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Timothy syndrome

Tracheal agenesis

orphan**a**nesthesia

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

SUPPLEMENT NR. 2 | 2019

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 OrphanAnesthesia 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.

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 recommendations 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.

Bisher in A&I publizierte Handlungsempfehlungen finden Sie unter:

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

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

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orphananesthesia

Anaesthesia recommendations for patients suffering from **Tracheal agenesis**

Disease name: Tracheal agenesis, Tracheal atresia

ICD 10: Q32.4

Synonyms: -

Tracheal agenesis is a rare, congenital anomaly that may present to the anaesthesiologist in the operating room or in the delivery suite [1]. It was first reported by Payne in 1900 [2]. Since then, more than 150 cases worldwide have been reported in literature [3,4]. The incidence is 1/50,000 births, male: female ratio is 2:1 [3]. Several types have been described in literature. It usually manifests in the new born with severe respiratory distress and an absent cry [5]. There is failure to perform endotracheal intubation. Ventilation via a bag-valve-mask and oesophageal intubation (i.e. only in the presence of a trachea-oesophageal fistula) may enable supportive ventilation until a complete evaluation is made. Further, the airway management of laryngeal agenesis and tracheal agenesis at birth, in the delivery room differs significantly [1].

Embryologically, during the 3rd and 4th week of gestation, the oesophagus and trachea begin their development. Ventral displacement of the trachea-oesophageal septum at this stage causes the tracheal agenesis. Various anomalies of the larynx or trachea or both may develop [3].

Medicine 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

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Disease summary

Floyd has described three types of tracheal agenesis (TA) ranging from short tracheal segment attached to oesophagus to complete absence of trachea, whereas Faro [7] has described seven types ranging from pulmonary atresia to tracheal stenosis. Faro's classification system of tracheal agenesis (TA) has been nicely depicted by de Groot-van der Mooren et al [3].

Faro type A - total pulmonary agenesis

Faro type B/Floyd type III - complete TA with a separate origin of the main bronchi from the esophagus

Faro type C - total TA with normal main bronchi fusing in the midline at the carina, a trachea-oesophageal fistula (TOF) is present

Faro type D - larynx joined by atretic strand to distal trachea with a TOF present

Faro type E/Floyd type I - agenesis of the proximal trachea with a normal caudal segment of the distal trachea and a TOF

Faro type F- agenesis of the proximal trachea with a normal caudal segment of the distal trachea and no TOF is present (not shown)

Faro type G: a short segment of TA.

It is important to differentiate laryngeal from tracheal agenesis but sometimes they may occur together. In case of laryngeal agenesis, there is absent cry at birth and the neonate may have associated tracheoesophageal fistula. The diagnosis is made on laryngoscopy and emergency tracheostomy is needed. In case of tracheal agenesis, there may be a weak cry at birth, CT scan may be necessary to make the diagnosis and oesophageal intubation is needed for short-term management. Tracheostomy may be lifesaving but this depends on the length of trachea available. Complete tracheal agenesis is incompatible with life and survival may be less than 6 weeks. The longest reported survival is up to 6 years of age [8]. Survival depends on the nature and severity of the agenesis. In the future, use of regenerative medicine and tissue engineering with amniotic fluid cells for reconstruction of trachea and larynx may improve outcomes in these patients [9].

Typical surgery

These patients may present at birth or in the new-born period or later for tracheal or oesophageal reconstruction [10–13]. If mask ventilation is successful at birth, there are three immediate possible surgical steps that may be considered:

- (1) oesophageal banding of the lower oesophagus for palliation with the aim to substituting the oesophagus for the trachea
- (2) cervical oesophagostomy to facilitate drainage or insertion of a tracheostomy tube
- (3) gastrostomy for enteral feeding.

Following initial palliative substitution of oesophagus for trachea, further management of the trachea-oesophageal fistula may be managed as follows [12]:

- (1) conservative management
- (2) deliberate insertion of the endotracheal tube into the fistula to facilitate ventilation
- (3) oesophageal reconstruction using small intestine or colonic interposition or a gastric tube

The patient may present for following procedures:

- emergency tracheostomy
- repair/ reconstruction of trachea-oesophageal fistula
- tracheal reconstruction
- oesophageal reconstruction
- oesophageal banding (to substitute oesophagus for trachea) and gastrostomy (for enteral feeding)
- ex-utero intrapartum therapy (EXIT procedure).

Type of anaesthesia

The neonate may present at birth or in the new-born period with other congenital anomalies that confounds the clinical signs and diagnosis [14]. Typically, general anaesthesia would be used for these procedures, however the airway management may be challenging. There is high mortality associated with this disorder and the decision for further management does need an inter-disciplinary team approach.

In case of laryngeal atresia/agenesis or partial tracheal agenesis, an emergency tracheostomy would be lifesaving and may be performed as an EXIT procedure since tracheal and laryngeal agenesis may be a cause for Congenital High Airway Obstruction syndrome (CHAOS) [15].

Necessary additional diagnostic procedures (preoperative)

Prenatal diagnostic procedures:

In the absence of a trachea-oesophageal fistula, both laryngeal and tracheal agenesis may present as congenital high airway obstruction syndrome (CHAOS) [15]. The characteristic antenatal ultrasound findings are polyhydramnios, foetal ascites, fluid in the upper airway, hyper echogenic lungs, flattened or inverted diaphragm, dilated airways distal to the obstruction and vigorous large amplitude foetal breathing. However, in the presence of a tracheo-oesophageal fistula, these signs are absent on the ultrasound examination because the fluid can pass into the stomach or amniotic sac via the fistula. Foetal MRI is used to confirm the diagnosis if there is suspicion for tracheal agenesis on the antenatal ultrasound and is useful for planning further management. Testing amniotic fluid for phospholipids is useful as an additional test, since these are secreted by the respiratory tract [16].

Postnatal diagnostic procedures:

A rigid bronchoscopy and an emergency CT scan image [17,18] to confirm the diagnosis is necessary for further management. A chest radiograph will reveal absence of tracheal shadow and posteriorly located endotracheal tube. Other diagnostic tests, as part of VACTERL work up will include echocardiography, and blood gases.

Particular preparation for airway management

This can be very challenging. Emergency tracheotomy at birth is life saving in case of laryngeal agenesis or partial tracheal agenesis. In case of tracheal agenesis, options include oesophageal intubation and ventilating the lungs through a broncho-oesophageal fistula, EXIT procedure with opening of mediastinum and intubating the bronchus. In case of proximal agenesis of trachea with intact caudal segment, oesophageal banding to temporarily substitute trachea for oesophagus and a gastrostomy for feeding has been reported in literature. Hence, adequate size face masks, endotracheal tube, flexible and rigid bronchoscopy must be made available. Also smaller size endotracheal tubes must be available. Care must be taken during intubation as there may be fragility of the oesophagus/fistula/ trachea and there is a risk of perforation and bleeding.

Particular preparation for transfusion or administration of blood products

There is no known specific bleeding diathesis that is associated with this condition. As with any complicated paediatric patient procedures in the operating room, bank blood may need to be made available.

Particular preparation for anticoagulation

There is no evidence to support the need for anticoagulation with this condition.

Particular precautions for positioning, transport or mobilisation

This can be challenging. Transport may be intra-hospital or inter-hospital when referring to a tertiary centre. Transport within the hospital may be from the delivery room to the Neonatal ICU, from the NICU to the operating room or to the CT scan suite, all of which may be located on different floors. Clinical concerns include maintaining a patent airway, oxygen saturation, ventilation strategies, and haemodynamic stability especially when there are other anomalies present and maintenance of normothermia (normal body temperature). Good communication between the neonatology, anaesthesia and surgery teams is paramount. Standard monitoring, especially capnography is essential in this case.

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

No data is available for this condition in particular. There are no reported interactions with anaesthetic drugs.

Anaesthesiologic procedure

Anaesthesia on a patient suffering from tracheal and/or laryngeal agenesis must be performed by an experienced team of paediatric anaesthesiologists. Due to the severity of the airway anomaly, establishing an airway management plan ahead of time with adequate discussion between neonatology, surgery and anaesthesia teams is absolutely essential.

The neonate may be transported to the operating room for tracheostomy, repair of trachea-oesophageal fistula or gastrostomy with oesophageal banding. The neonate may be already intubated in the delivery room with an oesophageally placed endotracheal tube for lung ventilation via the fistula. A fiberoptic or a rigid laryngoscopy/ bronchoscopy is performed in the operating room to confirm the diagnosis. Further, anaesthesia induction and maintenance is performed with inhalational agents. The use of muscle paralyzing agents is based on individual clinical judgement, however with difficulties establishing a secure airway, it is prudent to have the neonate breathe spontaneously and intubate under deep inhalational anaesthesia. Tracheostomy in the delivery room is lifesaving and can be performed under local anaesthetic. Again, this will depend on the length of the trachea that is available.

If an EXIT (Ex-utero Intrapartum Therapy) procedure is planned for partial agenesis, a lot of planning will be needed [3,19]. Two anaesthesia teams (maternal and paediatric), emergency neonatal resuscitation team, obstetric and surgery team will be needed. Maternal anaesthesia may be challenging and will need to focus on adequate maternal anaesthesia and maintaining uterine relaxation until the cord is clamped. This is usually achieved with general endotracheal anaesthesia with volatile anaesthetic agents but neuraxial anaesthesia and nitroglycerine for uterine relaxation may also be used. The foetus is delivered after maternal hysterotomy, while maintaining uterine relaxation, the procedure for securing the foetal airway is performed followed by clamping of the umbilical cord. It is important to remember that the neonate's lungs must not be ventilated until the foetal surgery is completed and the umbilical cord is divided. Premature foetal/ neonatal ventilation may result in transitional neonatal circulation thereby losing the benefits of placental support during this procedure. A neonatal team must be prepared to receive the neonate when the procedure is successfully completed. If not successfully completed, a second prepared operating room will be needed to complete the surgery on the neonate. The paediatric anaesthesia team then follows the neonate to the operating room and must be prepared for receiving a critically ill neonate with airway compromise, without intravenous access or with active bleeding.

After initial palliation surgery, these patients may need surgical management of the fistula in the form of anastomosis or external stent placement. Again, the anatomy and the surgical plan will need to be discussed with the surgical and the intensive care teams.

Particular or additional monitoring

Standard monitoring will include ECG, pulse oximetry, non-invasive blood pressure, capnography and body temperature. Invasive arterial line monitoring e.g. umbilical arterial line monitoring may be useful. During maternal anaesthesia for EXIT procedures, foetal monitoring may be achieved with pulse oximetry and heart rate.

Possible complications

The most severe and life threatening complication is inability to establish a patent airway and death from hypoxia and airway compromise. The other complications include ventilation problems and feeding difficulties. Long term there is a risk of oesophageal or tracheal stenosis, oesophageal perforation, bleeding and nutritional issues [8].

Postoperative care

This will be complicated by the severity of the disease and the airway management. The neonate will need to be in the neonatal intensive care unit (NICU) for further management including mechanical ventilation therapy. Post operatively, the neonate may need intermittent mandatory ventilation or CPAP (continuous positive airway pressure). There may be tracheomalacia, stenotic fistula which may collapse during inspiration and may need external stent placement. Weaning from the ventilator is challenging in these cases. In the long-term after staged procedures, the child may need home oxygen and/or ventilator therapy.

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 diseases, e.g.:

As mentioned before, the emergency situation presents at birth or shortly after birth in patients with tracheal agenesis. Tracheal agenesis may occur with or without laryngeal agenesis, the management may differ significantly as isolated laryngeal atresia or agenesis may be managed with a tracheostomy. In case of tracheal agenesis, while a tracheostomy may be lifesaving, its success will depend on the tracheal length that is available. The diagnosis may be antenatal or postnatal with high index of clinical suspicion and confirmation with foetal MRI.

Ambulatory anaesthesia

Not applicable with this anomaly due to the complexity of the clinical condition.

Obstetrical anaesthesia

Because of the short life-expectancy and poor long term prognosis for infants in case of tracheal agenesis, there are no reports pertinent to obstetric anaesthesia in these patients.

Literature and internet links

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