orphanaesthesia

Anaesthesia recommendations for

Lenz-Majewski hyperostotic dwarfism

Disease name: Lenz-Majewski hyperostotic dwarfism

ICD 10: Q87.1

Synonyms: Dystrophinopathy

Disease summary: Lenz-Majewski hyperostotic dwarfism is a very rare syndrome caused by a de novo heterozygous mutation of phosphatidylserine synthase 1 (PTDSS1) gene (Sousa et al. 2013). It was first described by Braham in 1969 as the Camurati-Engelman syndrome but soon later became the Braham-Lenz syndrome until it was re-classified by Lenz and Majewski in 1974. Only thirteen reported cases are described in the Anglo-Saxon literature.

The most important feature is a disproportionately large head in relation to a reduced size trunk and limbs. Large fontanels and widely separated sutures that close lately in childhood are present. The skin is loose, wrinkled, and atrophic with prominent veins, especially in the scalp. The ears are large and floppy, and frequently associated are choanal atresia or stenosis, nasolacrimal duct obstruction, and, in boys, cryptorchidism and inguinal hernia. There is craniofacial dysmorphic evolution with age, leading to progressive macroglossia, prognathism, midface hypoplasia, and upper airway obstruction.

The disorder is also characterized by failure to thrive and severe mental retardation. This condition can be mistaken for craniometaphyseal and craniodiaphyseal dysplasias. However, the absence of cranial nerves impingement confirms the diagnosis. Enlarged ventricles and hydrocephalus have been described in some patients.

The skeletal alterations are striking with the following radiographic features:

– progressive sclerosis of the skull, facial bones, and vertebrae
– broad clavicles and ribs
– short or absent middle phalanges
– diaphyseal undermodeling and midshaft cortical thickening
– metaphyseal and epiphyseal hypostosis
– retarded skeletal maturation

Tooth enamel is also defective.

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

Musculo-skeletal disorders procedures are common.

Type of anaesthesia

There are no specific precautions before anaesthesia except those associated with other medical conditions with the patient. However, because of the nature of the disease, facial dysmorphism is very common, and one must carefully evaluate the airway and plan accordingly for a potential difficult airway management and tracheal intubation.

This disease can involve considerable musculoskeletal and skin features that will require special attention during positioning. Finally, due to mental retardation, the use of a regional anaesthetic technique should be complemented by controlled sedation or general anaesthesia. In both cases, proper monitoring of the airway is mandatory.

No specific anaesthetic considerations are reported for this medical condition besides the usual patient management. However, in presence of a difficult airway, induction of anaesthesia with a volatile anaesthetic technique, maintenance of spontaneous breathing and placement of an intravenous catheter must be performed safely. Tracheal intubation under safe conditions is highly recommended. The use of a laryngeal mask is an alternative only for procedure where access to the airway is always available.

There are no known pharmacological implications for this syndrome.

Neuroaxial or peripheral block is an interesting option for anaesthetic management. The use of ultrasound-guided nerve blocks enhances success rate.

Necessary additional pre-operative testing (beside standard care)

Due to the low incidence of this syndrome and the lack of clinical evidence, no specific recommendation besides the usual preoperative preparation must be done. However, as the child gets older and thoracic deformity worsens, lung function tests might be considered (when feasible) to establish the pulmonary involvement.

Particular preparation for airway management

Not reported.

Particular preparation for transfusion or administration of blood products

Not reported.

Particular preparation for anticoagulation

Not reported.

Particular precautions for positioning, transportation and mobilisation

Positioning might require special attention particularly because of leg problems and the exquisite pain manifested by these patients in the post-operative period.

Interactions of chronic disease and anaesthesia medications

Not reported.

Anaesthetic procedure

There are no pharmacological known implications with this medical condition.

Particular or additional monitoring

Standard monitoring is indicated. The use of invasive monitoring should be dictated by the surgical procedure alone and not the medical condition.

Possible complications

Although they are not directly reported, given facial dysmorphism, potential complications could be related to difficult airway management.

Postoperative exquisite pain has been reported postoperatively and should be an important consideration in the management of these patients.

Post-operative care

The need of postoperative care is related to the kind of surgery and to preexisting co-morbidity.

Disease-related acute problems and effect on anaesthesia and recovery

Not reported.

Ambulatory anaesthesia

There are no contraindications to same-day anaesthesia and surgical procedures. Each patient must be evaluated carefully for co-morbidity and/or airway issues that might suggest that a hospitalization will be necessary for the safety of the postoperative care.

Obstetrical anaesthesia

Not reported.

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Authors

Adrià Font Gual, Anaesthesiologist, Hospital de la Santa Creu i Sant Pau. Barcelona, Spain
AFontG@santpau.cat

Tobias Koller, Anaesthesiologist, Hospital de la Santa Creu i Sant Pau. Barcelona, Spain

Juan Ignacio Casas Vila, Department of Anaesthesiology. Hospital de la Santa Creu i Sant Pau. Barcelona, Spain

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Reviewers

Bruno Bissonnette, Director of Neurosurgical Anesthesia, University of Toronto, Canada
bruno@brainstorm-anesthesia.com

Duang Wattanasirichaigoon, Paediatrician, Ramathibodi Hospital, Mahidol University, Bangkok, Thailand
duangrurdee.wat@mahidol.ac.th

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Tino Münster, Anaesthesiologist, Department of anaesthesiology and intensive care medicine, Hospital Barmherzige Brüder, Regensburg, Germany
Tino.Muenster@barmherzige-regensburg.de

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