orphanaesthesia

Anesthesia recommendations for

Fraser syndrome

Disease name: Fraser syndrome

ICD 10: Q87.0

ORPHAcode: 2052

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].

Diagnosis may be incorrect; if uncertainty exists, the diagnosis should be re-evaluated.

Every patient is unique; individual circumstances must always guide clinical care.

Medicine is in progress; new clinical knowledge may not be yet reflected in this guideline Perhaps new knowledge.

Recommendations are not rules or laws; they provide a framework to support clinical decision-making. Although this recommendation has passed a structured review process, it does not meet the formal criteria of a guideline.

Translations may not always reflect the most recent updates of the English version.

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

Emergency information

A	**AIRWAY / ANESTHETIC TECHNIQUE**	Known incidence of impossible intubation. Prepare for emergency tracheostomy. Otherwise, asymptomatic impossible intubation described in the literature.
B	**BLOOD PRODUCTS (COAGULATION)**	No known problems.
C	**CIRCULATION**	Syndrome associated with cardiac defects – ASD, VSD and pulmonary artery defects reported in the literature.
D	**DRUGS**	No known problems.
E	**EQUIPMENT**	Prepare for smaller than expected airway anatomy – especially subglottic stenosis.

Typical surgery and procedures

- Ophthalmic surgery for cryptophthalmos
- Hand surgery for syndactyly
- Urological [7] and gynecological 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 anesthesia

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

Necessary additional preoperative 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 [11,12,13]. Assessment by an ENT surgeon prior to the first general anesthetic should be considered. An

ENT surgeon may need to be available for the first induction of anesthesia 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%
- Micrognathia – 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 anesthetizing these children and the full range of difficult airway equipment made immediately available for use.

Emergency tracheostomy and retrograde intubation techniques have been described [9,10,11,13].

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 mobilization

No reported issues.

Interactions of chronic disease and anesthesia medications

No reported issues and no reported cases of anesthetic agent reactions.

Anesthetic 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.

Disease-related acute problems and effect on anesthesia and recovery

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

Ambulatory anesthesia

No specific contraindications to ambulatory anesthesia.

Obstetrical anesthesia

No documented literature on obstetric anesthesia with Fraser syndrome patients.

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