orphanaesthesia

Anesthesia recommendations for

De Barsy Syndrome

Disease name: De Barsy syndrome

ICD 10: Q87.7

OMIM: 614438

ORPHAcode: 2962

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)

Disease summary: 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 orthopedic 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 [1]. The diverse collection of clinical manifestations in De Barsy syndrome includes: intra-uterine growth retardation (IUGR), postnatal growth delay, motor delay, cognitive impairment, hypotonia, athetoid movements, malformations, microcephaly, wormian bones (extra cranial small bones and sutures), large fontanelles, facial dysmorphism, cataracts, corneal clouding, thin/wrinkled skin, easy bruising, sparse hair, joint laxity, osteopenia, and inguinal hernias.

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

A	**AIRWAY / ANESTHETIC TECHNIQUE**	Recommend video laryngoscopy as facial features include midface hypoplasia and microcephaly. Associated tracheomalacia, sleep apnea and restrictive lung disease. No reported issues with GA or RA.
B	**BLOOD PRODUCTS (COAGULATION)**	No known special necessary product formulations. Vascular fragility can lead to easy bruising. No known coagulation or hemostasis problems.
C	**CIRCULATION**	Cases of aortic root dilation, double aortic arch, vascular ring formation and cardiomyopathy reported. Consider preoperative ECHO and EKG. Challenging peripheral IV access reported.
D	**DRUGS**	Unknown interactions with patient's long-term medications and anesthetic agents. No reported cases of MH.
E	**EQUIPMENT**	Caution peripheral nerve injury with positioning. Monitor temperature closely as there are known cases with intraoperative hyperthermia.

Typical surgery and procedures

- Ophthalmologic procedures (cataract, eye exam)
- Orthopedic procedures (hip arthrogram/open reduction, joint stabilization, spinal fusions, capsulodesis)
- Skin biopsy
- Wound check
- Radiologic imaging (MRI)
- GI procedures (Nissen fundoplication, g-tube placement, EGD)
- ENT procedures (myringotomy and tube placement)
- Urologic procedures (orchiopexy, circumcision)
- Hernia repair

Type of anesthesia

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

Necessary additional preoperative testing (beside standard care)

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 ECG 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 [2,3].

In another published case series of De Barsy patients [1], 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 [4-2]. Midface hypoplasia and microphephaly malformations suggest caution with regard to airway manipulation. However, during 64 anesthetics 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 preparation 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 hemostasis.

Particular precautions for positioning, transportation and 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 anesthesia. No cases of perioperative nerve injury have been documented.

Interactions of chronic disease and anesthesia medications

Unknown.

Anesthetic 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 in approximately 10% of cases with temperatures exceeding 38ºC degrees Celsius [1]. 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 [11,12].

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 postoperative 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.

Disease-related acute problems and effect on anesthesia and recovery

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

Ambulatory anesthesia

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

Obstetrical anesthesia

Unknown.

References

1. Aponte EP, Smith HM, Wanek BJ and Rettke SR. Anesthesia considerations for patients with de Barsy syndrome. J Clin Anesth 2010;22:499-504
2. Lin DS, et al. Compound heterozygous mutations in PYCR1 further expand the phenotypic spectrum of de Barsy syndrome. Am J Genet A 2011;155a :3095-3099
3. Dutta AK, et al. De Barsy syndrome type B presenting with cardiac and genitourinary abnormalities. Clin Dysmorphol 2016;25:190-191
4. Aldave AJ, et al. Congenital corneal opacification in De Barsy syndrome. Arch Ophthalmol 2001;119:285-288
5. Al-Owain M, et al. A case of de Barsy syndrome with a severe eye phenotype. Am J Med Genet A 2012;158:2364-2366
6. Arazi M, Kapicioglu MI and Mutlu M. The de Barsy syndrome. Turk J Pediatr 2001;43:79-84
7. Bartsocas CS, et al. De Barsy syndrome. Prog clin Biol Res 1982;104:157-60
8. Burck, U. De Barsy syndrome, a further case. Klin Padiatr 1974;186:441-444
9. Dutta AK, et al. De Barsy syndrome type B presenting with cardiac and genitourinary abnormalities. Clin Dysmorphol 2016;25:190-191
10. Dimopoulou A, Fischer B, Gardeitchik T, et al. Genotype-phenotype spectrum of PYCR1-related autosomal recessive cutis laxa. Mol Genet Metab. 2013;110 :352-361
11. Dearlove O, Harper N. Costello syndrome. Paediatr Anaesth 1997;7:476-477
12. Furderer S, Stanek A, Karbowski A and Eckardt A. Intraoperative hyperpyrexia in patients with osteogenesis imperfecta. Z Orthop Ihre Grenzgeb 2000;138:136-139
13. Guerra D, et al. The de Barsy syndrome. J Cutan Pathol 2004;31:616-624
14. Hoekx J, et al. The de Barsy syndrome. Tijdschr Kindergeneeskd 1989;57:43-57
15. Karnes PS, et al. De Barsy syndrome : report of a case, literature review, and elastin gene expression studies of the skin. Am J Med Genet 1992;42:29-34
16. Kivuva EC, et al. De Barsy syndrome: a review of the phenotype. Clin Dysmorphol 2008;17:99-107
17. Kunze J, et al. De Barsy syndrome- an autosomal recessive, progeroid syndrome. Eur J Pediatr 1985;144:348-354
18. Leao-Teles E, et al. De Barsy syndrome and ATP6V0A2-CDG. Eur J Hum Genet 2010;18:526
19. Pontz BF, et al. Biochemical, morphological and immunological findings in a patient with a cutis laxa-associated inborn disorder (de Barsy syndrome). Eur J Pediatr 1986;145:428-434
20. Stanton RP, et al. Orthopaedic manifestations in de Barsy syndrome. J Pedatr Orthop 1994;14:60-62
21. Sybert VP. Genetic Skin Disorders 2nd ed. Oxford University Press NY, NY 2010:644-646
22. Wolthuis DF, van Asbeck E, Mohamed M, et al. Cutis laxa, fat pads and retinopathy due to ALDH18A1 mutation and review of the literature. Eur J Paediatr Neurol 2014;18:511-514

23. Zampatti S, et al. De Barsy syndrome : a genetically heterogenous autosomal recessive cutis laxa syndrome related to P5CS and PYCR1 dysfunction. Am J Med Genet A 2012;158a :927- 931

24. From internet: Eva Morava, MD, PhD. De Barsy Syndrome. National Organization For Rare Disorders. Available at: http://rarediseases.org/rare-diseases/de-barsy-syndrome/ Accessed on: August 5th, 2016 .

Date last modified: **February 2026**

This recommendation was prepared in October 2017 by:

Author(s)
Hugh M. Smith, Anesthesiologist, Mayo Clinic Rochester, Minnesota, USA
smith.hugh2@mayo.edu

Lindsay Warner, Anesthesiologist, Mayo Clinic Rochester, Minnesota, USA
Warner.Lindsay@mayo.edu

Disclosure: The authors has no financial or other competing interest to disclose. This recommendation was unfunded.

This recommendation was reviewed by:

Reviewer(s)
Robert Śmigiel, Specialist of pediatrics and neonatology, Specialist of clinical genetics, Department of Pediatrics and Rare Disorders Wroclaw Medical University, Wroclaw, Poland
robert.smigiel@umed.wroc.pl

Fowzan S. Alkuraya, Professor of Human Genetics, Principal Clinical Scientist and Senior Consultant, Head, Developmental Genetics Unit, King Faisal Specialist Hospital and Research Center, Riyadh, Saudi Arabia
falkuraya@kfshrc.edu.sa

Saud Alsahli, Department of Genetics, King Faisal Specialist Hospital and Research Center, Riyadh, Saudi Arabia
alsahlisauda@gmail.com

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

Disclosure: The reviewers have no financial or other competing interest to disclose.

Update and revision (2026)
Elizabeth McKenna, Pediatric Anesthesia Fellow, Mayo Clinic Rochester, Minnesota, USA
Mckenna.elizabeth@mayo.edu

Lindsay Warner, Pediatric Anesthesiologist, Mayo Clinic Rochester, Minnesota, USA
Warner.Lindsay@mayo.edu

Reviewer
Carlos L. Errando, Anesthesiologist, Consortium General University Hospital of Valencia, València, Spain

Editorial Review
Christine Gaik, Anesthesiologist, Department of Anesthesiology and Intensive Care Medicine, University Hospital Giessen and Marburg, Campus Marburg and Philipps University of Marburg, Germany
gaikc@med.uni-marburg.de