orphananesthesia

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 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 in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

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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 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 previously been described [9,10].

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

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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.

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Literature and internet links

1. Francannet C, Lefrançois P, Dechelotte P, Robert E et al. Fraser syndrome with renal agenesis in two consanguineous Turkish families. Am J Med Genet 2005; 36 (4): 477-479
2. Hoefele J, Wilhelm C, Schiesse M, Mack R et al. Expanding the mutation spectrum for Fraser Syndrome: Identification of a novel heterozygous deletion in FRAS1. Gene 2013; 520: 194-197
3. Gupta SP and Saxena RC. Cryptophthalmos. Brit J Ophthalmol 1962; 46: 629-32
4. Fraser GR. Our genetic 'load'. A review of some aspects of genetical variation. Ann Hum Genet 1962; 25: 387-415
5. Slavotinek A and Tifft C. Fraser syndrome and cryptophthalmos: review of the diagnostic criteria and evidence for phenotypic modules in complex malformation syndromes. J Med Genet 2002; 39: 623-633
6. Barisic I, Odak L, Loane M, Garne E, et al. Fraser Syndrome: Epidemiological study in a European population. Am J Med Genet Part A 2013; 161A: 1012-1018
7. Andiran F, Tanyel F, Hiçsönmez. Fraser Syndrome Associated With Anterior Urethral Atresia. Am J Med Genet 1999; 82: 359-361
8. Dakin M and Bingham R. Anaesthetic considerations in patients with Fraser syndrome. Anaesthesia 1995; 50: 746
9. Crowe S, Westbrook A, Bourke M, Lyons B, et al. Impossible laryngeal intubation in an infant with Fraser syndrome. Paediatr Anaesth 2004; 14: 276-278
10. Jagtap SR, Malde AD, Pantvaidya S H. Anaesthetic considerations in patients with Fraser syndrome. Anaesthesia 1995; 50: 39-41
11. Rose J and Ketterick R. Subglottic stenosis complicating the anaesthetic management of a newborn with Fraser syndrome. Paediatr Anaesth 1993; 3: 383-385.

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These guidelines have been prepared by:

Authors
Jonathan Mathers, Anaesthesiologist, Great Ormond Street Hospital, London, United Kingdom
jonathanmathers@me.com

Jonathan Smith, Great Ormond Street Hospital, London, United Kindgom

Peer revision 1
Suzanne Crowe, Anaesthesiologist, Our Lady's Hospital for Sick Children, Dublin, Ireland
Suzanne.Crowe@amnch.ie

Peer revision 2
Kaarthigeyan Kalaniti, Paediatrician, The Hospital for Sick Children (SickKids), University of Toronto, Canada
kaarthigeyank@yahoo.com

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