

# orphananesthesia

## Anaesthesia recommendations for patients suffering from **Apert syndrome**

**Disease name:** Apert syndrome

**ICD 10:** Q87.0

**Synonyms:** ACS 1, acrocephalosyndactyly type 1

Apert syndrome was named after the French paediatrician Eugène Apert, who first described the collection of signs in 1906. It is a congenital disease that is a form of acrocephalosyndactyly, and is characterized by malformations of the skull, hands, feet and face.

It is a rare disease with an incidence of around 1 per 160,000 live births. It is an autosomal dominant complaint and affects both females and males equally. Interestingly however the vast majority of cases are due to sporadic mutations, but there is an association with increased paternal age.

It is thought that the affected chromosome is chromosome 10, and there are two main identified gene defects affecting fibroblast growth factor receptor 2 gene. The resultant abnormal receptor prevents apoptosis of cells, and so in the case of those affected by Apert syndrome, digits on both hands and feet may be fused. These fusions can be either cutaneous or bony. Receptors found in the cranium are also affected and thus cause premature fusion of sutures resulting in craniosynostosis.

Apert syndrome is always apparent at birth. This is due to the characteristic hand and foot deformities, although the facial deformity may be less obvious in some cases.

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Medicine in progress



Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

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The syndrome is characterized by a number of typical features. Firstly it accounts for 4.5% of all craniosynostosis cases. Premature fusion of suture lines can be of both the skull and the face, and these can be variable in number and sites affected. Where there is fusion of sutures there is reduced bone growth, and this is compensated for in areas where there is no fusion. Thus Apert children tend to have characteristic shaped skulls. Brachycephaly is common, this is where the coronal sutures fuse prematurely resulting in a reduced distance from the front of the skull to the back. Usually both coronal sutures are affected. The severity and rate of progression of the skull will depend on the sutures affected.

Other facial characteristics include mid-face hypoplasia, hypertelorism and choanal stenosis. If growth of the brain surpasses skull development, then this will ultimately lead to an increase in intracranial pressure. Another result of abnormal skull development is that the orbits tend to be shallow resulting in protrusion of the eyes, and sometimes inability of the lids to close. This is known as exorbitism.

Other characteristic features include a multi-digit hand and foot syndactyly, which is bilateral, but not always symmetrical. Children with Apert syndrome are also described to have other associated anomalies. These include cardiac defects, polycystic kidneys and pyloric stenosis, all of which are rare.

The major anaesthetic concerns relate to the airway. Bag mask ventilation can sometimes be difficult due to mid-face hypoplasia. Often these children suffer from obstructive sleep apnoea, and therefore are prone to becoming obstructed on induction and emergence. This is usually easily alleviated by the use of airway adjuncts. Some Apert children may also suffer from central apnoea.

It has been reported that these children suffer a higher incidence of bronchospasm, although this was only detailed in one paper. Children with Apert syndrome have profuse secretions that may cause wheeze and contribute to an increase in airway irritability. There is an association with fusion of the cervical vertebra (C5/6), however this does not compromise intubation.

Another challenge for the anaesthetist is intravenous access. This can be made more difficult when one or more limbs are being operated on. As these children may return to theatre for repeat procedures this issue can become increasingly more problematic. As a result, some anaesthetists feel that for short procedures, e.g. change of dressings or CT scans, intravenous access is not mandatory. In an emergency an intra-osseous needle may be placed, or intramuscular drugs can be used.

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#### Typical surgery

Syndactyly release; craniosynostosis; dental surgery; orthopaedic procedures; change of dressings (COD); fronto-orbital correction.

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#### Type of anaesthesia

There is no definite recommendation for general or regional anaesthesia. There are no known contraindications to specific anaesthetic drugs known. Techniques can involve both general and regional techniques.

However, as the incidence of obstructive sleep apnoea (OSA) is high in this group of patients, it is advisable to employ regional techniques where possible in order to avoid intra and post-operative opiates, and therefore reduce the risks of obstruction and apnoea postoperatively. The option of doing unilateral, rather than simultaneous bilateral hand surgery also takes this issue into account.

Regional anaesthesia may sometimes pose a challenge to the anaesthetist, as the anatomy of the shoulder in an Apert child may not necessarily be normal, and can be associated with reduced abduction in the teenage years and older.

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#### **Necessary additional diagnostic procedures (preoperative)**

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If clinically symptomatic, a sleep study may be necessary. An ECG and echocardiography may be warranted if cardiac signs and symptoms are present

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#### **Particular preparation for airway management**

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Atropine has been suggested as a premedication in order to reduce secretions but this is probably unnecessary. As some of these children may have developmental delay an anxiolytic may be considered, although this should be measured against the risk of potential airway obstruction.

Bag-mask ventilation may be difficult due to mid-face hypoplasia. This is easily overcome with an oral airway but there may be difficulty in achieving sufficient depth of anaesthesia for one to be accepted. It may be helpful to use a two-person technique. Children with Apert syndrome do not generally have an increased risk of difficult laryngoscopy and intubation however.

Due to the increased incidence of supraglottic obstruction, a nasopharyngeal airway may be useful in the immediate postoperative period, although it may be difficult to place due to reduced nasopharyngeal volume. If the child is CPAP/BiPAP dependent, then they must be in an appropriate ward setting to accommodate this.

If the child has undergone mid-face advancement or has a distraction frame applied, airway management may be more challenging.

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#### **Particular preparation for transfusion or administration of blood products**

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There is no evidence to demonstrate any specific issues related to blood product administration.

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#### **Particular preparation for anticoagulation**

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There is no evidence for any particular anticoagulation regime in this set of patients. There is nothing to suggest that they are at an increased risk of deep vein thrombosis.

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#### Particular precautions for positioning, transport or mobilisation

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Eyes are particularly susceptible to damage due to inadequate lid closure. It is important to lubricate the eyes, and ensure they are also taped and padded. There are multiple limb abnormalities and care must be taken to avoid pressure points.

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

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Not reported.

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#### Anaesthesiologic procedure

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Avoid excessive use of opiates due to increased risk of obstructive sleep apnoea in this group of patients. Therefore, as far as possible, use regional techniques to reduce opiate use.

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#### Particular or additional monitoring

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The nature of the surgery will dictate the degree of invasive monitoring. Craniocynosis surgery will require invasive arterial monitoring and possibly central venous access.

Apert patients tend to sweat a lot, and unlike other patients undergoing syndactyly surgery do not appear to need warming when undergoing peripheral surgery to their limbs. If they are actively warmed there is a risk of pyrexia, so temperature should always be monitored.

Postoperatively, it is important to monitor for signs of airway obstruction.

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#### Possible complications

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Sedative drugs will potentially cause upper airway obstruction.

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#### Postoperative care

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Due to the increased incidence of supraglottic obstruction, a nasopharyngeal airway may be useful in the immediate postoperative period (see above). If the child is CPAP/BiPAP dependent, then the child must be sent to an appropriate ward setting to accommodate this.

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#### Information about emergency-like situations / Differential diagnostics

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*caused by the illness to give a tool to distinguish between a side effect of the anaesthetic procedure and a manifestation of the disease*

This will be dictated by the nature of the surgery. In cases where the child is intubated, a sudden increase in airway pressures may be the result of blockage of the endotracheal tube

by mucous plugs and secretions. There are no other common disease-related emergency-like situations that may arise.

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#### **Ambulatory anaesthesia**

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As Apert children are at an increased risk of obstructive sleep apnoea, the decision to undergo ambulatory surgery should be a measured one, but dressing changes and minor peripheral surgery not requiring opiate administration may be conducted on a day stay basis.

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#### **Obstetrical anaesthesia**

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There is no evidence of increased risk involved in the obstetric patient.

### Literature and internet links

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