orphanaesthesia

Anaesthesia recommendations for

Fraser syndrome

Disease name: Fraser syndrome

ICD 10: Q87.0

Synonyms: Cryptophthalmos syndrome

Disease summary: Autosomal-recessive inherited congenital disorder of cryptophthalmos, ear and facial abnormalities, cutaneous syndactyly and genital malformations [1]. Classical Fraser syndrome is caused by mutation of the FRAS1 gene located on chromosome 4 at 4q21.21 [1]. Mutations of FREM1, FREM2 and GRIP1 genes can cause a similar clinical phenotype to Fraser syndrome [2]. First described by Zehender and Manz in 1872 [3] as cryptophthalmos alone but the complete syndrome was described by Fraser in 1962 [4]. Diagnosis of Fraser syndrome is complex and there is debate on the criteria for a diagnosis [5]. Current incidence in Europe is 2 per million live births with 27.8% of infants with Fraser syndrome from consanguineous parents [6].

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

- Ophthalmic surgery for cyptophthalmos
- Hand surgery for syndactly
- Urological [7] and gynaecological surgery for ambiguous genitalia
- Craniofacial reconstruction for facial deformities
- ENT assessment for airway abnormalities and tracheostomy
- Rectal surgery for anorectal malformations
- Neurosurgery for VP shunt

Type of anaesthesia

General anaesthesia with or without regional anaesthesia as appropriate for the procedure.

Necessary additional pre-operative testing (beside standard care)

Difficult and impossible laryngeal intubation has been reported in the literature [6,9,10,11]. Of particular note, severe subglottic stenosis without clinical signs has been described [11,12]. Assessment by an ENT surgeon prior to the first general anaesthetic should be considered. An ENT surgeon may need to be available for the first induction of anaesthesia if there is any evidence of airway compromise such as stridor.

13% of Fraser syndrome children have an associated congenital heart defect – ASD, VSD and pulmonary artery anomalies have been reported so a pre-operative ECHO is mandatory [6].

Particular preparation for airway management

Epidemiological data from 16 countries in Europe from 1990 – 2008 have shown the following associated airway complications [6]:

- Cleft palate – 8%
- Micrognathism – 8%
- Laryngeal stenosis – 21%
- Subglottic stenosis – 4%

Impossible laryngeal intubation from a congenital laryngeal web has also been reported [9].

Rescue ventilation via face mask and supra-glottic airway devices have been successfully performed. A careful assessment of the airway should be performed prior to anaesthetising these children and the full range of difficult airway equipment made immediately available for use.

Emergency tracheostomy and retrograde intubation techniques have been described [9,10,11].

A case review of 125 anaesthetics delivered to ten children across 30 years with Fraser syndrome identified difficult intubation in one child and impossible intubation in a second. The impossible intubation was due to a laryngeal inlet that was narrowed and too small for an endotracheal tube. Both children were easy to ventilate by mask or LMA and had no evidence of airway obstruction prior to induction of anaesthesia [11].

In children without airway disorders standard airway techniques can be used [8].

Particular preparation for transfusion or administration of blood products

No reported issues.

Particular preparation for anticoagulation

No reported issues.

Particular precautions for positioning, transportation and mobilisation

No reported issues.

Interactions of chronic disease and anaesthesia medications

No reported cases of anaesthetic agent reactions.

Anaesthetic procedure

Gaseous or IV induction as deemed appropriate. Particular attention to the child with even minimal stridor on preoperative assessment – this may be a herald sign of airway compromise.

Particular or additional monitoring

None required.

Possible complications

No specific complications known.

Post-operative care

No specific postoperative care issues.

Disease-related acute problems and effect on anaesthesia and recovery

No specific emergency-like situations known apart from airway problems mentioned earlier.

Ambulatory anaesthesia

No specific contraindications to ambulatory anaesthesia.

Obstetrical anaesthesia

No documented literature on obstetric anaesthesia with Fraser syndrome patients.

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