orphanaesthesia

Anaesthesia recommendations for

Oculo-ectodermal syndrome

Disease name: Oculo-ectodermal syndrome

ICD 10: -

OMIM: 600268

Synonyms: Aplasia cutis congenita-epibulbar dermoids syndrome

Disease summary: Oculo-ectodermal syndrome (OES) is an extremely rare disease (< 1:1,000,000) first described in 1993 by Toriello et al. Approximately 20 cases have been described in the literature. OES is a mosaic developmental pathology recognised by its typical oculo-cutaneous manifestations. The aetiology of OES is post-zygotic missense variants in the proto-oncogene KRAS (on chromosome 12p12.1), in the RAS/MAPK signalling pathway. In at least 7 individuals, post-zygotic variants in the proto-oncogene KRAS, in the RAS/MAPK signalling pathway, have been identified. Many of these involve codon p. Ala146. OES is thought to be a mosaic RASopathy and shares many features with other syndromes of this group (Encephalo-cranio-cutaneous lipomatosis, Linear sebaceous nevus syndrome). The variable severity of this disease is explained by the precocity of the mutational event leading to a higher mutation burden. The hallmark clinical signs are a combination of congenital scalp lesions, referred to as aplasia cutis congenita and epibulbar dermoids. Due to mosaicism, there is a large variability in the phenotypic expression of the syndrome. They are:

- Hamartomas (myxovascular hamartoma, smooth muscle hamartoma) associated with regions of alopecia,
- Long bone non-ossifying fibromas and giant cell granulomas of the jaws have repeatedly been observed and seem to be age-dependent, becoming a common manifestation from the age of 5 years,
- Growth failure,
- Skeletal abnormalities: skull abnormality, length discrepancy in long bones, short neck, facial asymmetry,
- Ocular anomalies: defects or skin tags of the upper eyelid, corneal opacity, optic nerve or retinal abnormality, microphthalmia and glaucoma,
- Urogenital defects: bladder exstrophy, epispadias,
- Arachnoid cysts in the brain,
- Lymphoedema,
- Cardiovascular anomalies: hypertrophic cardiomyopathy, atrial septal defect, persistent ductus arteriosus and aortic coarctation,
- Moyamoya disease,

- Neurodevelopment symptoms can include developmental delay, learning difficulties, behavioural abnormalities or epilepsy.

An increased risk of malignancy cannot be excluded in particular for rare specific variants, but so far there is insufficient evidence for a recommendation to screen for specific tumours. Consequently, regular clinical follow-up should be recommended for an early oncologic diagnosis. So far, there is little data on disease progression, malignancy risk or RAS-targeted therapies in OES and, for that reason, future research is required.

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

The phenotypic expression is highly variable.

Surgical options might include removal of epibulbar dermoids and skin grafting in aplasia cutis congenita. Additional ocular anomalies, such as skin tags of the upper eyelid, may be corrected. Surgical removal of benign tumour-like lesions such as non-ossifying fibromas of the long bones and giant cell granulomas.

Congenital upper eyelid colobomas with corneal ulcer or with impending perforation are a non-traumatic oculoplastic emergency that may be present in the first few days of life.

Multi-disciplinary approach is advisable to provide the best possible outcomes.

Type of anaesthesia

There are no reports of any recommendations for either general or regional anaesthesia.

Based on the limited data available, there seem to be no contraindications to general, monitored anaesthetic care (MAC), regional or neuraxial anaesthesia.

The type of anaesthesia should be patient-tailored based on their age, co-existing diseases and surgery risk.

Most of the patients will probably be in the paediatric age, so patient safety and paediatric anaesthesia still require a special understanding of anatomic, psychological and physiological development.

Sedation might be required for diagnostic procedures.

Necessary additional pre-operative testing (beside standard care)

Due to the variability of the phenotypic expression, these patients can have multiple underlying defects (for instance skeletal, cardiovascular or soft tissue) so further work-up should be considered on a case-by-case basis, depending on the location, size of the defect and other clinical findings. As the possibly associated cardiac defects are generally paucisymptomatic, these children should have at least one echographic cardiac examination in their files.

Particular preparation for airway management

Optimise oxygenation, airway management, and tracheal intubation keeping in mind the challenges of different patient presentations. In case of Moyamoya disease, both hypo- and hypertension as well as hypo- and hypercarbia should be avoided.

Particular preparation for transfusion or administration of blood products

Based on the limited data available, there is no evidence of platelet function deficiency,

abnormal red blood cell counts, and altered coagulation in this population. Therefore, normal transfusion standards and guidelines should be followed.

Particular preparation for anticoagulation

Based on the limited data available, there is no evidence for particular anticoagulation therapy.

Particular precautions for positioning, transportation and mobilisation

Eyes seem to be particularly susceptible to damage. It is important to lubricate the eyes and ensure they are also taped and padded.

Interactions of chronic disease and anaesthesia medications

Not reported.

Anaesthetic procedure

From our experience with a 7-year-old girl undergoing a surgical eye biopsy, the airway management was uneventful and there were no complications with the use of opioids (fentanyl), intravenous anaesthetic (propofol), inhalational anaesthetic (sevoflurane) for the induction and maintenance, dexamethasone and acetaminophen.

At one month of age, the patient had presented for surgical repair of a unilateral lid coloboma. This defect was repaired under general anaesthesia with inhalational anaesthetics (sevoflurane) uneventfully.

Particular or additional monitoring

Standard anaesthesia monitoring, which includes ECG, NIBP, pulse oximetry, temperature, anaesthetic gases analysis and end-tidal CO2.

Possible complications

Not reported.

Post-operative care

The degree of post-operative monitoring and care depends on the patients´ pre-operative condition and the surgical procedure.

Disease-related acute problems and effect on anaesthesia and recovery

Not reported.

Ambulatory anaesthesia

Ambulatory surgery should be tailored for the individual patient based on their age, co-existing problems and the risk of surgery.

Obstetrical anaesthesia

Not reported.

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