orphanaesthesia

Anesthesia recommendations for patients suffering from

De Barsy syndrome

Disease name: De Barsy syndrome

ICD 10: Q87.7; OMIM 614438

Synonyms: DBS, De Barsy-Moens-Dierckx syndrome, Progeroid syndrome of De Barsy, Autosomal recessive cutis laxa Type 3
*With 2 gene subdivisions:
ARCL3A: caused by a ALDH18A1 mutation
ARCL3B: caused by a PYCR1 mutation

DeBarsy syndrome is a rare clinical syndrome characterized by cutis laxa, ophthalmic opacification, skeletal malformations, as well as mental and growth retardation. This disease is genetically transmitted in an autosomal recessive fashion. Affected patients often require surgical correction of ophthalmic and orthopaedic abnormalities. This syndrome was first described by A.M. De Barsy in 1967 and less than 100 known cases are documented in the medical literature. Very little has been published on this rare disorder and only a single article has addressed anesthesia case outcomes and management strategies (Aponte, 2010).

The diverse collection of clinical manifestations in De Barsy syndrome includes: intra-uterine growth retardation (IUGR), postnatal growth delay, motor delay, cognitive impairment, hypontonia, athetoid movements, malformations, microcephaly, wormian bones, large fontanelles, facial dysmorphism, cataracts, corneal clouding, thin/wrinkled skin, easy bruising, sparse hair, joint laxity, osteopenia, and inguinal hernias.

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

- Eye procedure (cataract, eye exam)
- Orthopaedic procedure (hip arthrogram/open reduction, joint stabilization, spinal fusions, capsulodesis)
- Skin biopsy
- Wound check
- Radiologic imaging (MRI)
- GI procedures (Nissen, g-tube placement, EGD)
- ENT procedure (myringotomy and tube placement)
- Urology (orchiopexy, circumcision)
- Hernioplasty

Type of anaesthesia

- General anaesthesia
- Regional anaesthesia for pain control
- Monitored anaesthesia care

Necessary additional diagnostic procedures (preoperative)

Given the wide variation of disease severity and clinical presentation seen in De Barsy patients, no consensus exists regarding standardized preoperative testing. Providers may wish to consider electrocardiographic and echocardiographic screening for cardiac anomalies based upon previously published reports. In two separate publications, De Barsy patients were noted to have progressive aortic root dilation (Lin, Chang et al. 2011) (Dutta, Ekbote et al. 2016).

In another published case series (Aponte, Smith et al 2010), of De Barsy patients, neonatal cardiac anomalies were discovered in one patient who went on to have division of a double aortic arch and vascular ring repair. In another patient, echocardiography revealed patent foramen ovale, trivial tricuspid regurgitation and possible biventricular hypertrophy, later diagnosed as idiopathic cardiomyopathy. The medical records of a third patient with De Barsy syndrome revealed pulmonary branch stenosis, however, no surgical intervention was required and serial echocardiographic studies remained normal.

Particular preparation for airway management

There is a paucity of literature with regard to De Barsy syndrome patients and airway management. The midface hypoplasia and microphephaly malformations suggest caution with regard to airway manipulation. However, during 64 anaesthetics for three patients performed at the Mayo Clinic, a single difficult airway was reported. This case involved fiberoptic bronchoscope intubation after several failed attempts with standard laryngoscopy. A variety of perioperative respiratory difficulties have been documented including asthma exacerbation, tracheomalacia, obstructive sleep apnea, central sleep apnea, aspiration pneumonia and restrictive lung disease. The vast majority of cases were performed with video laryngoscopy and this technique is recommended for De Barsy syndrome patients.

Particular precautions for transfusion or administration of blood products

The presence of cutis laxa, or the absence of normal elasticity and skin tone, predisposes De Barsy patients to easy bruising because of vascular fragility. Skin laxity contributes to challenging peripheral intravenous access, and peripherally inserted central catheters should be considered where multiple procedures, blood transfusion, or a prolonged hospital stay is likely.

Particular preparation for anticoagulation

De Barsy patients are not known to present problems with coagulation or surgical haemostasis.

Particular precautions for positioning, transport or mobilization

Patients with De Barsy syndrome have increased joint ligament and tendon laxity, in addition to reduced subcutaneous fat padding. These conditions may increase the risk for peripheral nerve injury and pose musculoskeletal problems with intraoperative positioning.

Awake alignment in positions of comfort may be prudent prior to induction of anaesthesia. No cases of perioperative nerve injury have been documented.

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

Unknown.

Anaesthesiologic procedure

- Regional block with bupivacaine
- Central line (femoral, right internal jugular)
- Arterial line (right and left radial)

Particular or additional monitoring

Both arterial and central line placement and monitoring have been used without complication in De Barsy syndrome patients. Close temperature monitoring is also recommended since these patients have been found to have elevated intraoperative temperatures.

Possible complications

Intraoperative hyperthermia has been reported (Aponte, Smith et al 2010) in approximately 10% of cases with temperatures exceeding 38 degrees Celsius. While these cases had no evidence of malignant hyperthermia (muscle rigidity or end-tidal carbon dioxide increases) they were associated with tachycardia. Consequently, close monitoring of body temperature is warranted. While patient overwarming via forced air devices or heat lamps may explain patient pyrexia, it is possible that these cases represent a form of non-malignant hyperthermia similar to that manifested in patients with other congenital diseases states such as Costello syndrome and osteogenesis imperfect (Dearlove, 1997) (Furderer, 2000).

Younger patients may have urea cycle disturbances leading to hyperammonemia and amino acid disturbances (hypoornithinemia, hypocitrullinemia, hypoargininemia and hypoprolinemia). This could be a potential cause of delayed awakening though this has never been reported in the literature (National Organization for Rare Disorders). If patients have feeding problems (gastrostomy or feeding by PEG) there may also be nutritional deficits.

Postoperative care

De Barsy syndrome patients have functional cognitive limitations which pose challenges in assessing post-operative pain. Providers should be aware that seizures and other involuntary movements are associated with this syndrome, and patients may have baseline writhing movements that may be confused with agitation and discomfort.

Information about emergency-like situations / Differential diagnostics

The differential diagnosis of intraoperative hyperthermia should always include malignant hyperthermia even though DBS and MH have never been reported together.

Ambulatory anaesthesia

Ambulatory anaesthesia has been performed without complications for radiologic studies such as magnetic resonance imaging and computed tomography.

Unknown.

Literature and internet links

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Please note that this guideline has not been reviewed by an anaesthesiologist, but by two disease experts instead.