orphananesthesia

Anesthesia recommendations for

Pfeiffer syndrome

Disease name: Pfeiffer syndrome

ICD 10: Q87.0

ORPHAcode: 710

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

Disease summary: 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 based on the typical phenotype, supported by molecular genetic testing.

Diagnosis may be incorrect; if uncertainty exists, the diagnosis should be re-evaluated.

Every patient is unique; individual circumstances must always guide clinical care.

Medicine is in progress; new clinical knowledge may not be yet reflected in this guideline Perhaps new knowledge.

Recommendations are not rules or laws; they provide a framework to support clinical decision-making. Although this recommendation has passed a structured review process, it does not meet the formal criteria of a guideline.

Translations may not always reflect the most recent updates of the English version.

Find more information on the disease, its centers of reference and patient organizations on Orphanet: www.orpha.net

A	**AIRWAY / ANESTHETIC TECHNIQUE**	Airway obstruction is common following induction of anesthesia. Maintain spontaneous ventilation till airway is secured. Smaller size ETT may be required. Avoid neck movement during intubation. Avoid nasopharyngeal airway. Plan extubation. Tracheostomy is common and you need to manage patients with tracheostomy for various procedures Both GA/RA can be used as per need.
B	**BLOOD PRODUCTS (COAGULATION)**	No Special preparation / storage of blood products needed. If there is a bleeding history, advanced tests are recommended (e.g., thromboelastography, coagulation factors).
C	**CIRCULATION**	Not a primary cardiac disorder. Congenital heart defects like VSD, ASD, PDA etc. may occur as a part of multisystem disorder.
D	**DRUGS**	Cautious use of sedatives like opioids, benzodiazepines due to increased risk of airway obstruction. Avoid drugs that may increase ICP like nitrous oxide and high dose ketamine which may increase airway secretions.
E	**EQUIPMENT**	Difficult IV access due to limb deformities. Arrange ultrasound and vein viewer to facilitate venous cannulation. Arrange for CPAP and tracheostomy due to increased risk of postoperative airway obstruction. Prepare for difficult airway cart including fiberoptic and video-laryngoscopes.

Abbreviations: VSD, ventricular septal defect; ASD, atrial septal defect; PDA, patent ductus arteriosus; ICP, intracranial pressure; CPAP, continuous positive airway pressure; GA, general anesthesia; RA, regional anesthesia; ETT, endotracheal tube.

Typical surgery and procedures

- Decompressive craniectomy
- Decompressive skull remodeling
- Ventricular peritoneal shunt surgery
- Diagnostic and therapeutic bronchoscopy
- Skull expansion surgery
- Midfacial advancement
- Naso-maxillary-zygomatic complex advancement
- Frontofacial distraction surgery to correct maxillary hypoplasia
- CAVE: extensive surgical trauma: tertiary level care required!
- Syndactyly release of fingers and toes
- Orthodontic surgery
- Ocular surgery e.g., strabismus and retinal surgery
- Sedation / General anesthesia for radiological procedures

Type of anesthesia

Anesthesia 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 [1].

Since they have an increased risk of OSA (obstructive sleep apnea), regional anesthetic techniques are preferable to avoid opiate-induced postoperative obstruction and apnea. Given that most major surgery will require opiates, these patients should be sent to a High Dependency Unit (HDU) or Intensive Care Unit (ICU) postoperatively.

However, regional anesthesia may be difficult due to multiple limb abnormalities and associated difficult positioning. Use of ultrasound is recommended since it may facilitate and improve block success in view of anticipated difficulty.

Necessary additional preoperative testing (beside standard care)

These patients frequently are admitted for various diagnostic, medical, and surgical procedures and may manifest high levels of anxiety. There is great need for counseling of these patients and their relatives to allay their anxiety.

The previous anesthetic management records should be checked for any significant perioperative event.

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 the 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.

Particular preparation for airway management

Vertebral fusion may be present, limiting cervical spine movement and presenting a potentially difficult airway

Patients may have a difficult bag and mask ventilation due to midface hypoplasia or secondary nasal obstruction due to a deviated nasal septum and choanal atresia [1,2]. Snoring and the presence of obstructive sleep apnea may suggest airway obstruction [3]. The obstruction may be overcome with the use of an oropharyngeal airway device.

Tracheal anomalies such as tracheal cartilaginous sleeve, tracheal ring and laryngeal web have also been reported, which may lead to respiratory distress [4-6].

Preexisting hydrocephalus with increased intracranial pressure may make positioning for intubation difficult [7].

Children with Pfeiffer syndrome can have an increased risk of difficult laryngoscopy and intubation. These children are prone to airway obstruction immediately after induction and following extubation. Preparations for a difficult airway are advisable at both these critical points.

The bag and mask ventilation is considered more challenging due to facial deformity and vertebral anomalies. The upper airway obstruction is common in patients with OSA, a history of previous episodes of upper airway obstruction, and those requiring domiciliary non-invasive ventilation [8].

Intubation is usually straightforward. Supraglottic airway devices are the most commonly used airway devices in pediatric anesthesia in modern era and can be used effectively in these children too.

The primary and alternative airway management plans should be ready with all possible devices available for intubation in case of need [9]. The nasopharyngeal airway or nasal intubation should be used with caution due to possible nasal stenosis.

A videolaryngoscope or fiberoptic bronchoscopy should be used early for intubation in view of anticipated difficulty and to reduce neck movement.

Extubation should be planned well and staggered/staged in case of difficult cases.

These children have a high incidence of tracheostomy due to the need of long-term airway support.

Particular preparation for transfusion or administration of blood products

No data available.

Particular preparation for anticoagulation

No data available.

Particular precautions for positioning, transportation and mobilization

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 well especially for surgeries in non-supine position.

Interactions of chronic disease and anesthesia medications

Not reported.

Anesthetic procedure

IV cannulation may be difficult due to limb abnormalities. Ultrasound or vein viewer guidance should be sought to increase success.

Use sedative premedication cautiously in patients with pre-existing features of airway obstruction and OSA. Further, avoid benzodiazepines and long-lasting opioids intraoperatively due to increased risk of OSA. Consider the use of shorter acting agents, such as desflurane, propofol and remifentanil, to facilitate a reliable return of airway reflexes and spontaneous ventilation at the end of the procedure.

Supplement general anesthetics with local anesthetics (local infiltration/nerve blocks/central neuraxial blocks) is recommended 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.

Particular or additional monitoring

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.

Possible complications

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

Postoperative care

An oropharyngeal 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.

Disease-related acute problems and effect on anesthesia and recovery

These arise from the underlying disease and should be considered to distinguish between a side effect of the anesthetic procedure and a manifestation of the disease itself.

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

Ambulatory anesthesia

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

Obstetrical anesthesia

No data available.

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