

Anaesthesia recommendations for **Beals syndrome**

Disease name: Beals Syndrome

ICD 10: Q68.8

Synonyms: Congenital contractural arachnodactyly (CCA), Beals syndrome, Beals-Hecht syndrome.

Disease summary: Beals syndrome was first described by Beals and Hecht in 1971 [1].

Beals syndrome is an extremely rare connective tissue disorder, characterised by multiple flexion contractures, arachnodactyly, severe kyphoscoliosis, abnormal pinnae and muscular hypoplasia [2,3]. The clinical features are similar to Marfan's syndrome. It differs from Marfan's syndrome in that the incidence of cardiac abnormalities like aortic root dilatation are much lower in Beals syndrome and the presence of multiple flexion contractures is characteristic of Beals syndrome. However, patients with Beals syndrome may present with mitral valve prolapse and other congenital heart disease.

Beals syndrome is an autosomal dominant condition associated with mutation in *FBN2* gene on chromosome region 5q23. The incidence of Beals syndrome is unknown and prevalence is difficult to estimate due to the overlap in phenotype with Marfan's syndrome [4]. Males and females are equally affected. Individuals with Beals syndrome are expected to be cognitively normal. Delay in the motor development is common due to contractures.

Medicine is in constant progress; new clinical knowledge may not be in this text.



Recommendations are not rules or laws; they are a framework for clinical decision-making.

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

The diagnosis may be wrong; if questionable, the diagnosis should be confirmed.



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

Typical surgery

Scoliosis correction surgery, contracture release, reduction of long bone fractures.

Type of anaesthesia

There is no definite recommendation for general anaesthesia or regional anaesthesia.

Beals syndrome is associated with difficult intravenous access, difficult airway and difficult positioning due to multiple contractures. There are reported cases of Beals syndrome children with difficult laryngoscopy and intubation, due to dysmorphic features.

Little information is available in literature regarding neuraxial blockade and regional anaesthesia in these patients. The presence of scoliosis and/ or kyphosis can present a significant technical challenge. Regional anaesthesia may be challenging due to contractures and difficulties in positioning.

Necessary additional pre-operative testing (beside standard care)

Respiratory function should be assessed preoperatively as persons with Beals syndrome can have restrictive lung disease.

Children with Beals syndrome can present with various heart defects such as septal defects (ASD, VSD), interrupted aortic arch and mitral valve prolapse. A preoperative echocardiogram should be done to rule out the presence of cardiac defects and its effects.

Although ocular involvement is yet unclear, a thorough ophthalmologic evaluation is recommended.

Particular preparation for airway management

There are reported cases of difficult airway in children with Beals syndrome [5,7]. Difficult laryngoscopy and intubation is reported due to restricted mouth opening, micrognathia and high arched palate. A thorough preoperative evaluation of the airway and an adequate management plan should be in place before anaesthetising these children. Preparations for difficult airway management are advisable ranging from simple (oropharyngeal/ nasopharyngeal airways) to the advanced (video laryngoscope/ fiberoptic bronchoscope).

Particular preparation for transfusion or administration of blood products

There is no evidence for specific transfusion practices in children with Beals syndrome. As in any scoliosis correction surgery, excessive bleeding and need for blood transfusion should be anticipated in children undergoing scoliosis correction surgery and general measures such as tranexamic acid and cell salvage should be considered.

Particular preparation for anticoagulation

No information on specific disease related pathophysiology.

Particular precautions for positioning, transportation and mobilisation

Special care should be taken while positioning and adequate padding of all bony protuberances should be ensured.

Interaction of chronic disease and anaesthesia medications

Not reported.

Anaesthetic procedure

Both inhalational and TIVA techniques may be used. There is no evidence favouring any particular induction or maintenance agent. There are reported cases of difficult intubation in children with Beals syndrome. So a thorough preoperative evaluation of airway and an adequate management plan should be in place before anaesthetising these children.

Regional anaesthesia can be challenging due to multiple joint contractures, scoliosis and spine deformities.

Particular or additional monitoring

Intraoperative monitoring needs to be tailored to the procedure and presence of comorbidities. Consider cardiac monitoring in children with cardiac comorbidities.

Possible complications

- Difficult airway
- Postoperative respiratory complications can occur due to concurrent restrictive lung disease, and poor airway control
- Cardiac complications – cardiac monitoring as necessary shall be performed
- Careful positioning due to multiple joint contractures.

Post-operative care

Depending on presence of comorbidities (congenital heart disease) and type of surgery performed (scoliosis correction); they may need special care and monitoring (HDU, ICU) postoperatively.

Disease-related acute problems and effect on anaesthesia and recovery

Airway problems as mentioned above.

Ambulatory anaesthesia

Minor procedures especially in patients without comorbidities can be carried out as day case procedures.

Obstetrical anaesthesia

No documented literature on Obstetric anaesthesia with Beals syndrome patients.

References

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