ribs and dextrocardia. Electrocardiogram and congenital color doppler echocardiogram revealed dextrocardia, coarctation of aorta and *situs inversus.* Ear, nose and throat evaluation showed that she had moderate amount of hearing loss (70 db). Ultrasound of the abdomen was done to look for any renal abnormalities. Routine investigations done at our department included: anterior occlusal radiograph, orthopantamograph and lateral cephalograph.

The clinical evaluation and results of investigative procedures indicated that the patient could have either Turner's syndrome or Klippel Feil syndrome. But the chromosomal analysis of her blood sample showed 46,XX configuration, resulting in the diagnosis of the case as Klippel Feil Syndrome.

Prior to the cleft repair an elective artography and left heart catheterization was carried out. The patient was on prophylactic antibiotic regimen for a period of 10 days during which the cleft closure was done. After a period of one month complete oral rehabilitation was rendered under antibiotic prophylaxis. The dental treatment given comprised of extraction of primary root stumps, restoration of mandibular right first primary molar with restorative glass ionomer cement (Ketac Molar, 3M). The lower first permanent molars were restored with posterior composite material (3M) and a semi-fixed lingual holding arch was delivered.

Preventive dental treatment given to the patient included oral prophylaxis and topical fluoride application. Pit and fissure sealants were applied to the upper first permanent molars. Instructions on proper oral hygiene maintenance were given to both parents and child. A fluoride mouth rinse was also prescribed. Patient was referred for speech therapy and is currently under regular follow up at our hospital.

DISCUSSION

Normal human development results in an asymmetrical arrangement of the organs within the chest and abdomen. Typically, the heart lies on the left side of the body (levocardia), the liver and spleen lie on the right and the lung on the left have two lobes while the lung on the right has three lobes. This normal arrangement is known as *Situs solitus*.[6] However, in about 1 in 8,500 people, the organs of the chest

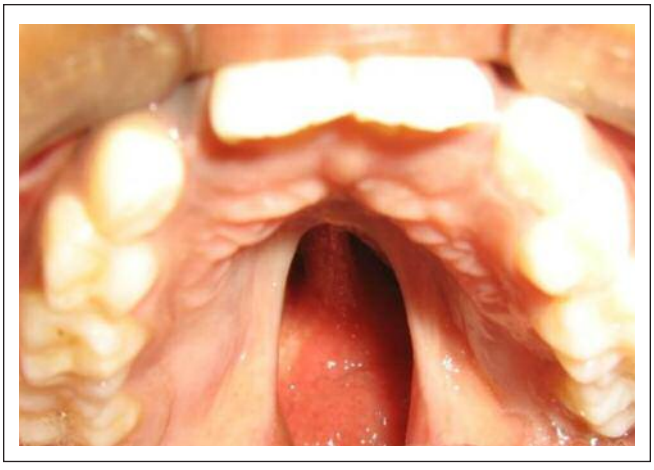

Figure 3. Preoperative photograph of maxilla showing cleft palate.

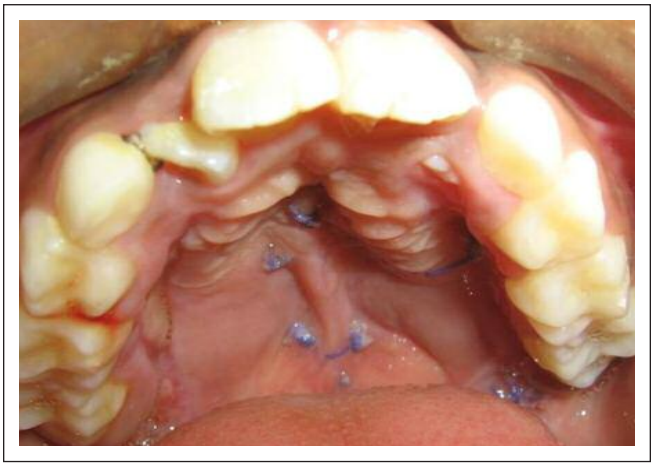

Figure 4. Post-operative photograph of maxilla showing surgical closure of cleft palate

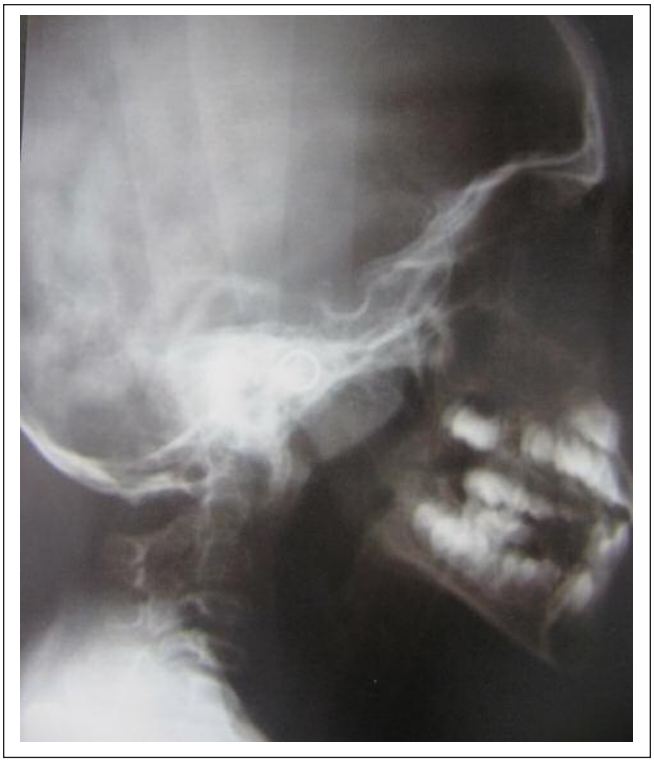

Figure 5. Lateral cephalogram

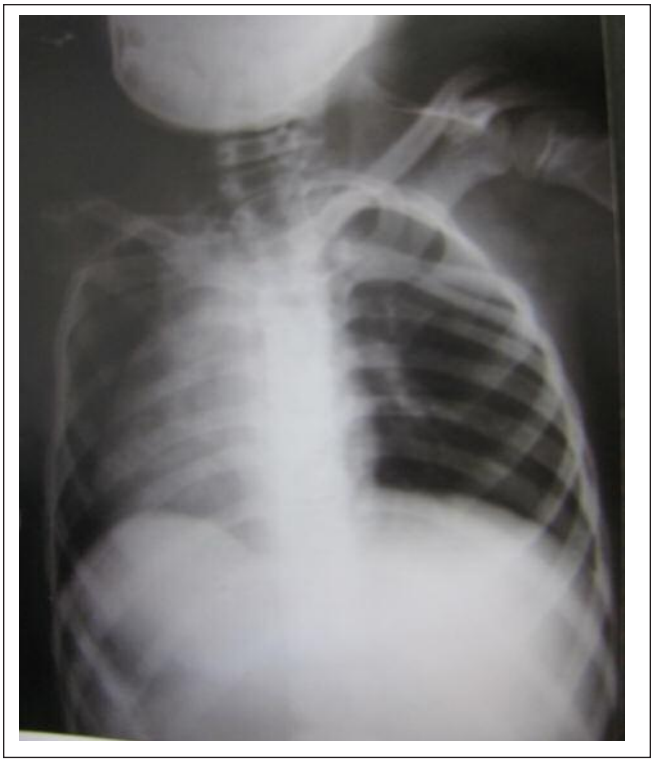

Figure 6. Radiograph of chest

and abdomen are arranged in the exact opposite position: the heart is on the right (dextrocardia), as is the two-lobed lung, and the liver, spleen, and three-lobed lung are on the left. This arrangement, called *Situs inversus,* is a perfect mirror image and since the relationship between the organs is not changed, functional problems rarely occur.[6]

Dextrocardia with Situs inversus is present at birth. The condition affects males and females in equal numbers. Approximately 0.01% of infants are born with this disorder. Autosomal recessive genes transmit the condition. The primitive loop in the embryo moves into the reverse direction of its normal position during fetal development, causing displacement of organs. Human traits including the classic genetic disorders are a product of interaction of two genes for that condition, one received from the father and one from the mother.[7]

Clefting of the secondary palate is present in 15% to 20% of the cases of Klippel Feil Syndrome. Hearing loss is seen in 25 to 50% of these patients. The combination of cleft palate and hearing loss explains the hypernasality in some patients with this syndrome. Congenital heart disease occurs in 4.2%[8] and ventricular septal defect is the most frequent heart anomaly documented. Whereas, in this patient congenital dextrocardia and coarctation of aorta were seen, which are very rarely found in association with Klippel Feil syndrome.

Sprengel deformity is a complex anomaly that is associated with malposition and dysplasia of the scapula. Congenital elevation of the scapula is caused by an interruption in the normal caudad migration of the scapula. This produces both cosmetic and functional impairment and probably occurs between the 9th and 12th week of gestation. An arrest in the development of bone, cartilage, and muscle also occurs. This condition also involves regional muscle hypoplasia or atrophy, which causes disfigurement and limitation of shoulder movement .The trapezius, rhomboid, or levator scapulae muscle may be absent, hypoplastic, or contain multiple fibrous adhesions. The serratus anterior muscle may be weak, leading to winging of the scapula. Other muscles, such as the pectoralis major, latissimus dorsi, or the sternocleidomastoid, may be hypoplastic and similarly involved.[9] The flared trapezius muscles in Klippel Feil anomaly give the patient an appearance that may be similar to that seen in Turner syndrome or Noonan syndrome (seen only in males).

Since this syndrome is mainly characterized by the fusion and occasionally agenesis of some parts of two or more cervical vertebrae, it affects the position of head and neck. The associated Sprengel deformity could also be the reason for inability to flex the neck and hand.

CONCLUSION

Thus the present case showed a rare association of Klippel Feil anomaly and Sprengel anomaly with cleft palate and congential cardiac deformities. Early diagnosis and timely intervention through an interdisciplinary approach is very essential in the management of such patients.

ACKNOWLEDGEMENTS

We thank team of doctors of Mahaveer Jain Hospital, Bangalore and the St Johns Medical College and Hospital, Bangalore for their help and support rendered during the medical management of the patient.

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