orphanaesthesia

Anaesthesia recommendations for

Sjögren-Larsson Syndrome

Disease name: Sjögren-Larsson syndrome

ICD 10: Q87.1

Synonyms: Fatty aldehyde dehydrogenase deficiency; Fatty acid alcohol oxidoreductase deficiency

Disease summary: Sjögren-Larsson syndrome (SLS) is a rare inherited neurocutaneous disorder resulting from mutations in the ALDH3A2 gene encoding fatty aldehyde dehydrogenase (FALDH), an enzyme responsible for catalysing the oxidation of fatty aldehyde to fatty acid. This mutation leads to the accumulation of fatty alcohols and aldehydes causing abnormalities within the skin, eyes, and brain.

Clinical Features: The classic clinical triad is characterized by ichthyosis, intellectual disability, and spastic diplegia, or less commonly, spastic tetraplegia. The first symptom of SLS is typically generalized ichthyosiform hyperkeratosis, which is often present from birth or early infancy. The skin condition tends to be associated with pruritus, and is prevalent in the neck, trunk, and flexures, sparing the face. Heat intolerance secondary to hypohydrosis has been reported. The neurologic symptoms present as developmental delay in the first one to two years of life, often with delayed motor milestones. Spasticity with weakness in the lower limbs, more than the upper limbs, reflects spastic diplegia and most individuals require assistance with walking. The intellectual disability varies from mild to severe. Seizures occur in about 40% of patients, but are usually well controlled with antiepileptic medications. Language problems prevail and include dysarthria and speech delays. The distinct ophthalmologic finding is crystalline retinopathy which manifests as glistening white dots in the macula, usually in the parafoveal area. Patients often suffer from impaired visual acuity and photophobia. Kyphoscoliosis and short stature have been reported. The prevalence of SLS is around 0.4 in 100,000.

Diagnosis and Treatment: Due to phenotypic variability and presentation following a typical age-dependent pattern, diagnosis can be difficult in young children. The classic triad is often not present until age 3. DNA diagnosis is now preferred, though originally fibroblast cultures were grown from skin biopsies for enzymatic testing. Brain MRI shows periventricular white matter abnormalities, which may represent dysmyelination. There is currently no treatment for SLS other than supportive care which may include topical treatments for ichthyosis, antiepileptic medications for seizures, and surgical procedures or botulinum toxin injections to relieve spasticity. Physical therapy and speech therapy are important to optimize daily functioning.

Medicine is in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnosis is wrong

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

Typical surgery

Common procedures in patients with SLS include MRI, ophthalmologic procedures, tendon lengthening, botulinum injection for spasticity, and orthopaedic procedures for contracture release or scoliosis.

Type of anaesthesia

General anaesthesia, either with volatile anaesthetics or total intravenous anaesthesia, is considered safe. There are no contra-indications to the use of conventional anaesthetic agents.

There are no contra-indications to regional anaesthesia. Regional anaesthesia may be complicated by the presence of contractures or scoliosis.

Necessary additional pre-operative testing (beside standard care)

No additional pre-operative testing is necessary. Assessment of neurologic function and presence of cognitive delays will help determine the need for an anxiolytic premedication.

Particular preparation for airway management

The airway of patients with SLS is typically described as normal.

Particular preparation for transfusion or administration of blood products

No particular preparation necessary for transfusion or administration of blood products.

Particular preparation for anticoagulation

No particular considerations in patients with SLS.

Particular precautions for positioning, transportation and mobilisation

Patients may have joint contractures and care should be given to careful positioning and adequate padding of extremities. Patients with SLS may also have photophobia; therefore, one should remain cognisant of possible aggravation with bright operating room environments.

Interactions of chronic disease and anaesthesia medications

Patients with a history of seizures should have medications reviewed for potential interactions with anaesthetic agents. In individuals with poorly controlled seizures, care should be taken with the use of epileptogenic anaesthetic agents. Neuromuscular blocking agents have been used safely in patients with SLS.

Anaesthetic procedure

Anaesthesia has been safely performed in patients utilizing either a total intravenous anaesthesia technique or with volatile anaesthetics.

Particular or additional monitoring

Due to dermatologic manifestations, monitoring of patients with SLS can be difficult as adhesives do not reliably stay attached to skin. Use of self-adherent bandage wrap to secure electrocardiogram leads, pulse oximeters, and intravenous catheters is helpful. Because the ichthyosis typically spares the face, securing an endotracheal tube or laryngeal mask airway is typically not a problem.

Patients with SLS may have a relative heat intolerance, likely due to hypohydrosis, and therefore temperature should be carefully monitored and care taken not to cause hyperthermia.

Monitoring of neuromuscular function is recommended.

Possible complications

Patients with SLS typically do not have complications from routine anaesthetic medication usage. However, care should be maintained to avoid hyperthermia.

Post-operative care

Postoperative monitoring is dependent upon surgical procedure, but no specific additional monitoring is necessary in patients with SLS.

Disease-related acute problems and effect on anaesthesia and recovery

None reported.

Ambulatory anaesthesia

Ambulatory anaesthesia may be performed in patients with SLS in selected cases.

Obstetrical anaesthesia

No particular concerns for obstetrical anaesthesia. Pregnancy in patients with SLS has not been reported.

References

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