orphanaesthesia

Anaesthesia recommendations for patients suffering from

Gomez-Lopez-Hernandez syndrome

Disease name: Gomez-Lopez-Hernandez syndrome (OMIM 601853)

ICD 10: Q07.8

Synonyms: Cerebellotrigeminal dermal dysplasia

Gomez-Lopez-Hernandez syndrome is a rare form of cerebellotrigeminal dermal dysplasia, first documented by Gómez and reported by López-Hernández. It comprises mainly neurological, dysmorphic and cutaneous symptoms. The genetic basis of this sporadic disorder GLH is not yet clear. De novo chromosomal arrangements or spontaneous dominant mutations may represent possible explanations. Identification of this syndrome is possible after birth, in view of rhomboencephalosynapsis, alopecia and trigeminal anaesthesia. Rhombencephalosynapsis (RES), a rare hindbrain malformation, manifesting as a fusion of the cerebellar hemispheres, the dentate nuclei and the superior cerebellar peduncles, is associated with agenesis/ hypogenesis of the vermis. RES may occur in isolation or in conjunction with VACTER-L or as part of Gomez-Lopez-Hernandez syndrome. Anaesthesia considerations include dealing with intellectual, psychiatric and behavioural problems such as hyperactivity, depression, self-injurious behaviour and bipolar disorder. Different degrees of trigeminal anaesthesia leading to recurrent corneal and facial scarring have been described. Parietal or parieto-occipital alopecia is usually bilateral, may be hidden by surrounding scalp hair. These findings are best evaluated and documented prior to initiation of anaesthesia care.

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

Typical surgery

Reported cases are sporadic and depicted at different ages, from the postnatal period until adulthood. Patients may present for different types of surgeries, especially neurosurgery (VP shunt and cranial vault reconstruction for craniosynostosis), ophthalmic surgery (trauma) and plastic surgery.

Type of anaesthesia

Each patient should be evaluated on individual basis. Because of the nature of the disease with possible existence of facial dysmorphism, a careful evaluation and management of a possible difficult airway should be planned. In the presence of a difficult airway, induction of anaesthesia with maintenance of spontaneous ventilation and tracheal intubation under safe conditions is highly recommended.

Possible presence of psychiatric and behavioural problems such as hyperactivity, depression, self-injurious behaviour and bipolar disorder may challenge the anaesthesiologist in dealing with these patients perioperatively. Baseline behavioural status should be reported in the postoperative management to help differentiation with postoperative complications.

Ventriculomegaly and hydrocephalus due to aqueductal stenosis are common, and therefore neurological evaluation to rule out an increase in intracranial pressure is important during the assessment of these patients.

This syndrome can involve parietal or parieto-occipital alopecia and degrees of trigeminal anaesthesia. These should be documented prior to surgery and anesthesia with special attention during positioning including measures for eyes protection, especially for lengthy cases.

Necessary additional diagnostic procedures (preoperative)

Findings to be considered and documented prior to initiation of anaesthesia care: Ventriculomegaly and hydrocephalus. Different degrees of trigeminal anaesthesia. Parietal or parieto-occipital alopecia.

Baseline mental and intellectual impairment should be assessed. Psychiatric and behavioural problems such as hyperactivity, depression, self-injurious behaviour and bipolar disorder. Full neuro examination with regions of anesthesia are important, as is developmental assessment. The performance of patients with GLH can range from moderate-severe impairment to cognitively normal.

Particular preparation for airway management

Careful evaluation for difficult tracheal intubation secondary to dysmorphic features. Patients tend to have midfacial retrusion so the facial structures will be different when compared to each other.

Muscular hypotonia may precipitate upper airway obstruction.

Particular preparation for transfusion or administration of blood products

Not reported.

Particular preparation for anticoagulation

Not reported.

Particular precautions for positioning, transport or mobilisation

The presence of alopecia and trigeminal anaesthesia should be documented and taken into consideration when caring for these patients.

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

There are no known special pharmacological implications for this syndrome.

Anaesthesiologic procedure

Consideration and preparation for a possible difficult airway.

Possibility of neurological pathologies (such as intracranial hypertension).

Particular or additional monitoring

Individual basis if presence of intracranial hypertension.

Possible complications

Special attention to the following points: Neurological impairment/trigeminal anaesthesia, behavioural problems and a potential difficult airway.

Postoperative care

Documentation and stabilisation:

Psychiatric and behavioural problems such as hyperactivity, depression, self-injurious behaviour and bipolar disorder.

Ventriculomegaly and hydrocephalus usually due to aqueductal stenosis are common. Different degrees of trigeminal anaesthesia leading to recurrent corneal and facial scarring.

Parietal or parieto-occipital alopecia.

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

Not reported.

Ambulatory anaesthesia

Each patient must be evaluated carefully for co-morbidity and/or airway issues. Anaesthesia and surgery to be performed in a medical facility with capacity of taking care of potential complications.

Obstetrical anaesthesia

Anaesthesia and surgery to be performed in a medical facility with capacity of taking care of potential complications. The etiology or transmission of this condition is actually unknown and obstetrical management is speculative since no patients with GLH are known to have reproduced.

Literature and internet links

1. Gómez MR. Cerebellotrigeminal and focal dermal dysplasia: a newly recognized neurocutaneous syndrome. Brain Dev. 1979; 1(4):253-6
2. López-Hernández A. Craniosynostosis, ataxia, trigeminal anaesthesia and parietal alopecia with pons-vermis fusion anomaly (atresia of the fourth ventricle). Report of two cases. Neuropediatrics. 1982; 13(2):99-102.
3. Poretti A, Bartholdi D, Gobara S, et al.: Gomez-Lopez-Hernandez syndrome: An easily missed diagnosis. Eur. J. Med. Genet. 2008; 51(3):197-208
4. Sukhudyan B, Jaladyan V, Melikyan G, et al.: Gomez-Lopez-Hernandez syndrome: reappraisal of the diagnostic criteria. Eur. J. Pediatr. 2010; 169(12):1523-1528
5. Abdallah C.: Anaesthesia and orphan disease: Anaesthesia for Gomez-Lopez-Hernandez syndrome. Eur. J. Anaesthesiol. 2015; 32(3):218-20.

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These guidelines have been prepared by:

Author
Claude Abdallah, Anaesthesiologist, Children's National Mecial Center, Washington, USA
cabdalla@cnmc.org

Peer revision 1
Eric T. Rush, Assistant Professor of Pediatrics and Internal Medicine, Munroe-Meyer Institute for Genetics and Rehabilitation, University of Nebraska Medical Center Omaha, Nebraska USA
erush@unmc.edu

Peer revision 2
Eugen Boltshauser, Neuropaediatrican, Neuromuscular Centre Zurich, University Hospital and Childrens' Centre Zurich, Switzerland
Eugen.Boltshauser@kispi.uzh.ch

Please note that this guideline has not been reviewed by an anaesthesiologist but by two disease experts instead.