

Anaesthesia recommendations for patients suffering from **Fraser Syndrome**

Disease name: Fraser Syndrome

ICD 10: Q87.0

Synonyms: Cryptophthalmos syndrome

Autosomal-recessive inherited congenital disorder of cryptophthalmos, ear and facial abnormalities, cutaneous syndactyly and genital malformations¹. 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 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

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

Type of anaesthesia

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

Necessary additional diagnostic procedures (preoperative)

Difficult and impossible laryngeal intubation has been reported in the literature [6,9,10]. Of particular note, subglottic stenosis without clinical signs has been described [11]. 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 anomolies 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 has 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 has 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 previously been described [9,10].

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, transport or mobilisation

No reported issues.

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

No reported cases of anaesthetic agent reactions.

Anaesthesiologic 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.

Postoperative care

No specific postoperative care issues.

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.:

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.

Literature and internet links

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8. Dakin M and Bingham R. Anaesthetic considerations in patients with Fraser syndrome. *Anaesthesia* 1995; 50: 746
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