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LOBAR AGENESIS OF THE LEFT UPPER LUNG IN A PATIENT WITH KLIPPEL-FEIL SPRENGEL'S DEFORMITY: A CASE REPORT

Paediatrics

***Ajeet Kumar Yadav** MD., Department of Pediatrics; Dr. RML Hospital, New Delhi, India. *Corresponding Author

Ramkesh Meena MD., Department of Pediatrics; Dr. RML Hospital, New Delhi, India.

Surat Nabum MD., Department of Pediatrics; Dr. RML Hospital, New Delhi, India.

ABSTRACT

Klippel-Feil syndrome (KFS) is a rare disorder, presenting with craniofacial, other skeletal deformity and various other organ system anomalies. We report here a case of agenesis of upper lobe of left lung in a patient with Klipple-Fleil Sprengel's deformity.

KEYWORDS

Introduction:
Klippel-Feil syndrome (KFS) is characterized by congenital fusion of two or more cervical vertebrae. It was first described by Maurice Klippel and Andre Feil in 1912. Patients with KFS has classical clinical triad of short webbed neck, restriction of neck movements and low posterior hairline (1). Affected individuals may have other skeletal abnormalities like craniofacial abnormalities, cervical rib, bifid ribs, scoliosis, abnormal placements of scapula (Sprengel deformity)(2,3). The disorder is congenital and cases are identified later during hospital visits or when symptoms referring to other organ systems become apparent. KFS is estimated to occur in 1 of every 42,000 births, and 60% of cases are in females(3).

KFS may have variety of additional symptoms and other organ system abnormalities like, hearing impairment, genitourinary abnormalities, congenital heart defects, spina bifida or lung defects (4-6).

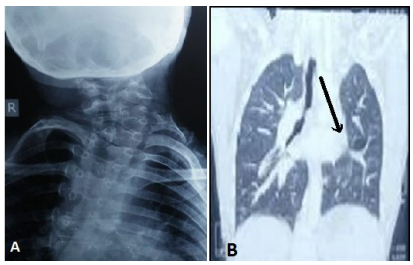
Pulmonary hypoplasia or aplasia is a rare congenital abnormality that represents primary embryogenic defect or secondary to situations that restrict fetal lung growth. Majority of reported cases had associated anomalies like skeletal, cardiovascular, gastrointestinal and genitourinary system (6,7). Lung agenesis in Klippel-Feil syndrome is an extremely rare entity and very few patients are known (7). Our case is agenesis of upper lobe of left lung in a patient with Klipple Fleil Sprengel's deformity.

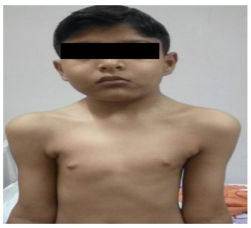
Case report: A 13-year-old boy admitted to our unit for the workup of anaemia. He presented with long history of easy fatigability, breathlessness and palpitation for two to three years and progressive paleness of whole body for two to three months. Easy fatigability, breathlessness and palpitation occurring on exertion and were not associated with any clinical features suggestive of heart disease. Paleness of whole body for two to three months without any localizing cause or blood transfusion in past. Patient had history of multiple episodes of hospitalization for pneumonia during pre-school age. Child was born as a product of non-consanguineous twin pregnancy. On clinical examination, patient was hemodynamically stable and systemic examination did not add any significant finding. Physical examination revealed facial asymmetry with short neck, low posterior hairline, restriction of side to side neck movement and mouth opening, left scapula was placed at higher level, discrepancy in level of nipples (right slightly at lower plane) and scoliosis was present. Laboratory tests found haemoglobin of 4.2 gm%, hematocrit of 12.9%, total leukocyte count of 5900 cumm, and platelet count of 2,60,000/cumm. His blood differential showed 56% neutrophils, 42% lymphocytes, 2% eosinophils, and peripheral smear shows moderate anisopoikilocytosis with presence of predominant microcytes and hypochromia. Liver, kidney functions, Ultrasound abdomen and Echocardiography were normal. X-ray neck and upper thorax showed finding suggestive of fusion of C4-C5 and C6-C7 vertebrae and scoliosis. CECT neck with chest showed hypoplastic posterior arch of atlas, fusion of C2-C3, C4-C5 vertebral bodies (block vertebrae), non-fusion of spinous process of C5 vertebrae, and C6 hemivertebrae noted with fusion of and C6-C7. CECT thorax showed non-visualisation (Agenesis) of upper lobe of left lung (Fig-1). Patient was managed conservatively and discharged on satisfactory background.

Discussion:
KFS is a rare musculoskeletal anomaly involving defect in cervical vertebral segmentation, which involve fusion of 2 segments or the entire cervical spine. KFS appears to be a failure of the normal segmentation and fusion processes of the mesodermal somites, which appear between the third and seventh week of embryonic development. Webbed neck, elevation of the scapula (sprengel's deformity) and congenital heart defects are frequently associated with this spinal anomaly(5).

Lobar agenesis of the lung is a very rare congenital anomaly, that may be observed in isolation or may be associated with other congenital defects of the cardiovascular, musculoskeletal, or gastrointestinal systems (9). Isolated lobar agenesis is usually asymptomatic and detected in adulthood when evaluated for an abnormal chest radiograph(10). Our patient had lobar agenesis of left lung, which was suspected as atelactesis of left upper lobe on chest roentogram and confirmed on CECT scan chest.

To conclude, in a patient with congenital anomaly of musculoskeletal system with respiratory symptoms demonstrating an abnormal chest radiograph, we should always suspect congenital anomaly of the lung. CECT may be useful in determining the extent of the lesion and to define associated abnormalities.





References:

1. Nagib MG, Maxwell RE, Chou SN. Identification and management of high-risk patients with Klippel-Feil syndrome. J Neurosurg. 1984 Sep;61(3):523–30.
2. Naikmasur VG, Sattur AP, Kirty RN, Thakur AR. Type III Klippel-Feil syndrome: case report and review of associated craniofacial anomalies. Odontology. 2011 Jul;99(2): 197–202.
3. Boraz RA, Irwin DH, Van Blarcom C. The dental rehabilitation of a patient with Klippel-Feil syndrome and Sprengel's deformity. Spec Care Dent Off Publ Am Assoc Hosp Dent Acad Dent Handicap Am Soc Geriatr Dent. 1986 Feb;6(1):22–4.
4. Falk RH, Mackinnon J. Klippel-Feil syndrome associated with aortic coarctation. Br Heart J. 1976 Nov;38(11):1220–1.
5. Mahiroğullari M, Ozkan H, Yildirim N, Cilli F, Güdemez E. [Klippel-Feil syndrome and associated congenital abnormalities: evaluation of 23 cases]. Acta Orthop Traumatol Turc. 2006;40(3):234–9.
6. Balachandran G. Klippel-Feil syndrome and anterior cervical meningomyelocele: a rare case report. AJNR Am J Neuroradiol. 2009 Oct;30(9):E130.
7. Mujeeb Rahman. A Case Report of Right Lung Agenesis With Klippel-Feil Syndrome. In Shanghai; 2016.
8. Hensinger RN, Lang JE, MacEwen GD. Klippel-Feil syndrome; a constellation of associated anomalies. J Bone Joint Surg Am. 1974 Sep;56(6):1246–53.
9. Clements BS. 1999. Congenital malformations of the lungs and airways. Pp. 1106–1136 in L. M. Taussing, and L. I. Landau, eds. Pediatric respiratory medicine. Mosby, St. Louis, MO, USA.
10. Zach MS, Eber E. Adult outcome of congenital lower respiratory tract malformations. Thorax. 2001 Jan;56(1):65–72.