

Anaesthesia recommendations for **CHARGE syndrome**

Disease name: CHARGE syndrome

ICD 10: Q87.8

Synonyms: CHARGE association; Hall-Hittner syndrome

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

- Coloboma
- Heart defect
- Atresia choanae (choanal atresia)
- Retarded growth and development
- Genital hypoplasia
- Ear 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 [1]. In 2004, mutations in the CHD7 gene were identified as the major cause. The inheritance pattern is autosomal dominant with variable expressivity. Almost all mutations occurs de novo, but parent-to-child transmission has occasionally been reported [2].

Clinical criteria for CHARGE syndrome [1]

Major criteria:

- Coloboma
- Choanal Atresia
- Cranial nerve anomalies
- Abnormalities of the inner, middle, or external ear

Minor criteria:

- Cardiovascular malformations
- Genital hypoplasia or delayed pubertal development
- Cleft lip and/or palate
- Tracheoesophageal defects
- Distinctive CHARGE facies
- Growth retardation
- Developmental delay

Occasional:

- Renal anomalies: duplex system, vesicoureteric reflux
- Spinal anomalies: scoliosis, osteoporosis
- Hand anomalies

- Neck/shoulder anomalies
- Immune system disorders

Individuals with all four major characteristics or three major and three minor characteristics are highly likely to have CHARGE syndrome [1].

CHARGE syndrome occurs in approximately 1 in 10000 newborns [3]. In more than 90% of patients fulfilling the clinical criteria for CHARGE syndrome mutations in the CHD7 gene in chromosome 8q12 can be detected [4].

Medicine is 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

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 pre-operative testing (beside standard care)

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

Optional:

- Abdominal ultrasound for renal anomalies
- Chest x-ray in the presence of lower respiratory tract infection
- Assessment of cranial nerve function
- Evaluation of obstructive sleep apnoea in case of sleep disturbances
- Cranial MRI
- EEG when seizures are observed

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 [5]. Up to 50% of the patients need tracheotomy not only for associated airway abnormalities but also for salivatory retention, swallowing disorders and chronic aspiration [6].

Common airway abnormality in the CHARGE association:

- Choanal atresia
- Cleft lip and palate
- Micrognathia
- Laryngomalacia
- 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. 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, transportation and mobilisation

Not reported.

Interactions of chronic disease and anaesthesia medications

Not reported.

Anaesthetic procedure

Because cooperation is limited often in these children, sedative premedication and the presence of the parents during induction may be helpful. If obstructive sleep apnoea syndrome is present benzodiazepines as premedication should be avoided.

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 [7].

Particular or additional monitoring

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

Possible complications

A difficult airway should be anticipated in every child with CHARGE syndrome. Postoperative airway events i.e. decreased oxygen saturations, excessive secretions resulting in airway obstruction, aspiration, prolonged crackles and wheezing decrease in respiratory rate, stridor, atelectasis and pneumothorax occur after up to 35% of anaesthetics [8].

Surgeries with the most adverse 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-fundoplicatio or a gastrostomy tube insertion have a significantly higher risk for anaesthetic airway events [8].

Post-operative care

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.

Disease-related acute problems and effect on anaesthesia and recovery

Not reported.

Ambulatory anaesthesia

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

Obstetrical anaesthesia

Not reported.

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