orphananesthesia

Anaesthesia recommendations for patients suffering from

Lamellar ichthyosis

Disease name: Lamellar ichthyosis

ICD 10: Q80.2

Synonyms: Autosomal recessive congenital ichthyosis, bathing-suit ichthyosis (limited to the trunk), LI

The ichthyoses comprise a group of keratinisation disorders characterised by generalised scaling of the skin. Lamellar ichthyosis (LI) is usually manifested at birth as a collodion baby with ectropion/eclabion. Collodion membrane means encased in a tight shiny covering. It is characterised afterwards by non-bullous scaling of the whole integument with variable erythroderma. LI is a major subtype of autosomal recessive congenital ichthyosis with an estimated prevalence of 1 : 200,000–300,000. The most common cause of LI is inactivating mutations in the TGM1 gene, which encodes transglutaminase-1 (TGase1), a key enzyme that cross-links proteins in the cornified cell envelope in the dermis. Production of the cornified cell envelope is one of the essential events during terminal differentiation of epidermal keratinocytes and skin barrier formation. Although life expectancy is normal, those affected with LI have impaired growth because of a defective skin permeability barrier, which results in impaired vitamin D production and increased loss of water and calories.

Medicine in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

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Typical surgery

Orthopaedic surgery, syndactyly repair, cataract surgery.

Type of anaesthesia

There is no definite recommendation for either general or regional anaesthesia. There has been description of anaesthesia in only three cases and total five anaesthetics (general anaesthesia in all five) in English literature. One case has been described where the patient received epidural labour analgesia.

Both intravenous and volatile inhalational anaesthetic agents should be safe.

Necessary additional diagnostic procedures (preoperative)

In addition to a haemogram, serum electrolytes including calcium, protein and albumin levels may be indicated if there is associated malnutrition. Serum parathyroid hormone level, 25-hydroxyvitamin D 3 level may be required to confirm the presence of vitamin D deficiency, but of little interference with anaesthesia. These investigations are not mandatory to anaesthetic care.

Particular preparation for airway management

There are a very few reports of anaesthetic management of patients with LI and only one report of anticipated difficult airway. However, all these patients were younger than 12 years (a 20-month-, a 3-year- and a 12-year-old). Progress of the disease with age and more severe involvement of different tissues may lead to restriction of mouth opening and neck mobility. Therefore, possibility of a difficult airway must be kept in mind and appropriate preparation made, but usually LI patients have no particular airway difficulties.

Particular preparation for transfusion or administration of blood products

Because of malnutrition, there may be anaemia and a smaller total blood volume leading to reduced overall reserve. Thus, there may be increased requirement for blood and blood products for moderate to major surgery.

Particular preparation for anticoagulation

There are no reports to suggest the need for perioperative anticoagulation. However, these patients may be bedridden because of the secondary musculoskeletal deformities necessitating anticoagulation following a major surgery.

Particular precautions for positioning, transport or mobilisation

Because of the associated malnutrition and rickets, there may be severe musculoskeletal deformities with restricted mobility of joints. A careful and gentle approach during transportation is very important to avoid injuries.

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

None reported.

Anaesthesiologic procedure

If LI patients have associated malnutrition (a rare event, because LI is not involving the GI tract, only the epidermis) and consequently, reduced serum protein levels, the free form of drugs used during an anaesthetic, especially those which are highly plasma protein bound, may be high. A careful consideration of the dosage of the drugs is essential because of the possible alteration in pharmaco-dynamics and -kinetics. Similarly, dosage of local anaesthetics should be limited to avoid toxicity.

Because of abnormal keratinisation of the skin and limb deformities, venous access could be difficult. Fixation of the intravenous cannula to the skin is difficult as the normal adhesive plasters do not stick well to the hyperkeratinised skin. Attempt to increase the adhesiveness may be met with difficult removal. Similarly, electrodes of an ECG may not stick well and there may be poor quality of signal because of reduced conductivity.

Using a tube holder is useful to secure the endotracheal tube.

These patients are susceptible to develop perioperative hypo- as well as hyperthermia because of the abnormal keratinisation.

These patients usually develope ectropion, and cataract due to constant exposure to light. Protection of eyes from exposure keratitis and direct trauma, especially during prolonged surgery and procedures done in position other than supine. Replacement tear drops and protective goggles may be required.

Particular or additional monitoring

Monitoring core temperature is very important as these patients are susceptible to both hyper- and hypothermia because of hyperkeratinisation.

Possible complications

Patients with LI are at risk of developing perioperative hypo- as well as hyperthermia.

Fractures during transportation and positioning are possible because of the musculoskeletal deformities and restricted joint movements developing as a consequence of rickets.

They are susceptible to eye injuries due to direct trauma as well as exposure because of the ectropion and inability to close the eyes completely.

Postoperative care

Keep the patient warm by using active heating methods like body warmers and monitor body temperature to maintain normothermia.

These patients may need to be carefully monitored for respiratory depression because of the altered pharmaco-kinetics and -dynamics due to severe malnutrition making them susceptible to overdose of anaesthetics, opioids and neuromuscular blocking agents.

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 disease-triggered emergent situations.

Ambulatory anaesthesia

Procedures of short to moderate duration may be undertaken and managed according to the standard guidelines for ambulatory anaesthesia.

Obstetrical anaesthesia

Patients may require epidural labour analgesia, which may be associated with technical difficulties during the procedure and abnormal spread of local anaesthetic if associated with spinal deformities like kyphoscoliosis.

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Literature and internet links

1. Vahlquist A. Pleomorphic ichthyosis: proposed name for a heterogeneous group of congenital ichthyoses with phenotypic shifting and mild residual scaling. Acta Derm Venereol 2010;90: 454-460
2. Oji V, Traupe H. Ichthyoses: differential diagnosis and molecular genetics. Eur J Dermatol 2006;16:349-359
3. Kubota R, Miyake N, Nakayama H, et al. Anesthetic management of a patient with non-bullous congenital ichthyosiform erythroderma. Masui 2003;52:1332-1334
4. Boku A, Tachibana K, Takeuchi M, Kinouchi K, et al. Anesthetic considerations for a boy with non-bullous ichthyosiform erythroderma. Masui 2011;60:258-261
5. Akiyama M. Harlequin ichthyosis and other autosomal recessive congenital ichthyoses: the underlying genetic defects and pathomechanisms. J Dermatol Sci 2006;42:83–89
6. Hegde HV, Annigeri VM, Pai VV. Anesthetic challenges in lamellar ichthyosis. Paediatr Anaesth 2012;22:492-4
7. Moskowitz DG, Fowler AJ, Heyman MB, Cohen SP, Crumrine D, Elias PM, Williams ML. Pathophysiologic basis for growth failure in children with ichthyosis: an evaluation of cutaneous ultrastructure, epidermal permeability barrier function, and energy expenditure. J Pediatr. 2004;145:82-92
8. Thacher TD, Fischer PR, Pettifor JM, Darmstadt GL. Nutritional rickets in ichthyosis and response to calcipotriene. Pediatrics. 2004;114:e119-23
9. Sathish Kumar T, Scott XJ, Simon A, Raghupathy P. Vitamin D deficiency rickets with Lamellar ichthyosis. J Postgrad Med. 2007;53:215-7
10. Nayak S, Behera SK, Acharjya B, Sahu A, Mishra D. Epidermolytic hyperkeratosis with rickets. Indian J Dermatol Venereol Leprol. 2006;72:139-42.

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