

orphan^ainesthesia

Anesthesia recommendations for patients suffering from **CHARGE syndrome**

Disease name: CHARGE syndrome

ICD 10: Q87.8

Synonyms: CHARGE association; Hall-Hittner syndrome

CHARGE syndrome was initially defined as a non-random association of anomalies

- **C**oloboma
- **H**ear defect
- **A**tresia Choanae
- **R**etarded Growth and Development
- **G**enital Hypoplasia
- **E**ar Anomalies/Deafness

In 1998, an expert group defined the major (the classical 4C's: *Choanal atresia, Coloboma, Characteristic ear and Cranial nerve anomalies*) and minor criteria of CHARGE syndrome. Individuals with all four major characteristics or three major and three minor characteristics are highly likely to have CHARGE syndrome [1].

Medicine in progress



Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong



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

Minor criteria:

- Cardiovascular malformations
- Genital hypoplasia
- Cleft lip and palate
- Tracheoesophageal fistula
- Distinctive CHARGE facies
- Growth deficiency
- Developmental delay

Occasional:

- Renal anomalies: Duplex system, vesicoureteric reflux
- Spinal anomalies: Scoliosis, osteoporosis
- Hand anomalies
- Neck/shoulder anomalies

In many children with CHARGE Syndrome mutations in the CHD7 gene in chromosome 8q12 can be detected. Most cases are sporadic [2].

Typical surgery

- Heart (total repair, shunts, vascular ring and patent ductus ligations)
- Ears (examination, myringotomy tubes)
- Nose and throat (choanal atresia repair, cleft lip/palate repair, tracheostomy)
- Gastrointestinal tract (closure of tracheoesophageal fistula, fundoplication, gastrostomy)
- Genitourinary system (vesicoureteral reflux, hypospadias, cryptorchidism)
- Diagnostic scopes (nasopharyngoscopy, laryngoscopy, bronchoscopy)
- Diagnostic procedures (MRI, CTscan, scintigraphy)
- Eyes (examination)
- Other (hernia repair, circumcision, cochlear implant, removal of granuloma, nephrotomy tube insertion)

Type of anaesthesia

Choice of the anaesthesiologic procedure is dependent mainly on the individual phenotypic features and the presence of organ manifestations.

General anaesthesia can be done with either volatile or intravenous anaesthetics.

Necessary additional diagnostic procedures (preoperative)

- Echocardiography
- Blood gases analysis, creatinin, electrolytes and calcium

Optional:

- Abdominal ultrasound for renal anomalies
- Chest x-ray in the presence of lower respiratory tract infection
- Neurological examination for presence of hypotonia or facial palsy
- Cranial MRI and EEG

Particular preparation for airway management

Up to 56% of the patients with CHARGE association have upper airway abnormalities apart from choanal atresia and cleft lip and palate [3]. Up to 50% of the patients need tracheotomy not only for associated airway abnormalities but also for salivatory retention, swallowing disorders and chronic aspiration [4].

Common airway abnormality in the CHARGE association are

- Choanal atresia
- Cleft lip and palate
- Micrognathia
- Laryngomalazia
- Subglottic stenosis
- Bulbar palsy
- Laryngeal cleft
- Recurrent laryngeal nerve palsy
- Tracheomalazia
- Tracheo-oesophageal fistula

Before anaesthesia the presence of active lower respiratory tract infections should be determined and treated. A careful search for airway abnormalities should be accomplished. Consultation of an ear, nose, and throat specialist is recommended. Planning of the airway management is dependent on the existing airway abnormality and on the presence of gastro-oesophageal reflux and chronic aspiration. In an urgent situation a difficult airway should be anticipated.

Particular preparation for transfusion or administration of blood products

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

Particular preparation for anticoagulation

Not reported.

Particular precautions for positioning, transport or mobilisation

Not reported.

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

Not reported.

Anaesthesiologic procedure

Because cooperation is limited often in these children, sedative premedication and the presence of the parents during induction may be helpful.

Gaseous induction of anaesthesia may be difficult in patients who are severely affected with drooling because of decreased ability to swallow secretions, but may be preferable for patients with a tracheotomy and difficult intravenous access.

Due to a high incidence of gastro-oesophageal reflux some anaesthetists prefer a rapid sequence induction but there is no evidence to suggest that this increases safety.

There are no studies of different airway techniques in this group. One case report describes successful airway management with the use of a laryngeal mask airway in a patient with CHARGE syndrome and a Cormack Lehane score of IV in direct laryngoscopy [5].

Particular or additional monitoring

Monitoring is dependent on the individual phenotypic CHARGE features and the surgical procedure.

Possible complications

Postoperative adverse airway event i.e. decreased oxygen saturations, excessive secretions resulting in airway obstruction, aspiration, prolonged crackles and wheezing decrease in respiratory rate, stridor, atelectasis and pneumothorax.

Arrhythmias and other heart rate abnormalities [6].

Postoperative care

Postoperative airway events of individuals with charge syndrome occur after up to 35% of anaesthetics.

Surgeries with the most airway events involve the heart, the gastrointestinal tract and airway diagnostic scopes. Combining multiple surgical procedures under one anaesthetic does not increase the risk of postoperative airway events. Individuals who require a Nissen

funduplicatio or a gastrostomy tube insertion have a significantly higher risk for anaesthetic airway events [6].

Prolonged postoperative mechanical ventilation may be required. Prolonged supervision in the recovery room or in an intermediate care or intensive care unit is highly recommended.

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.:

Not reported.

Ambulatory anaesthesia

Due to the significantly higher risk for postanaesthetic adverse airway events ambulatory anaesthesia is not recommended.

Obstetrical anaesthesia

Not reported.

Literature and internet-links

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3. Stack CG, Wyse RK: Incidence and management of airway problems in the CHARGE Association. Anaesthesia 1991; 46(7): 582-5.
4. Naito Y, Higuchi M et al. : Upper airway obstruction in neonates and infants with CHARGE syndrome. American Journal Of Medical Genetics Part A 2007; 143A (16): 1815-20.
5. Hara Y et al.: Successful airway management with use of a laryngeal mask airway in a patient with CHARGE syndrome. J Anesth 2009; 23: 630 - 632
6. Blake K, MacCuspie J et al.: Postoperative airway events of individuals with CHARGE syndrome. International Journal Of Pediatric Otorhinolaryngology 2009; 73(2): 219-26..
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