Klippel-Feil Syndrome: When Using Fiberoptic Bronchoscopy Guide, a Case Report

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Abstract

Klippel-Feil syndrome is a rare disease with congenital musculoskeletal condition characterized by faulty segmentation of cervical vertebrae and consists of cervical vertebra fusions with limitation of head movements, short neck and low posterior hairline. In several cases the syndrome is associated with cardiovascular malformations. Patients affected by Klippel-Feil syndrome could be an anesthetic challenge, not only during cardiac surgery. We are presenting a case of Klippel-Feil Syndrome in an adult patient, who was operated on for a pulmonary valve insufficiency in a previously corrected Tetralogy of Fallot Syndrome. We are going to discuss the features of this rare syndrome.

Keywords: Klippel-Feil syndrome; Anesthesia; Cardiac surgery; Tetralogy of fallot; Intubation; Vertebral fusion

Case Presentation

A 43-year-old man was referred by our Centre for evaluation of chronic pulmonary regurgitation. The man was affected with Tetralogy of Fallot, which was surgically corrected at an early age by preservation of his pulmonary valve. After a long follow-up course, he came to our attention with evidence of severe pulmonary valve regurgitation and right ventricle enlargement, which was diagnosed on echocardiogram. In order to clarify indications for surgery, he underwent an MRI scan, which confirmed the correct time and indication to surgery because of enlargement volume of right ventricle beyond 160 ml/m^2, a PR regurgitation fraction > than 35, an increased RV size (RV/LV ratio >1.5) and subjective and objective impairment in exercise capacity. The pulmonary valve was anatomically unsuitable for percutaneous pulmonary valve insertion (RVOT diam. > than 22 mm). No lung anomalies or disease were detected. The patient underwent an elective pulmonary valve replacement. Informed consent was taken. On the day of the procedure, the patient was placed on the operating table in supine position. The oro-tracheal intubation was performed by a senior anesthetist by using a fiberoptic bronchoscopy guide, as previously planned, due to the difficulty expected in relation to the Klippel-Feil syndrome associated with the Tetralogy of Fallot. A transesophageal (TOE) probe was also positioned, taking care regarding the cervical spine deformity, in order to assess the heart and the pulmonary valve before and after the surgical procedure. The operation was performed through a redo median sternotomy on cardiopulmonary bypass, on normothermia and on the beating heart, without clamping the aorta because there was no evidence of any residual inter-atrial or inter-ventricular shunts - potential sources of air embolism. Upon incising the pulmonary infundibulum, the pulmonic valve was visualized showing three cusps, but the cusps were thin and a lack of coaptation was noted - a finding often seen in patients with a history of Tetralogy of Fallot. The valve was replaced with a 25 mm Carpentier-Edwards tissue valve, well seated onto the annulus and secured by a single Prolene running suture. The patient came off cardiopulmonary by-pass without any difficulty, and the procedure was completely uneventful. TOE showed a good surgical result. The patient had an uneventful post-operative course in the ICU. He was extubated few hours after the surgical procedure without complications and transferred to the ward the day after in a stable condition. No damages were reported at the level of the cervical spine once extubated. He was discharged home on the fifth post-operative day.

Discussion

Klippel-Feil syndrome (KFS) was first described by Klippel and Feil in 1912 [1] and it is a

congenital malformation starting around the beginning of the fifth week of foetal life [2]. The incidence is about 1/40-42.000 births, and most of patients are women [1]. The classic triad of KFS consists of a short neck, low posterior hairline and restriction of cervical motion due to cervical vertebrae fusion. This last finding is the main clinical expression of the syndrome, which ranges from mild cosmetic deformity to severe neurological disabilities [3]. According to the primary description, the syndrome can be classified in 3 types [4]:

1)   Type 1: fusion of many cervical and upper thoracic vertebrae;

2)   Type 2: fusion (s) at only one or two cervical interspaces;

3)   Type 3: fusions in the cervical spine accompanied by fusions in the lower thoracic or upper lumbar vertebrae;

Different physiological defects, such as congenital scoliosis, rib abnormalities, deafness, genito-urinary abnormalities, Sprengel's deformity (congenital elevation of a scapula), synkinesia, and cervical ribs can occur in patients with KFS1. In some patients, the congenital synostosis can be observed as a part of a larger syndrome, such as Wildervank, Rokitansky-Kuster-Huser, or Goldenhar [5,6]. For example, in Wildervank syndrome one would normally find a Klippel-Feil anomaly, facial asymmetry, oral mucosal cleft, hearing loss, Duane syndrome, Chiari malformation and accessory bone mass in mandibular branch, leading to the clinical diagnosis of cervico-oculo-acoustic syndrome (COAS), also called Wildervank syndrome [7]. Other findings can be observed in different combinations, leading to the discovery of new syndromes.

Embryology and Genes of Spinal Development

KFS and other congenital spinal defects may occur by mutations, re-arrangements or disruptions in genes regulating segmentation and resegmentation. Fourteen days after conception, the gastrulation process generates mesenchymal cells responsible for creating head, heart, and lateral and paraxial mesoderm [8]. During 20th to 30th days the paraxial mesoderm is subdivided into spherical segmental structures called somites. As they mature, they form three sub sections: the sclerotome, which forms the adult vertebrae, the myotome, which forms the adult axial musculature, and the dermatome, which basically forms the adult dermis manner [9]. Geneticists use multiple tools to identify the genes involved in these process, but heterogeneous composition of patients, the large spectrum of anomalies associated with sporadic cases of KFS make complex research. KFS may be caused by sporadic mutation in the GDF6 or GDF3 gene and inherited in an autosomal dominant manner or it may be caused by the mutation in the MEOX1 gene and inherited in an autosomal recessive manner. The medical genetics literature contains descriptions of defects of known etiology, like fibroblast growth factor receptor-3 gene (FGFR3) [10], Notch pathway genes Notch1, D11 1, D11 3, Hes7, Psen1 which distrupt somite segmentation in mice [11,12], PAX family genes Pax1 and Pax9 which have been shown to regulate somite segmentation in mice [13,14], and other important genes such as those involved in HOX complex required for the normal specification and identity of vertebrae [15]. At the moment, there is no evidence of genes linking cardiac malformation with the cervical spine segmentation defects. Another important issue is the association of a bilateral (sometimes only unilateral) thoracic bifurcation of the common carotid artery (CCA) associated with KFS anomaly [16,17]. In this context we observe bifurcation of the CCA within the thoracic cavity between T-2/T-4, whereas normally we find the bifurcation between C-3/C-5. Moreover, at the bifurcation, the internal carotid artery (ICA) is posterior and medial to the external carotid artery (ECA) [18]. Genes involved into these processes might be responsible of KFS anomaly and perhaps of other abnormalities such as cardiovascular abnormalities. Currently there are only a few cases and reported studies, which is insufficient to draw a conclusion. However, we believe in this interesting hypothesis: probably even retardation during the involving processes of carotid bifurcation development, with a normal morphologic character of CCA axis, can lead to KFS anomaly. In this way, because of a very accurate timing of molecular processes, even a small delay can be very significant in segmentation process.

Cardiovascular Anomalies

For a cardiac surgeon the Klippel-Feil syndrome could be an interesting field. In 14% to 19% of cases the KFS is associated with cardiovascular anomalies where ventricular septal defects seem to be the most prevalent [19,20]. Morrison and al., reported a series of 505 patients with 24 affected by congenital cardiovascular defects (4.2%) [21]. Septal defect, abnormally positioned heart and patent ductus arteriosus were found in this group. Franzen, et al. [19], reported a case of aortic coarctation in a young man [22]. Bhagat, et al. [20] described a case of a patient with KFS and associated agenesis of right upper and middle lobes, hypoplasia of right lower lobe of the lung, and Lovn-Ganong-Levine syndrome [23]. An association with cor triatrium has been recently described [23]. Even the association of KFS, dextrocardia and situs inversus has been reported [24].

Conclusion

The anomalies to be found in Klippel-Feil syndrome are no more problematic for the surgeon than the single anomalies out of context of the syndrome. There are no different surgical techniques or approaches. The "real" problem, approaching these patients, could be an anesthetic problem as the airway management is. The literature reveals some reports of difficult intubation requiring fiberoptic bronchoscopy after several attempts, using direct laryngoscopy [25,26] and when dealing with patients affected by KFS, we consider it is important to keep in mind the possibility of performing the intubation directly with a fibroscope. An expert anesthetist would be able to perform the intubation avoiding any kind of complications, but these patients present infrequently, and we therefore need to be alert to the fact that they may have underlying hidden abnormalities. Further vigilance is required, not only during the airway management, but during the positioning of the patient both for the vulnerability of cervical spine/high neurologic injury risk and for possible low respiratory reserve.

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