orphanaesthesia

Anaesthesia recommendations for patients suffering from

Larsen syndrome

Disease name: Larsen syndrome

ICD 10: Q74.8; OMIM 150250

Synonyms: -

Disease summary:

First described in 1950, Larsen syndrome (LS) is a rare hereditary condition, characterised by multiple joint dislocations and characteristic facial, hand and feet abnormalities. Both autosomal dominant and recessive forms exist, although the former is much more common. Incidence of the disease is 1:100,000, with equal gender distribution. The autosomal dominant form of the disease is caused by mutations of the gene encoding filamen B, in a region containing human type VII collagen. This leads to abnormal collagen fibre formation, resulting in musculoskeletal and cardiac anomalies. The vertebrae are affected, causing cervical spine instability and kyphoscoliosis. Joints are prone to dislocation. Cardiac abnormalities can exist. Respiratory abnormalities occur due to decreased rigidity of cartilages in the airway and rib cage, leading to laryngotracheomalacia, bronchomalacia and lung hypoplasia. Repeated surgical interventions for spinal and musculoskeletal abnormalities are common, warranting careful anaesthetic evaluation of the airway, cervical spine, cardiovascular, respiratory and neurological function. Despite the multisystem organ involvement, patients can have productive lives with early corrective surgery and support.

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

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Typical surgery

Talipes correction and craniofacial surgery are typically performed in early life. As the bones grow, these patients may require repeated orthopaedic surgeries to correct skeletal dislocations or deformities. Spinal surgery such as cervical spine fusion and scoliosis correction are also common. Multiple revisions of spinal fusions may be required due to progression of the deformity.

Type of anaesthesia

General and regional anaesthesia can be used, although the disease involvement of the spine makes the latter technically challenging. Both intravenous and gas induction are safe. While there was one case report of malignant hyperthermia due to volatile agents, the evidence for this association was strongly disputed in the same journal. There is no contraindication to common anaesthetic drugs. Only in rare cases of distal muscle weakness due to cervical myelopathy should depolarising agents be avoided, due to the risk of causing hyperkalaemia. Regional analgesia such as caudal blocks have been described. [38]

Necessary additional diagnostic procedures (preoperative)

Additional tests are guided by a thorough assessment of the airway, cervical spine, respiratory, cardiovascular, neurological and musculoskeletal systems.

A thorough evaluation of the airway is necessary in these children. In clinically asymptomatic patients, indirect laryngoscopy will give a fair idea of the airway. Nasal fibreoptic endoscopy should be requested by ENT surgeons in uncooperative and symptomatic children to assess airway dynamics and to rule out the presence of laryngotracheomalacia and subglottic stenosis.

Because of the frequent involvement of the cervical spine, it is recommended to do a cervical X ray as a bare minimum investigation in all cases of LS. Cervical spine imaging, such as CT or MRI should be done if there are positive findings in the X Ray of the cervical spine or in the presence of neurological signs.

A pre-operative electrocardiogram should be considered. If a murmur is elicited on auscultation, cardiology review is required, and an echocardiogram may be necessary.

Pulmonary function tests may reveal a restrictive picture if kyphosis is significant.

Particular preparation for airway management

Difficult intubation should always be anticipated. Equipment such as paediatric bougies, supraglottic airway devices, videolaryngoscopes and fibreoptic endoscopes should be available. Previous anaesthetic charts should be reviewed due to the likelihood of repeated surgeries.

Manual in line stabilisation of the cervical spine is recommended even in the absence of neurological signs pre-operatively.

For short and uncomplicated procedures, consider the use of facemask or supraglottic airway devices to avoid the need for tracheal intubations. This would avert the risk associated with intubations and minimise manipulation of the neck. However, preparation for difficult intubation should remain in place.

Particular preparation for transfusion or administration of blood products

No special requirements for transfusion have been reported. Blood loss should be anticipated in complex and prolonged orthopaedic procedures.

Particular preparation for anticoagulation

There is no known association between LS and coagulopathy.

Particular precautions for positioning, transport or mobilisation

Careful position of patients with LS is extremely important. The cervical spine needs to be handled with care, especially during prone positioning for vertebral surgery. Large joints are at risk of dislocations, and needs to be carefully positioned.

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

Not reported.

Anaesthesiologic procedure

Premedication is generally safe. Both intravenous or gas induction can be used. Once asleep, cervical in line stabilisation should be maintained and difficult airway anticipated. Because of the the presence of laryngotracheomalacia commonly, it may be safe to use muscle relaxants only after confirming adequate bag mask ventilation. Common anaesthetic agents are safe to use. Additional monitoring may be required depending on type and duration of procedures. Positioning of transfer of patient need to be done carefully to minimise risk of dislocations. Extubation requires careful planning due to the high incidence of airway oedema and laryngotracheomalacia.

Particular or additional monitoring

For surgery involving the spine, intraoperative neurophysiological monitoring may be required, such as somatosensory or motor evoked potentials, in order to minimise the risk of spinal cord damage during surgery.

Invasive monitoring should be considered if the patient has severe cardiac involvement.

Careful monitoring of patient positioning during prolonged procedures would help to reduce the risk of musculoskeletal injury.

Possible complications

Complications arise mainly from known risks. Cervical injury due to excessive neck manipulation can occur, particularly if tracheal intubation is difficult. There have been numerous reports on respiratory complications after surgery. Post-extubation airway oedema may cause croup / stridor. The presence of laryngotracheomalacia or subglottic stenosis may further contribute to respiratory complications in the postoperative period. . Bronchomalacia and lung hypoplasia may contribute to respiratory failure during the post-operative period. Musculoskeletal injury can occur due to suboptimal positioning.

There was one case report of intraoperative cardiac arrest during spinal surgery under sevoflurane anaesthesia. This was attributed to a combination of a known pre-existing cardiac disease and stress of scoliosis surgery.

Postoperative care

High dependency or intensive care unit admission should be considered if the patient has significant organ involvement of the disease, or if surgical procedure is prolonged and complicated. Patients with severe kyphoscoliosis may require additional respiratory support post-extubation.

Information about emergency-like situations / Differential diagnostics

caused by the illness to give a tool to distinguish between a side effect of the anaesthetic procedure and a manifestation of the disease

There are no known complications reported in the literature.

Ambulatory anaesthesia

There are no reported day case procedures in the literature. However, this can be considered depending on the preoperative condition of the patient and type of surgery.

Obstetrical anaesthesia

Due to pelvic and hip abnormalities in LS, planned caesarean sections are preferred.

General anaesthesia is best avoided. Pregnancy induced changes to the airway and respiratory system would compound the problems in Larsen syndrome. However, spinal or epidural insertions would be technically challenging due to vertebral abnormalities. Time permitting, a regional technique consisting of an epidural catheter with gradual titration of local anaesthetic is ideal. This reduces excessive rostral spread, and minimises the risk of

respiratory complications. Difficult airway equipment should be prepared in case a general anaesthetic is required.

Neonatal cervical spine protection during delivery may be required if pre-natal screening reveals a likelihood of baby having LS.

Attachment 1
Table 1: Summary of anaesthetic considerations in Larsen syndrome

System	Anaesthetic considerations
Airway	Anticipate difficult airway obtain imaging for cervical spine Maintain manual in line cervical stabilisation during intubation Care with extubation
Respiratory	Potential complications due to tracheo-bronchomalacia and lung hypoplasia. Close monitoring post-operatively
Cardiovascular	Investigate for possible underlying cardiac disease Obtain ECG, consider cardiology review and transthoracic echocardiogram
Neurological	Meticulous handling of cervical spine throughout the case to reduce the risk of spinal cord injury
Musculoskeletal	Exercise care with transfer and positioning, especially large joints

Literature and internet links

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Internet links :

1. National Organisation for Rare Diseases
https://rarediseases.org/rare-diseases/larsen-syndrome/

2. Patient support in the UK :
http://www.cafamily.org.uk/medical-information/conditions/l/larsen-syndrome/

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