

Anaesthesia recommendations for **Hamamy syndrome**

Disease name: Hamamy syndrome

ICD 10: ./.
OMIM 611175

Synonyms: Craniofacial dysplasia-osteopenia syndrome

Disease summary: Hamamy Syndrome (HS) is a very rare disease initially described by Hamamy et al. HS results from a congenital homozygous mutation of the IRX5 gene on chromosome 16q12.2 [1]. At present, only 5 patients have been reported in the literature and one of them deceased. Mutations in the IRX5 homeobox cause a recessive congenital disorder affecting the face, brain, erythrocytes, heart, bone and gonadal development [2]. This suggests that IRX proteins may be crucial for the ontogeny and function of many organs both in new-borns and adults [2]. This syndrome is characterized by craniofacial dysmorphism including midface prominence (“bulging”), upslanting palpebral fissures with sparse lateral eyebrows, severe telecanthus, lacrimal-salivary apparatus agenesis, fronto-nasal abnormalities, thin upper vermillion border, protruding ears, myopia, mental retardation, sensorineural hearing impairment, enamel hypoplasia or hypodontia, gonadal anomalies, sloping shoulders, low posterior hairline, congenital heart anomalies (VSD, atrioventricular canal) with intraventricular conduction anomalies, pectus excavatum, hypochromic microcytic anaemia, and skeletal abnormalities of the long bones with recurrent fractures [2]. Anaemia, difficult airway and cardiac pathologies are the possible difficulties in the anaesthesia management of HS patients.

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

Orthopaedic surgery (correction osteotomies for multiple fractures), dental surgery and hernia repair.

Type of anaesthesia

There is no definite recommendation for either general or regional anaesthesia for HS patients, and there is only one case report on anaesthesia management in the literature [3]. The risk of malignant hyperthermia should be similar as in the normal population. HS involves multiple systems, clinical judgement regarding the type of anaesthesia can be made on a case-by-case evaluation.

Necessary additional pre-operative testing (beside standard care)

Preoperative evaluation of the patient's airway, cardiac, and haematological functions are crucial. We suggest preoperative ECG evaluation in order to detect possible cardiac and conduction anomalies. Haematological evaluation is also important because anaemia and blood anomalies can be expected in HS patients, and chest X-ray.

Particular preparation for airway management

The presence of craniofacial dysmorphism should always suggest difficulties in airway management (both ventilation with a facial mask and tracheal intubation) and techniques for difficult airway should be readily available. In the only published anaesthesia case report, mask ventilation was facilitated with the aid of an oropharyngeal airway and tracheal intubation was successful with direct laryngoscopy [3].

Particular preparation for transfusion or administration of blood products

HS patients may need transfusion for their operations (preoperative anaemia and/or surgical bleeding). Perioperative anaemia is a major concern mostly in orthopaedic surgery [4]. In paediatric surgery, ESA guidelines should be followed taking into account the presence of a cardiac anomaly [5]. Our suggestion is to correct anaemia in the preoperative period in case of elective surgery, and that one should be prepared for transfusion in case of urgency [3].

Particular preparation for anticoagulation

There is no evidence to support prophylactic anticoagulation. It should be considered according to the type of surgery, length of postoperative immobilization, and patient's age.

Particular precautions for positioning, transportation and mobilisation

Positioning requires caution because HS patients have severe osteoporosis, and the anaesthesiologist should be aware of possible fractures.

Interactions of chronic disease and anaesthesia medications

Not reported.

Anaesthetic procedure

Non-depolarizing neuromuscular blocking agents can be used safely [3]. Anaesthesiologist should show vigilance for possible arrhythmias. There is no evidence for any anaesthetic drug that may be insecure. Prophylactic postoperative ventilation is generally not necessary for routine cases.

Particular or additional monitoring

We strongly suggest monitoring body temperature to avoid shivering, because increased oxygen demand may be dangerous for cardiac HS patients.

Possible complications

Cardiac and haematological complications are possible.

Post-operative care

Unless required by the surgical procedure, intensive care in the postoperative period is not mandatory. We suggest ECG monitoring and haemoglobin level follow-up in the first post-operative 24 hours.

Disease-related acute problems and effect on anaesthesia and recovery

Disease triggered emergency-like situations are not common in HS patients.

Ambulatory anaesthesia

Dental procedures may be performed with ambulatory anaesthesia. We do not recommend orthopaedic surgeries with ambulatory anaesthesia.

Obstetrical anaesthesia

There are 4 HS patients alive and 1 of them is female. Since the syndrome is associated with abnormal reproductive cells, fertility at the moment is indeterminate.

References

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Please note that this recommendation has not been reviewed by an anaesthesiologist and a disease expert but by two anaesthesiologists instead.
