

# orphanainesthesia

Anaesthesia recommendations for patients suffering from

## Pfeiffer syndrome

**Disease name:** Pfeiffer syndrome

**ICD 10:** Q87.0

**Synonyms:** ACS5, Acrocephalosyndactyly type V, noack syndrome, cranio-facial-dermatological dysplasia

Pfeiffer syndrome is a rare autosomal dominant disorder, characterized by malformations of the skull, face, hands and feet. Crouzon, Apert and Pfeiffer syndromes are the most recognizable of the syndromic craniosynostoses. Diagnoses can be established from the typical phenotype accompanied by molecular genetic testing.

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Medicine in progress



Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

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Find more information on the disease, its centres of reference and patient organisations on Orphanet: [www.orpha.net](http://www.orpha.net)

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## Disease summary

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Most cases can be attributed to mutations in the fibroblast growth factor receptor genes FGFR-1 or FGFR-2. However there are phenotypes that cannot be related to FGFR abnormalities. Based on the severity of malformations it is divided into three clinical subtypes. Type 1 “classic” Pfeiffer syndrome is by far the most common subtype and presents with mild manifestations of brachycephaly, midface hypoplasia and broad fingers and toes; it is associated with normal intelligence and generally good outcome. Type 2 consists of cloverleaf skull, extreme proptosis, finger and toe abnormalities, elbow ankylosis or synostosis, airway malformations, developmental delay, neurological complications and decreased life span. Type 3 is similar to type 2 but without a cloverleaf skull. Clinical overlap between the three types may occur.

Craniosyostosis (premature fusion of one or more of the cranial sutures) most often involves coronal and lambdoid sutures. This prevents further growth of the skull, affects the shape of the head and face and may lead to increased intracranial pressures. Possible atresia of the external auditory canal will lead to conductive hearing loss. Fused vertebrae, Arnold-chiari malformation and seizures may be present. The underdeveloped maxillary results in very shallow orbitae and proptosis, which may lead to corneal damage from xerophthalmia.

Maxillary hypoplasia also results in a small nasopharynx and hypopharynx, which may restrict air passage, cause obstructive sleep apnoea, restrict the passage of food into the oesophagus and lead to gastric reflux and possibly recurring aspirations. Cleft palate and choanal atresia are common. In the rare cases of ACS5 Type 2 and 3, extensive airway affections such as tracheal stenosis, tracheo-cartilagenous sleeve and tracheobronchial malacia have been reported.

Hand and feet involvement may range from broad and radially deviated thumbs and big toes to syn- and brachydactyly.

Even though rare, the spectrum of internal organ anomalies is broad and may comprise congenital cardiac malformations.

The prognosis depends mainly on the severity of the associated anomalies of central nervous and respiratory systems. Patients with type 1 PS generally have good prognosis whereas type 2 and 3 PS is usually associated with early demise in infancy or childhood. An early and coordinated multidisciplinary medical and surgical approach is indicated.

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## Typical surgery

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- Decompressive craniectomy
- Decompressive skull remodeling
- Skull expansion surgery
- Midfacial advancement
- Naso-maxillary-zygomatic complex advancement
- CAVE: Extensive surgical trauma, tertiary level care required!

- Syndactyly release of fingers and toes.
- Orthodontic surgery
- Ocular surgery e.g. strabismus and retinal surgery

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### **Type of anaesthesia**

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Anaesthesia can involve both general and regional techniques. There are no known contraindications to specific anesthetic drugs or procedures.

Patients may have an increased risk of regurgitation, hence appropriate precautions should be taken. Since they have an increased risk of OSA (obstructive sleep apnoea), regional anaesthetic techniques are preferable to avoid opiate-induced postoperative obstruction and apnoea. Given that most major surgery will require opiates, these patients should be sent to an High Dependency Unit or Intensive Care Unit post-operatively.

However, regional anaesthesia may be difficult due to multiple limb abnormalities and associated difficult positioning.

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

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Pre-existing visual/hearing loss needs to be evaluated.

Neurosurgical assessment may be required in patients with features suggestive of raised ICP (intracranial pressure) to avoid damage to brain and eyes.

A thorough history and examination should be done to rule out specific organ dysfunction (especially congenital cardiac anomalies). If the patients are clinically symptomatic, then appropriate tests such as sleep study, ECG and/or echocardiography may be warranted.

No recommendation can be made for or against routine testing of all patients with Pfeiffer syndrome. A preoperative hematocrit and type and cross match should be performed for reconstruction procedures expected to cause significant blood loss.

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

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Patients may have a difficult bag and mask ventilation due to mid face hypoplasia or secondary nasal obstruction due to deviated nasal septum and choanal atresia. Snoring and the presence of obstructive sleep apnoea may suggest airway obstruction. The obstruction may be overcome with the use of an oropharyngeal airway device.

Tracheal anomalies such as tracheal cartilaginous sleeve and laryngeal web have also been reported which may lead to respiratory distress. Moreover, these patients may have reduced neck movement due to cervical fusion.

Children with Pfeiffer syndrome can have an increased risk of difficult laryngoscopy and intubation. Preparations for a difficult airway are advisable.

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

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No data available.

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

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No data available.

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

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Patients with Pfeiffer syndrome may have multiple limb abnormalities and care should be taken to avoid pressure damage during transportation and positioning for surgeries. Eyes are susceptible to damage due to inadequate lid closure and should be lubricated, taped and padded.

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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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IV cannulation may be difficult due to limb abnormalities.

Avoid benzodiazepines and long-lasting opioids due to increased risk of OSA. Consider the use of shorter acting agents, such as desflurane, propofol and remifentanyl, to facilitate a reliable return of airway reflexes and spontaneous ventilation at the end of the procedure. Sedative drugs may increase postoperative upper airway obstruction.

Supplement general anaesthetics with local anaesthetics whenever possible to reduce opioid requirements.

In the case of increased intracranial pressure the use of Nitrous Oxide is not advised. Any further increase in ICP should be avoided.

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#### **Particular or additional monitoring**

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The degree of monitoring will depend on the nature of surgery and possible cardiac and respiratory malformations. One should be vigilant for any signs of airway obstruction.

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### **Possible complications**

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- Potentially difficult ventilation and intubation. Intubation is generally more difficult after mid-face advancement. Emergency tracheostomy for airway obstruction.
- Increased risk of regurgitation and subsequent aspiration.
- Patients may have conductive hearing loss; communication may be difficult.
- Prepare for seizures

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### **Postoperative care**

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- An oro- or nasopharyngeal airway may be required to relieve airway obstruction. There is an increased risk of CPAP/ BiPAP requirements in the immediate postoperative period.
- Follow-up care in an ICU or HDU environment must be taken into consideration.

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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*

These patients may require urgent surgical intervention due to raised ICP.

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### **Ambulatory anaesthesia**

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These patients have an increased risk of difficult ventilation and intubation and also increased risk of obstructive sleep apnoea. Thus ambulatory cannot be recommended. Only minor procedures may be considered on a day care basis.

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### **Obstetrical anaesthesia**

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No data available.

## Literature and internet links

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