

Brief Communication

Anesthetic management of patients with Klippel-Feil syndrome, a case series

Kamran Mottaghi¹, Farhad Safari¹, Parisa Sezari¹, Naeime Gholizadeh¹, Masoud Nashibi^{1*}

Abstract

Klippel-Feil syndrome (KFS) is a rare entity which is characterized by failure of normal segmentation of cervical vertebrae resulting in short neck with restricted movement and cervical instability. This anomaly increases the risk of neurological damage during airway instrumentation like laryngoscopy and positioning for surgery. We report three patients, a 42 years old man with KFS who scheduled for craniocervical fusion under general anesthesia, a 6 years old girl candidate for cleft palate repair and a 26 years old woman candidate for craniocervical fusion. All the patients were successfully managed using fiberoptic bronchoscope in first two cases and gum elastic bougie (GEB) in the last case.

Keywords: Klippel-Feil syndrome; Fiberoptic Bronchoscope; Gum Elastic Bougie

Please cite this article as: Mottaghi K, Safari F, Sezari P, Gholizadeh N, Nashibi M. Anesthetic management of patients with Klippel-Feil syndrome, a case series. *J Cell Mol Anesth.* 2017;2(2):63-8.

1. Anesthesiology Research Center, Shahid Beheshti University of Medical Sciences, Tehran, Iran

Corresponding Author:

Masoud Nashibi, MD, Assistant Professor, Anesthesiology Research Center, Shahid Beheshti University of Medical Sciences, Tehran, Iran, E-mail: masoudnashibi@sbm.ac.ir

Introduction

Most of the rare genetic disorders are considered as great challenges for anesthesiologists (1-3). KFS is a rare congenital fusion of cervical vertebrae due to a failure in the segmentation process of the cervical vertebrae in the early weeks of gestation and also may have other associated anomalies. The clinical presentation is known as the classic triad of short neck, limited neck movements and a low posterior hairline (4, 5). There are three subtypes of the condition classified according to the extent of vertebral involvement. KFS appears to be an etiologically heterogeneous condition with sporadic occurrence, autosomal dominant and autosomal recessive modes of inheritance reported. Gunderson suggested that KFS is a genetic condition, whereas

Gray found a low incidence of inheritance (6). Two small studies suggested that mutations of the MEOX1 gene, which codes for mesenchyme homeobox 1, may cause a recessive subtype of this syndrome (7, 8).

The incidence of KFS is 1:42,000 births and is one of the common congenital causes of difficult airway (4, 6).

The spinal deformities and increased risk of neurological damage during airway management is a challenge for anesthesiologist (9); hence, their anesthetic management should be carefully planned considering other comorbidities. We report successful anesthetic management in patients with KFS and associated anomalies and also review the literature regarding the various methods used for airway management in these cases.

Brief Report

Case 1

A 42 years old man was scheduled for craniocervical fixation. He was a known case of KFS with a short webbed neck. On examination the patient had a pulse rate of 86 beats/ min and a blood pressure of 125/82 mmHg. Examination of the airway showed that there was limited flexion and extension of the upper cervical spine, he had an adequate mouth opening and his Mallampati grade was 3. The systemic examination was normal. His other investigations were all within normal limits, except Magnetic Resonance Imaging (MRI) of the cervical spine which showed fusion of the upper cervical spines and brain stem compression (figure 1), scoliosis (figure 2) and sabre shaped tibial bone (figure 3).

Since anatomical landmarks in the neck were distorted and a difficult airway was anticipated, an awake fiberoptic intubation was planned.

The day before surgery, the procedure was explained to the patient. He was kept nil by mouth for 8 hours. On the day of surgery the patient was brought to the operating room where his ECG, SpO₂ and BP were monitored and an intravenous access secured with an 18G cannula on his right hand. Atropine (0.25 mg/IV) injected to desiccate patient's saliva. After calculating the maximum safe dose of local anesthetic, the patient was given 2% lidocaine viscous gargles which he was asked to swallow so as to coat the posterior surface of the epiglottis. This was followed by aliquots of a 10% xylocaine spray which was installed into each nares, with the patient taking deep breaths. Minimal sedation with 1mg midazolam and 50µg fentanyl was given intravenously to keep the patient calm, while the reversal agents were at hand. Phenylephrine nasal drop applied to each nostril and 2% lidocaine gel sniffed by the patient. Fiberoptic bronchoscope advanced through the right nostril. After visualizing the vocal cords, two ml of 2% lidocaine was injected through the side port of the fiberoptic bronchoscope to anesthetize the airway and vocal cords. The fiberoptic bronchoscope was then introduced into the trachea, the tracheal rings and the carina identified, the endotracheal tube inserted and its position confirmed after removal of the fiberoptic

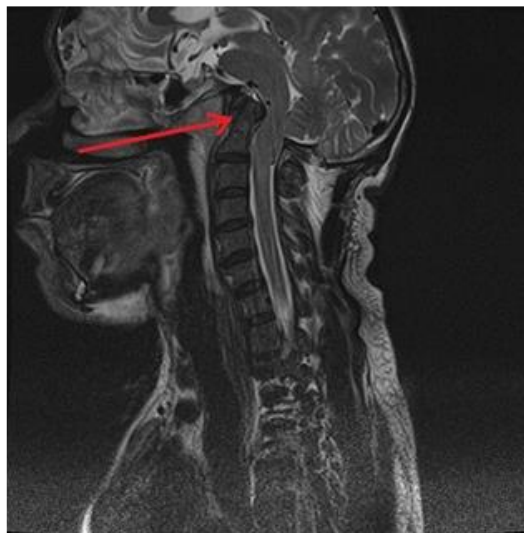


Figure 1. Brainstem compression.

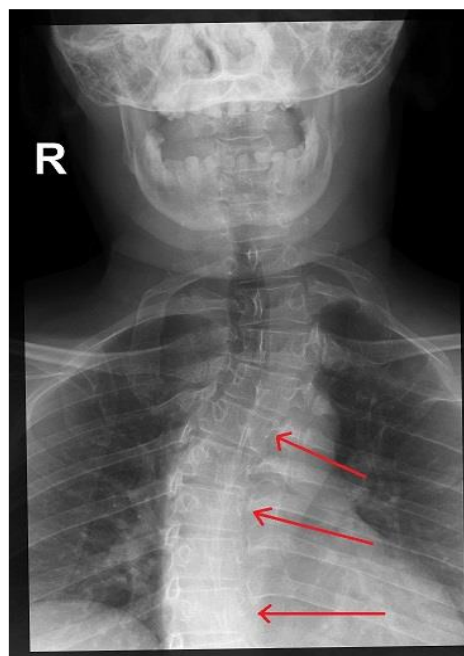


Figure 2. Thoracic scoliosis.

bronchoscope, by the chest movements as seen on inspection and bilateral symmetric air entry on auscultation. The EtCO₂ further confirmed correct placement of the endotracheal tube which was then fixed properly. Then Propofol 100 mg was given intravenously, the patient was then paralyzed with a non-depolarizing neuromuscular blocker (Atracurium) and anesthesia was maintained with isoflurane, 50% nitrous oxide and 50% oxygen, and fentanyl 50µg/hour.

Surgery proceeded uneventfully and patient



Figure 3. Sabre shaped tibia bone.



Figure 4. Webbed neck.



Figure 5. Fusion of cervical Vertebrae.

transferred to the Intensive Care Unit (ICU). On the next morning, he transferred to Operation theatre for extubation which was uneventful.

Case 2

A six years old girl, known case of KFS was a candidate for cleft palate repair. The patient had webbed neck (figure 4), cervical spine vertebrae fusion (figure 5), scoliosis, barrel chest (figure 6), dental deformities and severe limitation in head extension and rotation and mouth opening. The vital signs, lab data and respiration were normal. Since the patient was a six-year old girl, she was not cooperative enough to perform an awake fiberoptic tracheal intubation. On the other hand, we were concerned about airway management and probable difficulty in mask ventilation and tracheal intubation so we decided to intubate the patient using a fiberoptic bronchoscope after inhalational induction with sevoflurane. The incremental percentage of sevoflurane was used. After ten minutes patient's eyes were in central position, respiration had a regular pattern and Cerebral State Monitoring (CSM) showed the depth of anesthesia at 40. In the first attempt, we failed since the passage was obstructed by tongue. For the second attempt, jaw thrust performed which lifted the tongue and opened the passage and fiberoptic passed through the trachea and tracheal tube slit over. Surgery lasted about 2 hours. At the end the residual muscle relaxation reversed using neostigmine and atropine. The procedure completed uneventfully.

Case 3

A twenty-six years old woman complaining of mild weakness and paresthesia in all the four limbs was admitted for operative treatment of the cervical spine. The illness started one year back and had a progressive course necessitating her admission.

The patient had been diagnosed to have KFS several years previously and was a known case of hypothyroidism who was on levothyroxine treatment.

On examination the patient was a short, moderately built woman with a pulse rate of 85/minute and a blood pressure of 130/80 mmHg. She had a short webbed neck and a low posterior hairline with a limited extension of the neck (figure 7). Flexion and rotation were however not limited. She had an adequate mouth opening and her Mallampati grade was 3. Thyromental span was 6 cm. No dental abnormality or occlusion was present.

Cervical spine x-rays showed fusions at C2 to

C3 levels and C5 to C7 (figure 8). Chest x-ray showed thoracic kypho-scoliosis (figure 9).

Induction was conducted using sevoflurane 8% and 50% mixture of N₂O and O₂. When sufficient depth of anesthesia was reached, head fixation was applied gently by an attending neurosurgeon, a check laryngoscopy (Macintosh blade 4) was done and there was difficulty in visualization of the vocal cords so we used GEB as the first alternative technique in case of difficult intubation. The endotracheal tube slit over GEB and its position confirmed by the chest movements as seen on inspection and bilateral equal air entry on auscultation. The EtCO₂ further confirmed correct placement of the endotracheal tube.

After securing the orotracheal tube, the patient was rotated to the prone position with necessary precautions and maintenance of anesthesia managed with propofol infusion and non-depolarizing neuromuscular blocker.

After completion of the surgery, which lasted 300 min, the patient was returned back to the supine position and residual muscle paralysis reversed with neostigmine 4mg and atropine 1.2mg. Extubation of the trachea was accomplished after ensuring that the patient was awake and had adequate respiration. Post-operatively the patient was monitored in the recovery room after which she was sent to the ward.

Discussion

Rare congenital and genetic disorders are a great challenge for anesthesiologists. KFS occurs due to a failure of normal segmentation of the cervical spines in the 3rd to 8th week of gestation (10). Its etiology is unknown and clinical presentation is varied because of the different associated syndromes and anomalies that may occur. Other axial anomalies include cervical or fused ribs, cleft or hemi vertebrae, kyphosis, scoliosis, spina bifida occulta and sacral agenesis. Decreased range of movement of the neck with loss of rotation is the most consistent finding being more pronounced than the loss of flexion and extension. High cervical abnormalities can cause acute spinal cord compression following comparatively minor trauma. A Sprengel's anomaly occurs in 20-30% of patients. Renal anomalies are common in individuals with KFS. Laryngeal

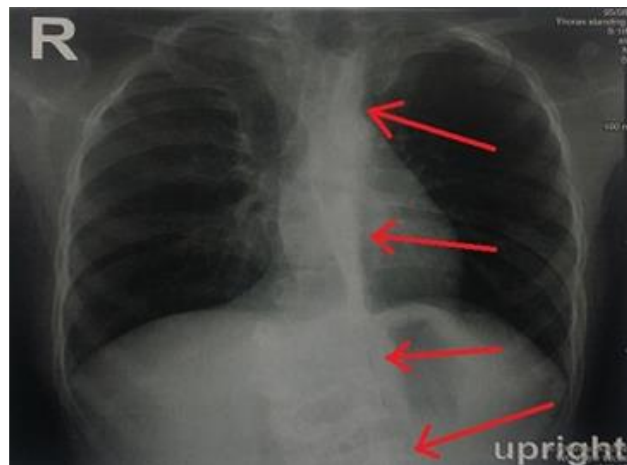


Figure 6. Scoliosis and barreled chest.



Figure 7. Limited head extension.

cartilages may be malformed, causing aphonia or other voice impairment. Cardiovascular anomalies occur in 14-29% of patients, the most common being Ventricular Septal Defect (11, 12). Other less common anomalies are congenital limb deficiencies, craniosynostosis, ear abnormalities and craniofacial abnormalities (13).

Out of three variants of KFS, Type I is an extensive abnormality in which several cervical and upper thoracic vertebrae are fused into a single block. In Type II variant, failure of complete segmentation occurs at one or two cervical interspaces. Associated anomalies were Sprengel's deformity and deafness. Type III variant includes Type I or II deformities with coexisting segmentation defects in the lower thoracic or lumbar spine. Most common form is Type II and it



Figure 8. Fusions at C2 to C3 levels and C5 to C7.

commonly involves C2-3 and C5-6 interspaces (13, 14).

In our first case, it is most likely type II variant with involvement of C2- C3 and C5 – C7 interspaces.

Patients with KFS may present for various surgical procedures at different ages. Commonly involved surgical procedures are cervical spine surgery, scoliosis correction, spinal canal stenosis surgery, renal surgery, cesarean section and eye or ear surgery.

The main anesthetic concern with these patients is the airway management during general anesthesia due to potential unstable cervical spine and abnormal atlanto-occipital junction, and an increased risk of neurological damage (15). Although several methods of having a secure airway have been reported in these patients, a specific standard recommendation on airway management does not exist. An awake fiberoptic intubation seems to be the most effective and safe method but sometimes difficult and time consuming as well (16).

Direct laryngoscopy with manual cervical stabilization has also been used successfully but even slight extension at atlanto-axial joint may cause damage to spinal cord (17). On the other hand, traction maneuver may limit the orolaryngeal alignment and make intubation more difficult. Neurological worsening following intubation under manual neck stabilization has not been reported (15). Cricothyrotomy is considered to be a good alternative,

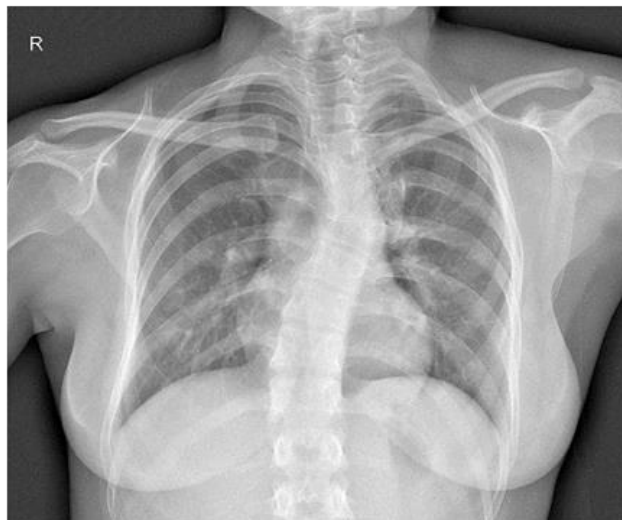


Figure 9. Thoracic kypho-scoliosis.

but an obstruction in the operative field remains a concern. Bullard laryngoscopy has been reported to be safe by some authors, but limited availability has made it less popular (18). The intubating LMA has also been used successfully for intubation without head and neck manipulation (19). But posterior displacement of the normal cervical spine due to cervical pressures generated by the laryngeal mask devices was also observed (20). O’Conner and Moysa reported that airway control could be temporarily lost after induction and an LMA might be required to secure the airway (21). Therefore, caution must be taken when dealing with an unstable cervical spine.

Due to earlier anticipation of difficult airway in these patients, it is indispensable to secure the airway before induction of general anesthesia especially in emergent patients. So we used awake fiberoptic intubation for airway management in our patients. This technique has some advantages, including but not limited to

- an awake, spontaneously breathing patient, who maintains own airway
- needs no spinal movement during intubation
- allows confirmation of tracheal tube placement
- a high success rate
- lower rate of complications
- better patient acceptance (19).

Despite the numerous advantages, failure of fiberoptic intubation with complications like massive subcutaneous emphysema or airway injury have also been reported (22, 23).

Conclusion

A thorough airway assessment and adequate preparation is a critical component for airway management in such cases. After considering all the modes of airway management, we advise awake fiberoptic intubation as one of the best and safest options for securing the airway in a patient with KFS, if it was not possible, inhalational induction and fiberoptic intubation would be a wise alternative.

Acknowledgment

The authors would like to thank the kind help of physicians and nurses, operating room and anesthesiology ward staffs, Loghman Hakim Hospital and Anesthesiology Research Center Shahid Beheshti University of Medical Sciences, Tehran, Iran for their kind help and support.

Conflicts of Interest

The authors declare that there are no conflicts of interest.

References

- Nashibi M, Tajbakhsh A, Mahdavi-pour Vahdati S, Safari F, Mottaghi K. Didmoad (Wolfram) Syndrome: Case Report. *Journal of Cellular & Molecular Anesthesia*. 2016;1(3):126-8.
- Nashibi M, Tajbakhsh A, Safari F, Mottaghi K. Anesthetic Consideration of Niemann-Pick Disease Type C. *Journal of Cellular & Molecular Anesthesia*. 2016;1(2):73-7.
- Safari F, Mottaghi K, Fallahinejadghajari R, Nashibi M. Crouzon Syndrome: a fibroblast growth factor receptor 2 gene mutation. *J Cell Mol Anesth*. 2017;2(1):15-8.
- Klippel M, Feil A. Un cas d'absence des vertèbres cervicales cage thoracique remontant jusqu'à la base du crâne. *Bulletins et Mémoires de la Société d'anthropologie de Paris*. 1912:101-2.
- Samartzis D, Lubicky JP, Herman J, Shen FH. Faces of Spine Care: From the Clinic and Imaging Suite. Klippel-Feil syndrome and associated abnormalities: the necessity for a multidisciplinary approach in patient management. *The spine journal : official journal of the North American Spine Society*. 2007;7(1):135-7.
- Gunderson CH, Greenspan RH, Glaser GH, Lubs HA. The Klippel-Feil syndrome: genetic and clinical reevaluation of cervical fusion. *Medicine*. 1967;46(6):491-512.
- Bayrakli F, Guclu B, Yakicier C, Balaban H, Kartal U, Erguner B, et al. Mutation in MEOX1 gene causes a recessive Klippel-Feil syndrome subtype. *BMC Genet*. 2013;14:95.
- Mohamed JY, Faqeih E, Alsiddiky A, Alshammari MJ, Ibrahim NA, Alkuraya FS. Mutations in MEOX1, encoding mesenchyme homeobox 1, cause Klippel-Feil anomaly. *Am J Hum Genet*. 2013;92(1):157-61.
- Khawaja OM, Reed JT, Shaefi S, Chitilian HV, Sandberg WS. Crisis resource management of the airway in a patient with Klippel-Feil syndrome, congenital deafness, and aortic dissection. *Anesthesia and analgesia*. 2009;108(4):1220-5.
- Kaplan KM, Spivak JM, Bendo JA. Embryology of the spine and associated congenital abnormalities. *The spine journal : official journal of the North American Spine Society*. 2005;5(5):564-76.
- Algom M, Schlesinger Z. Prolapse of the mitral valve in Klippel-Feil syndrome. *Chest*. 1981;79(1):127-8.
- Falk RH, Mackinnon J. Klippel-Feil syndrome associated with aortic coarctation. *Br Heart J*. 1976;38(11):1220-1.
- Chattopadhyay A, Shah AM, Kher A, Bharucha BA, Karapurkar AP. Craniosynostosis and Klippel-Feil syndrome: a rare association. *Indian J Pediatr*. 1996;63(6):819-22.
- Nagib MG, Maxwell RE, Chou SN. Identification and management of high-risk patients with Klippel-Feil syndrome. *J Neurosurg*. 1984;61(3):523-30.
- Whittle IR, Besser M. Congenital neural abnormalities presenting with mirror movements in a patient with Klippel-Feil syndrome. Case report. *J Neurosurg*. 1983;59(5):891-4.
- Daum RE, Jones DJ. Fiberoptic intubation in Klippel-Feil syndrome. *Anaesthesia*. 1988;43(1):18-21.
- Stallmer ML, Vanaharam V, Mashour GA. Congenital cervical spine fusion and airway management: a case series of Klippel-Feil syndrome. *J Clin Anesth*. 2008;20(6):447-51.
- Baraka A, Muallem M, Sibai AN, Louis F. Bullard laryngoscopy for tracheal intubation of patients with cervical spine pathology. *Can J Anaesth*. 1992;39(5 Pt 1):513-4.
- Al Zahrani T. Reverse LMA insertion in a neonate with Klippel-Feil syndrome--case report. *Middle East J Anaesthesiol*. 2007;19(3):625-9.
- Keller C, Brimacombe J, Keller K. Pressures exerted against the cervical vertebrae by the standard and intubating laryngeal mask airways: a randomized, controlled, cross-over study in fresh cadavers. *Anesthesia and analgesia*. 1999;89(5):1296-300.
- O'Connor PJ, Moysa GL, Finucane BT. Thoracic epidural anesthesia for bilateral reduction mammoplasty in a patient with Klippel-Feil syndrome. *Anesthesia and analgesia*. 2001;92(2):514-6.
- Maktabi MA, Hoffman H, Funk G, From RP. Laryngeal trauma during awake fiberoptic intubation. *Anesthesia and analgesia*. 2002;95(4):1112-4, table of contents.
- Richardson MG, Dooley JW. Acute facial, cervical, and thoracic subcutaneous emphysema: a complication of fiberoptic laryngoscopy. *Anesthesia and analgesia*. 1996;82(4):878-80.