orphanaesthesia

Anaesthesia recommendations for patients suffering

from

Klippel Feil syndrome

Disease name: Klippel Feil syndrome

ICD 10: Q76.1

Synonyms: Cervical vertebral fusion, Congenital cervical synostosis, Isolated Klippel-Feil syndrome, KFS

In 1912, Klippel and Feil (1) first reported on a patient with a short neck, a low posterior hairline, and severe restriction of neck movements due to complete fusion of the cervical spine, the classic clinical triad which is the hallmark of Klippel-Feil syndrome (KFS). It is estimated to occur in 1 in 40,000 to 42,000 newborns worldwide. Mutations in the GDF6 and GDF3 genes can cause KFS (2). But in some people there are no identified mutations in the GDF6 or GDF3 genes and the etiology remains unknown. Mutations in MEOX 1 have been found to occur in association with Klippel-Feil syndrome (3).

Most cases of Klippel Feil are sporadic. Some cases are due to autosomal dominant or autosomal recessive inheritance. It is a rare skeletal disorder primarily characterized by abnormal union or fusion of two or more cervical vertebrae. Other commonly associated anomalies include scoliosis, renal abnormalities, Sprengel deformity, deafness, synkinesia and congenital heart disease. The most common heart disease variant was ventricular septal defect. Less commonly associated were ptosis, lateral rectus palsy, facial nerve palsy and upper extremity anomalies.

Medicine in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

Find more information on the disease, its centres of reference and patient organisations on Orphanet: www.orpha.net

Disease summary

There are 3 variants of KFS. Type I is an extensive abnormality where elements of several cervical and upper thoracic vertebrae are incorporated into a single block. In Type II variant, failure of complete segmentation occurs at one or two cervical interspaces. Type III variant includes Type I or II deformities with coexisting segmentation errors in the lower thoracic or lumbar spine. Type II is considered to be the most common form. C2-3 and C5-6 are the interspaces commonly involved (4-12). The main anaesthetic concern with these patients is a potential unstable cervical spine and abnormal atlanto-occipital junction prone to an increased risk of neurological damage. Hence their anaesthetic management should be carefully planned with patient involvement keeping in mind the other abnormalities associated with the condition.

Typical surgery

Patients with Klippel-Feil syndrome present at different ages with an immense spectrum of clinical manifestations for various surgical procedures. Thus, the perioperative management of these patients varies individually. The majority of patients present for cervical spine surgery, scoliosis correction, spinal canal stenosis surgery, renal surgery, cleft palate repair, caesarean section, decompression surgery for degenerative disc disease (21,22) and eye surgery for Duane retraction abnormality (in some instances).

Type of anaesthesia

There are no recommendations for particular type of anaesthesia. Clinical judgement on a case-by-case basis is the key for safety.

General anaesthetic techniques are used in majority of the cases depending on the type of surgery. Airway management can be challenging in most of these cases mainly due to limitation in the range of neck movement due to cervical immobility and cervical instability could increase the risk of neurologic damage during intubation. Awake fibreoptic intubation is the method of choice for securing the airway in an adult patient (13). Patient should be well informed and the risks and benefits explained. A multispecialty involvement including ENT team stand-by for emergency tracheostomy is ideal. In paediatrics, airway management could include fibreoptic intubation after inhalational induction and a spontaneously breathing patient. Other methods of airway management will depend on the experience of the anaesthesiologist in- charge of the case.

Regional anaesthetic techniques including epidural, combined spinal and epidural, continuous spinal and single shot spinal have been used successfully for management of surgeries in these cases (14-20).

The anaesthesia produced by a neuraxial technique could be unreliable due to distortion of vertebral column and a compressed epidural or intrathecal space.Hence, vigilance and regular monitoring of the level of block is necessary for all patients (15).

Necessary additional diagnostic procedures (preoperative)

The extent of investigation will depend upon the age of presentation, the nature of the problem, presence of other associated abnormalities and the type of surgery.

Anteroposterior (AP) and lateral views of the cervical spine are required as a baseline. If abnormalities are present, other views may be required. CT scan of the head, in particular the base of skull and the cervical spine is a good tool for assessing bony abnormalities. MRI scan is useful to assess the spinal canal and any abnormalities of the spinal column, such as syringomyelia. CT myelography is another investigation, which will help assess the spinal canal and its contents.

Chest X-Ray may show cardiac abnormalities or fusion of ribs. A CT chest may be required depending on the presence of any abnormalities

Ultrasound is used for imaging of the urinary tract. Intravenous pyelogram (IVP) may also be required. All children should have an assessment of hearing.

Cardiac evaluation will include ECG and an echocardiography if there is suspicion of any other abnormalities. Other routine investigations including complete blood count, electrolytes, renal function tests and coagulation may be required.

If there is presence of any neurological abnormalities, a detailed neurological assessment and neurological review is recommended.

Particular preparation for airway management

Patient information, thorough airway evaluation and adequate airway preparation is the key to successful airway management.

Particular preparation for transfusion or administration of blood product

Blood loss may require replacement especially in major spine surgeries. However, the use of tranexamic acid and other pharmacological techniques (use of hypotensive agents) to minimise blood loss can avoid a potential blood transfusion (22-24).

Particular preparation for anticoagulation

There is no evidence to support the need of particular anticoagulation. However, if the patient is having a neuraxial technique then anticoagulation should be carefully reviewed before performing the procedure.

Particular precautions for positioning, transport or mobilisation

None reported.

Probable interaction between anaesthetic agents and patient's long-term medication

None reported.

Anaesthesiologic procedure

Airway management can be challenging in these cases. Hence, the experience of the anaesthesiologist and the type of surgery plays a key role. The safest option for securing the airway is awake fibreoptic intubation. The airway has to be prepared thoroughly before carrying out the fibreoptic intubation. Premedication with an anti-sialogogue helps to reduce the secretions.

Once the airway is secured, maintainance of general anaesthesia with either an inhalational anaesthetic or an intravenous technique is a matter of personal choice.

Full recovery of neuromuscular blockade if any should be achieved before extubation.

Patients who have surgery performed in prone position will require vigilance with reference to areas prone to hypoperfusion and pressure points.

Particular or additional monitoring

Standard monitoring should be applied in all cases. A few cases may require monitoring of temperature and neuromuscular blockade. The use of bispectral index monitoring is mandatory for total intravenous techniques after airway is secured.

Additionally some cases may require invasive arterial blood pressure monitoring and transoesophageal echocardiography.

Possible complications

Particular attention at the time of extubation will avoid possible post-operative airway complications. Careful extubation strategies should avoid the need for re-intubation. Adequate neuromuscular blockade reversal and monitoring during and after the recovery period is required for sedative or opioid side effects.

Postoperative care

The extent of postoperative monitoring depends on the surgical procedure and the pre-operative condition of the patient. High dependency or intensive care may be required, but not mandatory.

Information about emergency-like situations / Differential diagnostics

The possible emergency scenario is a patient for emergency caesarean section. An experienced anaesthesiologist who can manage airway should be in charge of the case. A neuraxial technique should be attempted if time permits.

Other emergencies are very unlikely.

Ambulatory anaesthesia

Patients with airway abnormalities should not be scheduled for ambulatory anaesthesia.

Obstetrical anaesthesia

Patients may present for management of pain relief during labour. There are reports of continuous epidural technique or spinal microcatheters for the management of pain relief. The use of remifentanil PCA may be considered.

Anaesthesia for caesarean section may require a general anaesthesia or regional technique depending on patient preference, technical issues with the back and extent of airway abnormalities (14-16,18,19).

Literature and internet links

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This guideline has been prepared by:

Author
Jameel Ahmed Khan, Anaesthesiologist, Royal Victoria Hospital Belfast, United Kingdom
drjameel@hotmail.co.uk

Peer Revision 1
Peter Tan, Anaesthesiologist, Sarawak General Hospital, Sarawak, Malaysia
petecstan@yahoo.com

Peer Revison 2
Philip F. Giampietro, Paediatrician, Waisman Center Madison, WI, USA
pfgiampietro@pediatrics.wisc.edu