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Deutsche Akademie für Anästhesiologische Fortbildung e.V. (DAAF)
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Pfeiffer syndrome

Phocomelia

orphan anesthesia

a project of the German Society
of Anaesthesiology and Intensive Care Medicine

SUPPLEMENT NR. 1 | 2018

OrphanAnesthesia –
ein krankheitsübergreifendes Projekt des Wissenschaftlichen Arbeitskreises Kinderanästhesie der Deutschen Gesellschaft für Anästhesiologie und Intensivmedizin e.V.

Ziel des Projektes ist die Veröffentlichung von Handlungsempfehlungen zur anästhesiologischen Betreuung von Patienten mit seltenen Erkrankungen. Damit will Orphan Anesthesia einen wichtigen Beitrag zur Erhöhung der Patientensicherheit leisten.

Patienten mit seltenen Erkrankungen benötigen für verschiedene diagnostische oder therapeutische Prozeduren eine anästhesiologische Betreuung, die mit einem erhöhten Risiko für anästhesieassoziierte Komplikationen einhergehen. Weil diese Erkrankungen selten auftreten, können Anästhesisten damit keine Erfahrungen gesammelt haben, so dass für die Planung der Narkose die Einholung weiterer Information unerlässlich ist. Durch vorhandene spezifische Informationen kann die Inzidenz von mit der Narkose assoziierten Komplikationen gesenkt werden. Zur Verfügung stehendes Wissen schafft Sicherheit im Prozess der Patientenversorgung.

Die Handlungsempfehlungen von OrphanAnesthesia sind standardisiert und durchlaufen nach ihrer Erstellung einen Peer-Review-Prozess, an dem ein Anästhesist sowie ein weiterer Krankheitsexperte (z.B. Pädiater oder Neurologe) beteiligt sind. Das Projekt ist international ausgerichtet, so dass die Handlungsempfehlungen grundsätzlich in englischer Sprache veröffentlicht werden.

Ab Heft 5/2014 werden im monatlichen Rhythmus je zwei Handlungsempfehlungen als Supplement der A&I unter www.ai-online.info veröffentlicht. Als Bestandteil der A&I sind die Handlungsempfehlungen damit auch zitierfähig. Sonderdrucke können gegen Entgelt bestellt werden.

Bisher in A&I publizierte Handlungsempfehlungen finden Sie unter:

www.ai-online.info/Orphsuppl
www.orphananesthesia.eu

OrphanAnesthesia –
a common project of the Scientific Working Group of Paediatric Anaesthesia of the German Society of Anaesthesiology and Intensive Care Medicine

The target of OrphanAnesthesia is the publication of anaesthesia recommendations for patients suffering from rare diseases in order to improve patients' safety. When it comes to the management of patients with rare diseases, there are only sparse evidence-based facts and even far less knowledge in the anaesthetic outcome. OrphanAnesthesia would like to merge this knowledge based on scientific publications and proven experience of specialists making it available for physicians worldwide free of charge.

All OrphanAnesthesia recommendations are standardized and need to pass a peer review process. They are being reviewed by at least one anaesthesiologist and another disease expert (e.g. paediatrician or neurologist) involved in the treatment of this group of patients.

The project OrphanAnesthesia is internationally oriented. Thus all recommendations will be published in English.

Starting with issue 5/2014, we'll publish the OrphanAnesthesia recommenations as a monthly supplement of A&I (Anästhesiologie & Intensivmedizin). Thus they can be accessed and downloaded via www.ai-online.info. As being part of the journal, the recommendations will be quotable. Reprints can be ordered for payment.

A survey of until now in A&I published guidelines can be found on:

www.ai-online.info/Orphsuppl
www.orphananesthesia.eu

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orphananesthesia

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 craniosynostosis. Diagnoses can be established from the typical phenotype accompanied by molecular genetic testing.

Medicine in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

▶ **Citation:** Gupta A, Gupta N, Prottengeier J: Pfeiffer syndrome. Anästh Intensivmed 2018;59:S1-S7. DOI: 10.19224/ai2018.S001

Disease summary

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.

Craniosynostosis (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 xerophtalmia.

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 œsophagus, 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.

Typical surgery

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

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

Type of anaesthesia

Anaesthesia can involve both general and regional techniques. There are no known contraindications to specific anaesthetic 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 postoperatively.

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

Necessary additional diagnostic procedures (preoperative)

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 haematocrit and type and cross match should be performed for reconstruction procedures expected to cause significant blood loss.

Particular preparation for airway management

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.

Particular preparation for transfusion or administration of blood products

No data available.

Particular preparation for anticoagulation

No data available.

Particular precautions for positioning, transport or mobilisation

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.

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

Not reported.

Anaesthesiologic procedure

Intravenous 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 remifentanil, 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.

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.

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Possible complications

- 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

Postoperative care

- 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.

Information about emergency-like situations / Differential diagnostics

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.

Ambulatory anaesthesia

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.

Obstetrical anaesthesia

No data available.

Literature and internet links

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I would like to participate in the project ○

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