orphanaesthesia

Anaesthesia recommendations for

Jarcho-Levin syndrome

Disease name: Jarcho-Levin syndrome

ICD 10: Q76

Synonyms: Spondylocostal dysostosis

Disease summary: Jarcho-Levin Syndrome (JLS) is an inherited costovertebral dysplasia. It has an autosomal dominant and recessive trait inheritance [1]. Saul Jarcho and Paul M Levin first described this disorder in 1938 [2]. Global incidence of JLS is 1/40,000 birth which makes it a rare genetic disorder [1]. It is related to SSRT/ARS2 (Serrate RNA effector molecule homolog or Arsenite-resistance protein-2 gene. It is caused by mutation in DLL3 gene mapped on 19q13 sequence [3]. The intelligence levels (IQ) remain normal in all reported cases. Its exact incidence in Asian continent is unknown, but a few cases of JLS are reported in India [4]. JLS has two phenotypes: Spondylothoracic dysplasia (STD), which is autosomal recessive and Spondylocostal dysplasia/dysostosis (SCD), which is autosomal dominant, both being associated with multiple other anomalies. Both types are due to failure of vertebral segmentation. Infants usually present with vertebral and rib anomalies along with a plethora of non-skeletal abnormalities including hydrocephalus, neural tube defects, tracheal abnormalities, cardiac, renal, gastrointestinal and urinary tract abnormalities [4,5].

Axial skeleton growth defects are manifested in the form of vertebral column malformations, intrinsic rib anomalies, small thoracic cavity, short stature, scoliosis and kyphosis, leading to protuberant abdomen and severe pulmonary complications [6].

Respiratory insufficiency, pulmonary hypertension and other multi-system anomalies are the major causes of mortality in these patients.

Medicine is in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnosis is wrong

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Typical surgery

Patients with JLS generally present in infancy with neurological problems which may require neurosurgical procedures (emergency or elective), like Ventriculo-Peritoneal (VP) shunt insertions. Thorough pre-operative evaluation should be done, with detailed systemic examination for multi-system afflictions of this rare syndrome.

They may also present for throacospinal corrective surgeries, performed as staged operations in prone position, with its attendant complications.

They can also present for orthopaedic correction surgeries, requiring repeated anaesthesia exposures. Rarely, they can also present for obstetric anaesthesia if there is uneventful survival till adulthood.

Type of anaesthesia

General anaesthesia is most commonly employed in these children. All considerations for neonatal/paediatric anaesthesia are required, with possibility of difficult airway and respiratory dysfunction. Controlled mechanical ventilation is usually preferred for adequate oxygenation. Central neuraxial anaesthesia is difficult due to spinal abnormalities and not recommended. Ultrasound-guided nerve blocks can be given to supplement anaesthesia for peri-operative analgesia.

Necessary additional pre-operative testing (beside standard care)

Besides standard pre-anaesthetic investigations, these children will need neurological work-up, including magnetic resonance imaging (MRI) of the brain and spine. Cardiac and abdominal evaluation may be required in children with extensive multi-system disease. However, emergency procedures may not give adequate time for detailed assessments.

Chest x-ray films may show crowding of ribs in a crab like pattern and dextrocardia in cardiac silhouette. Advanced radiological investigations like MRI of the spine and brain may reveal fusion defects in the lower back and upper trunk region with herniation of nerve roots, Arnold Chiari malformation and/or chronic hydrocephalus [7].

Particular preparation for airway management

Children with JLS have difficult airway due to skeletal growth defects, hydrocephalus and cranio-facial abnormalities. In addition, due to severe scoliosis and kyphosis, they have minimal respiratory reserves and increased chances of hypoxia during apnoeic episodes. In addition airway access can be challenging because of the thoracic lordosis and the compensatory cervical kyphosis that may make intubation very challenging even with proper positioning. Difficult airway cart needs to be prepared beforehand. A silicon or cotton donut with padding needs to be used to position the patient supine on operation table prior to induction for accommodating the meningomyelocele sac. Paediatric video laryngoscopes and fibre-optic bronchoscopes must be available. Adequate pre-oxygenation and presence of expert anaesthesiologist help are needed. Standard difficult airway guidelines and algorithms need to be followed for patient safety.

Particular preparation for transfusion or administration of blood products

There are no specific recommendations for blood or blood products transfusion. Adequate, cross-matched blood needs to be arranged for spinal corrective procedures.

Particular preparation for anticoagulation

There are no particular recommendations for anticoagulation in these patients. A coagulation profile may be done before major surgery in these children.

Particular precautions for positioning, transportation and mobilisation

Children with JLS have multiple deformities, hence positioning needs to be done carefully to avoid injuries. Padding of pressure points is mandatory to prevent neuropathies and pressure sores. Particular positioning difficulties can arise in the presence of meningomyelocele, where donuts (silicon or cotton) are used to cushion the posterior swelling during induction, to prevent their rupture.

Interactions of chronic disease and anaesthesia medications

There has been no reported literature on interactions of chronic diseases and anaesthetic medications in JLS. Nevertheless, it is better to avoid histamine-releasing agents.

Anaesthetic procedure

Due to distorted airway anatomy, difficult positioning and recurrent infections, the challenges for anaesthetic management become manifold. Anaesthetic management of any JLS case is challenging for anaesthesiologist due to distorted anatomy of rib cage, high incidence of lower respiratory tract infections and anomalies of tracheal and bronchial lumen (e.g., missing tracheal rings, narrow bronchial lumen, wider carina, short neck and small or distorted thorax) [8].

If dextrocardia [9] is present, one must remember to place the precordial stethoscope for auscultation on the right chest, rather than the usual left side. Oto-rhino-laryngology and paediatric consultation need to be obtained to rule out other congenital anomalies. Difficult airway cart must be prepared preoperatively for both induction and extubation. Airway of a JLS patient is very sensitive to infection because of distorted anatomy and high risk of aspiration. Intubation in JLS patients should always be considered difficult and all necessary arrangements to be made prior to taking the patient on the table. Positioning for induction and surgery of such children can also pose great difficulties. Gentle handling of children especially during laryngoscopy is warranted with diligent post-operative monitoring. Care and padding of spinal deformities and meningomyelocele sac is advisable.

Controlled mechanical ventilation under general anaesthesia is usually recommended, especially in paediatric patients undergoing spinal or neurosurgical procedures. In addition, ultrasound-guided nerve blocks can be given by trained anaesthesiologists for peri-operative pain management.

Particular or additional monitoring

Standard ASA (American Society of Anaesthesiologists) monitoring must be applied to all patients. In addition, neuromuscular monitoring, invasive arterial and central venous monitoring line can be instituted in major surgeries and spinal corrective procedures. Arterial blood gas measurements can be monitored serially for correcting metabolic and electrolytes derangement. Temperature monitoring is mandatory, especially in paediatric patients. Strict intake-output charting with hourly urine output monitoring is recommended.

Possible complications

Presence of vertebral, rib and tracheal anomalies lead to increased incidence of difficult airway and post-operative respiratory complications. These patients may require post-operative mechanical ventilation, prolonged ICU stay and its attendant complications. Neurological problems in patients of JLS can have a protracted course. The longest surviving reported case of JLS is of 33 years [9].

Paediatric patients with JLS undergoing spinal, neurosurgical and orthopaedic procedures may have multi-system disorders like respiratory failure, cardiovascular complications, renal dysfunction, problems due to blood transfusion and metabolic derangements. Other concerns like peri-operative laryngospasm and bronchospasm are common due to airway anomalies and frequent respiratory tract infections in these children.

Post-operative care

Patients of JLS will generally require post-operative ICU care and continued monitoring post-operatively. Post-operative care can be protracted with prolonged post-operative mechanical ventilation and management of other multi-system abnormalities. Padding of pressure points is required to prevent pressure sores. Care of invasive monitoring lines need to be continued in the ICU along with diligent monitoring.

Disease-related acute problems and effect on anaesthesia and recovery

Disease-related acute problems in JLS patients include distorted airway anatomy resulting in difficult airway and “cannot-ventilate, cannot-intubate” (CVCI) situation; frequent respiratory tract infections leading to increased incidences of laryngospasm and bronchospasm; rib, chest wall and vertebral anomalies causing prolonged mechanical ventilation, hypoxia and metabolic derangements; dextrocardia or other cardiac anomalies resulting in difficulty in cardiovascular monitoring and arrhythmias; spinal abnormalities resulting in neurological problems, stunted growth, pressure ulcers and repeated surgical/anaesthetic exposures. Anaesthesia can be quite challenging in these children and recovery can be delayed.

Ambulatory anaesthesia

It is best to avoid day-care or ambulatory surgery in paediatric patients with JLS, as there is a possibility of severe peri-operative complications due to multi-system involvement, restrictive

lung disease and neurological defects. There can also be incidences of post-operative apnoeas in preterm children of JLS.

Obstetrical anaesthesia

There has been some recent literature [10] regarding obstetric anaesthesia in patients with JLS. Though there has been no studies regarding fertility in these patients, pregnancy may be possible if patient survives till reproductive age. There are several challenges for obstetric anaesthesia in them. All standard precautions, monitoring and peri-operative care concerns with detailed pre-operative evaluation must be expedited for a safe maternal and foetal outcome. The difficult airway risk associated with the obstetric patient is accentuated in patients with JLS and difficult airway cart needs to be kept ready in all these cases. In view of vertebral anomalies, central neuraxial blockade may be difficult to perform for operative deliveries. Aspiration prophylaxis, adequate pre-oxygenation, meticulous monitoring, expert airway manger, post-operative ICU care and alternative plan of anaesthesia should be expedited on a case-to-case basis.

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