Klippel Feil syndrome with Endocardial Cushion Defect-A rare case report

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Abstract:
Klippel Feil syndrome KFS is a rare anomaly defined as fusion of two or more cervical vertebrae. It is been associated with multiple system anomalies, of which cardiovascular system being less commonly involved or reported. Here we present a case of KFL with a new anomaly of heart, not reported so far. An 18 yr old female was referred with history of NYHA class II dyspnea and restricted neck movements. On Clinical examination she was found to have wide fixed second heart sound, mitral regurgitation with significant restriction of neck movement in lateral flexion and extension. CT scan revealed a block vertebra and a omo-vertebra. 3D echo showed partial Endocardial Cushion Defect with ostium primum ASD with cleft AML, Grade-II MR, 3mm peri membranous VSD with left to right shunt. Aortogram revealed aortic arch anomaly. A final diagnosis of KFS type 2 with sprengel shoulder with partial ECD with aortic arch anomaly was made in our workup. An early diagnosis, search for anomaly and prompt management could improve the quality of life of the patient. A vigilant search for cardiovascular system anomalies should be mandatory in KFS which could yield less morbidity and mortality in KFS patients.

Keyword: Endocardial cushion defect (ECD), Multiple Cardiac anomaly-Klippel-Feil Syndrome, omo-vertebra, block vertebra Klippel-Feil syndrome [KFS] is defined as a congenital fusion of two or more cervical vertebrae with a classical clinical triad of low hairline, short neck, and restriction of head and neck movements (1). The cause of this syndrome is believed to be due mutations in the expand GDF6 and GDF3 genes, responsible for growth and maturation of bone and cartilage (2). The mutation of morphogenetic protein family can occur as early as 2-8 weeks of embryonic period leading to faulty maturation of bone. In few other KFS, the cause of the syndrome is unknown. It was originally described in 1912, by Maurice Klippel and André Feil from France [3]. The incidence of KFS is 1 in 40,000 live births[4]. The 18th Dynasty of Egyptian Pharaoh Tutankhamun is believed to have suffered from KFS. [5].
An Initiative of The Tamil Nadu Dr. M.G.R. Medical University
University Journal of Medicine and Medical Sciences

Case summary:
An 18 year old female was referred to our outpatient department of Cardiology, with history of NYHA Class II dyspnea and difficulty in neck movements. There was no other significant cardiac symptoms in past history. Her higher functions and cognition were normal. Her school performance was normal and there was no family history of heart disease. On examination she had a normal intelligence, double helix of ear, scoliosis, short neck and low occipital hairline. There was significant restriction of neck movement in both lateral flexion and extension. No focal neurological deficit was noted. Her hearing assessment and ophthalmological examination was normal.

X-ray Cervical spine revealed fusion of C5 & C6 vertebra with presence of an Omo-vertebral bone. X-ray of both hands had an arachnodactyly. CT Cervical spine revealed C4, C5 as block vertebrae, a short cervical rib on left side and Omo-vertebral bone that originated from C5-C6 spinous process to spine of scapula. Fig (1) ECG sinus rhythm, prolonged PR interval, LAD, rsR in V1 was consistent with features of Endocardial Cushion Defect. 3D ECHO examination showed a partial endocardial cushion defect with ostium primum ASD with cleft AML, Grade-II MR, 3mm peri membranous VSD with left to right shunt. (Fig 2-5) LV angiogram showed the classical gooseneck deformity implying wedged LVOT with cleft AML and perimembranous VSD. Aortogram showed aortic arch anomaly with double innominate artery with laterally displaced origin of LIMA from Left SCA.

Discussion: KFS has been associated with anomalies of multiple systems, which includes skeletal system (60%), urinary system (35%), hearing loss (30%), facial asymmetry and flattening of neck (20%), synkinesis or mirror movements (20%), Cardiovascular system (4.2% to 14%) [1]. Among cardiovascular anomaly, VSD is the most common association, others include Atrial Septal Defects, Dextrocardia, Aortic stenosis and Patent Ductus Arteriosus [6], [7], [8], [9]. We believe this is the first case report that documents KFS associated with partial Endocardial Cushion Defect, Ostium Primum ASD with cleft AML and Aortic arch anomaly. This case study adds a new entity to the spectrum of cardiac anomalies in KFL patients.

Conclusion
The heterogeneity of presentation of the KFS suggests that a vigilant approach to the symptoms and thorough search for other anomalies could help the management of symptoms and help the patient lead a quality life. The discovery of new link between KFS and AV canal defect, would help decode the genetics basis of similar congenital heart diseases.

References:


