orphanaesthesia

Anaesthesia recommendations for

Nager syndrome

Disease name: Nager syndrome

ICD 10: Q75.4
OMIM: 154400

Synonyms: Acrofacial dysostosis 1 (AFD1), Nager acrofacial dysostosis, Preaxial acrofacial dysostosis, Mandibulofacial dysostosis with preaxial limb anomalies

Disease summary: Nager syndrome is a rare syndrome that has an unknown prevalence. It shares some phenotypic features of Treacher-Collins syndrome. The literature describes sporadic cases (de novo mutation) as well as autosomal dominant or recessive and spontaneous inheritance patterns. It is caused by a genetic mutation in the SF3B4 gene (1q21.2), preventing the function of SAP49 protein, which is important for bone and cartilage maturation. This results in abnormal development of the 1st and 2nd brachial arches and limb buds. Clinical manifestations include: craniofacial malformations (malar hypoplasia, micrognathia, cleft palate, down-slanting palpebral fissures, absent eyelashes in the middle of the eyebrows, lower eyelid coloboma, external auditory defects, conductive hearing loss, choanal atresia) and pre-axial limb malformations predominately of the upper limb (hypoplastic/absent thumbs, clinodactyly or syndactyly, shortened or absent forearm, shortened humeral bone) and, less commonly, lower limb abnormalities. More rarely is an involvement of the heart (Fallot tetralogy, ventricular septal defect, atrial septal defect, patent ductus arteriosus), kidneys, genitalia and urinary tract. It is associated with normal cognitive function.

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

Craniofacial surgery, mandibular and/or midface advancement and distraction osteogenesis, temporo-mandibular joint reconstruction, genioplasty, cleft palate repair, orthognathic procedures; tracheostomy; gastrostomy insertion; limb surgery.

Type of anaesthesia

There is no recommendation for either general or regional anaesthesia. Caution should be utilized when considering sedation as it is associated with obstructive sleep apnoea and difficult airway. Difficult intravenous access is described.

Necessary additional pre-operative testing (beside standard care)

Due to the possible association with congenital cardiac disease and possible development of cor pulmonale in case of poorly managed OSAS, an echocardiogram should be performed. Genetic mutations with craniofacial manifestations can be associated with cervical spinal anomalies, so one author recommends preoperative radiological evaluation of the cervical spine. Choanal atresia should be evaluated: permeability of nasal passages, previous endo-nasal surgery.

Particular preparation for airway management

Meticulous pre-operative airway assessment is necessary. This syndrome is associated with difficult airway management due to midface hypoplasia, limited temporo-mandibular mobility or even ankylosis, and mandibular hypoplasia. Emergency and elective tracheostomy has been described as well as death from airway obstruction. A range of difficult airway equip-ment should be available. Intubation techniques using a laryngeal mask to facilitate fibreoptic intubation under anaesthesia and use of a C-MAC adult blade D partially inserted into the mouth has been described with success. Appropriate mask fit and bag mask ventilation can be difficult due to the association with cleft palate with midface deformities. Retrograde intubation has been described with success. Post-operative airway obstruction should be anticipated: in the absence of choanal atresia, inserting a nasopharyngeal airway before extubation is very helpful. Due to swallowing abnormalities they may be at increased risk of aspiration, however, most will have had a gastrostomy at an early stage.

Particular preparation for transfusion or administration of blood products

There is no evidence to support a difference in haematological marker or a difference in the administration of blood products.

Particular preparation for anticoagulation

There is no recommendation for the need for particular anticoagulation.

Particular precautions for positioning, transportation and mobilisation

Positioning may require extra support to prevent excessive stress on supporting joints to avoid neurovascular injury.

Interactions of chronic disease and anaesthesia medications

Not reported.

Anaesthetic procedure

This is a potential difficult oxygenation/difficult intubation situation: spontaneous breathing should thus be maintained until the airway is secured. Safe use of a pre-medication has been described in two cases without any consequence. Nitrous oxide and volatile agents have been used without complication. No reports on intravenous induction (with propofol, ketamine or dexmedetomidine) have been published so far. An opiate-sparing strategy may be advised due to the risk of post-operative airway obstruction. No interaction with non-depolarizing or depolarizing muscle relaxants has been published so far. There is no contra-indication to the use of local anaesthetic agents. Intra-operative use of volatile agents has been described without complication. Patients should be extubated fully awake. As residual anaesthesia can exacerbate airway obstruction, inserting a nasopharyngeal airway before extubation is very helpful. Everything should be ready for a difficult re-intubation especially if surgery involved the upper airway: inserting a Cook exchange catheter into the trachea before extubation should be considered in order to railroad the tracheal tube over it for re-intubation.

In case of major upper airway surgery, delayed extubation in the presence of an ENT surgeon (in the ICU or operating room) should be considered.

Particular or additional monitoring

No additional monitoring is described.

Possible complications

Sedative medications can lead to airway obstruction, it so should be used with caution.

Post-operative care

Post-operative airway obstruction can occur secondary to microstomia, micrognathia, restricted temporo-mandibular mobility and mandibular hypoplasia. Inserting a nasopharyngeal airway prevents upper airway obstruction. Monitoring is recommended.

Disease-related acute problems and effect on anaesthesia and recovery

No emergency-like situations have been reported.

Ambulatory anaesthesia

Due to potential post-operative airway obstruction, ambulatory anaesthesia is not recommended.

Obstetrical anaesthesia

If a prenatal diagnosis has been established, an EXIT procedure has been described as part of securing a definitive airway in newborns with severe upper airway obstruction.

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