Aktiv Druck & Verlag GmbH | ISSN 0170 - 5334 I 02330

www.ai-online.info

59. Jahrgang | Juli/August 2018

# A&I

# ANÄSTHESIOLOGIE & INTENSIVMEDIZIN

Offizielles Organ: Deutsche Gesellschaft für Anästhesiologie und Intensivmedizin e.V. (DGAI)
Berufsverband Deutscher Anästhesisten e.V. (BDA)
Deutsche Akademie für Anästhesiologische Fortbildung e.V. (DAAF)
Organ: Deutsche Interdisziplinäre Vereinigung für Intensiv- und Notfallmedizin e.V. (DIVI)

## Sickle cell disease

## Smith-McCort dysplasia (SMC)

orphanaesthesia

a project of the German Society
of Anaesthesiology and Intensive Care Medicine

## SUPPLEMENT NR. 12 | 2018

## OrphanAnesthesia –
### ein krankheitsübergreifendes Projekt des Wissenschaftlichen Arbeitskreises Kinderanästhesie der Deutschen Gesellschaft für Anästhesiologie und Intensivmedizin e.V.

Ziel des Projektes ist die Veröffentlichung von Handlungsempfehlungen zur anästhesiologischen Betreuung von Patienten mit seltenen Erkrankungen. Damit will Orphan Anesthesia einen wichtigen Beitrag zur Erhöhung der Patientensicherheit leisten.

Patienten mit seltenen Erkrankungen benötigen für verschiedene diagnostische oder therapeutische Prozeduren eine anästhesiologische Betreuung, die mit einem erhöhten Risiko für anästhesieassoziierte Komplikationen einhergehen. Weil diese Erkrankungen selten auftreten, können Anästhesisten damit keine Erfahrungen gesammelt haben, so dass für die Planung der Narkose die Einholung weiterer Information unerlässlich ist. Durch vorhandene spezifische Informationen kann die Inzidenz von mit der Narkose assoziierten Komplikationen gesenkt werden. Zur Verfügung stehendes Wissen schafft Sicherheit im Prozess der Patientenversorgung.

Die Handlungsempfehlungen von OrphanAnesthesia sind standardisiert und durchlaufen nach ihrer Erstellung einen Peer-Review-Prozess, an dem ein Anästhesist sowie ein weiterer Krankheitsexperte (z.B. Pädiater oder Neurologe) beteiligt sind. Das Projekt ist international ausgerichtet, so dass die Handlungsempfehlungen grundsätzlich in englischer Sprache veröffentlicht werden.

Ab Heft 5/2014 werden im monatlichen Rhythmus je zwei Handlungsempfehlungen als Supplement der A&I unter www.ai-online.info veröffentlicht. Als Bestandteil der A&I sind die Handlungsempfehlungen damit auch zitierfähig. Sonderdrucke können gegen Entgelt bestellt werden.

**Bisher in A&I publizierte Handlungsempfehlungen finden Sie unter:**

**www.ai-online.info/Orphsuppl**
**www.orphananesthesia.eu**

## OrphanAnesthesia –
### a common project of the Scientific Working Group of Paediatric Anaesthesia of the German Society of Anaesthesiology and Intensive Care Medicine

The target of OrphanAnesthesia is the publication of anaesthesia recommendations for patients suffering from rare diseases in order to improve patients' safety. When it comes to the management of patients with rare diseases, there are only sparse evidence-based facts and even far less knowledge in the anaesthetic outcome. OrphanAnesthesia would like to merge this knowledge based on scientific publications and proven experience of specialists making it available for physicians worldwide free of charge.

All OrphanAnesthesia recommendations are standardized and need to pass a peer review process. They are being reviewed by at least one anaesthesiologist and another disease expert (e.g. paediatrician or neurologist) involved in the treatment of this group of patients.

The project OrphanAnesthesia is internationally oriented. Thus all recommendations will be published in English.

Starting with issue 5/2014, we'll publish the OrphanAnesthesia recommenations as a monthly supplement of A&I (Anästhesiologie & Intensivmedizin). Thus they can be accessed and downloaded via www.ai-online.info. As being part of the journal, the recommendations will be quotable. Reprints can be ordered for payment.

**A survey of until now in A&I published guidelines can be found on:**

**www.ai-online.info/Orphsuppl**
**www.orphananesthesia.eu**

**Projektleitung**

**Prof. Dr. Tino Münster, MHBA**
Geschäftsführender Oberarzt
Facharzt für Anästhesie,
Spezielle Schmerztherapie,
Notfallmedizin
Anästhesiologische Klinik
Friedrich-Alexander-Universität
Erlangen-Nürnberg
Krankenhausstraße 12
91054 Erlangen, Deutschland
Tel.: 09131 8542441
Fax: 09131 8536147
E-Mail: muenster@kfa.imed.uni-erlangen.de

Deutsche Gesellschaft für Anästhesiologie & Intensivmedizin
**www.dgai.de**

A&I
ANÄSTHESIOLOGIE & INTENSIVMEDIZIN
**www.ai-online.info**

orphanästhesia

# Anaesthesia recommendations for patients suffering from

# Smith-McCort dysplasia (SMC)

**Disease name:** Smith-McCort dysplasia (SMC)

**ICD 10:** -

**Synonyms:** Smith-McCort dwarfism, Orpha no: ORPHA178355

SMC, which was first described in 1958, is an autosomal recessive skeletal dysplasia. The disease bears similarity to Dyggve-Melchior-Clausen (DMC) syndrome except that patients with SMC have normal intelligence function, whereas patients with DMC have moderate to severe intellectual disabilities. The DMC gene is mapped to the 18q12-12.1 chromosomal region. SMC (OMIM: 607326) also mapped to this same region and as it turns out both conditions have mutations in the DYM gene. DMC results from mutations in the gene that results in loss of function, whereas in SMC, the mutation is a missense mutation resulting in residual DYM activity, thus a less severe phenotype. The gene codes for a novel protein known as Dymeclin, which is involved in Golgi organisation and intracellular trafficking.

Recently, another gene, RAB33B, which decreases the level of another Golgi protein that is involved in retrograde transport of Golgi vesicles, has been implicated in the pathogenesis of SMC.

Medicine in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnosis is wrong

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

▶ **Citation:** Bansal S, Umamaheswara Rao GS: Smith-McCort dysplasia (SMC). Anästh Intensivmed 2018;59:S494-S499. DOI: 10.19224/ai2018.S494

## Disease summary

Patients with SMC have increased glycosaminoglycans (GAGs) in their urine but have normal electrophoretic patterns and enzyme assays. Lysosomal pathways are also normal in patients with SMC.

Patients with this syndrome have a severe generalised bony dysplasia with a short trunk, short stature, coarse facies, a short neck and a protuberant abdomen. Specific skeletal abnormalities include hypoplastic odontoid, pectus carinatum, kyphoscoliosis, platyspondyly, exaggerated lumbar lordosis, flared ribs, widened costochondral junction, small scapulas with concave inferior angles, small pelvis, hypoplastic acetabulums with small ilium, dislocated hips, widened symphysis pubis, deformed and displaced femoral neck, genu valgum or varum, rhizomelic limb shortening and limited joint extensibility. Pathognomic radiologic features include double-humped vertebral end plates and lace-like appearance of iliac crests.

## Typical surgery

Atlantoaxial dislocation-induced spinal cord compression is a serious and treatable problem in these patients. Common surgeries for which these patients might present include posterior cervical spine fusion, orthopaedic femoral osteotomy, total hip arthroplasty, early meniscectomy and realignment osteotomy. However, we recommend postponement of any surgery (unless emergent) till adolescence in view of risks associated with anaesthesia, and doubtful long term results in case of orthopaedic surgeries.

## Type of anaesthesia

No particular recommendations are available regarding the type of anaesthesia, i.e., general versus regional. Congenital spine abnormalities must be ruled out if spinal or epidural block is planned. Moreover, if associated cervical spine disease has led to muscle weakness or spasticity, the same should be documented before contemplating regional anaesthesia.

## Necessary additional diagnostic procedures (preoperative)

Chest radiographs and pulmonary function tests should be done if there are co-existing kyphoscoliosis and chest abnormalities. A blood gas analysis on room air may be useful for comparison at a later stage. Electrocardiography and echocardiography will help to rule out any congenital cardiac anomalies or cardiac dysfunction.

## Particular preparation for airway management

A difficult airway may be expected in patients with Smith-McCort dysplasia as these individuals may have macroglossia, short neck and abnormality of neck flexion. But surprisingly, laryngoscopy may not be problematic. However, intubation may be possible only with an endotracheal tube smaller than expected. Therefore, several sizes of endotracheal tubes should be readily available.



© Anästh Intensivmed 2018;59:S494-S499 Aktiv Druck & Verlag GmbH

In a case report of anaesthesia in a patient with DMC, the authors did not find any airway difficulties. With one SMC patient with whom we have been involved, we anticipated difficulty with anaesthesia and planned for fibre-optic intubation. However, on direct laryngoscopy, the vocal cords could be visualised without difficulty. We, however, could negotiate only a 5mm uncuffed endotracheal tube into this 18-year-old patient.

In cases where a tracheostomy is required, post tracheostomy airway problems should be anticipated. In SMC patients, the biggest challenge is maintaining the position of the artificial airway because of distorted airway anatomy. Hence, wherever possible, direct extubation is preferable and tracheostomy should be performed only in unavoidable circumstances. Once a tracheostomy has been performed, all measures should be directed at appropriate positioning to prevent accidental misalignment of the tracheostomy tube.

## Particular preparation for transfusion or administration of blood products

No recommendations are available.

## Particular preparation for anticoagulation

No recommendations are available.

## Particular precautions for positioning, transport or mobilisation

Not reported.

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

Not reported.

## Anaesthesiologic procedure

Because the patient with Smith-McCort dysplasia has dwarfism, anthropometric measurements such as height and weight should be recorded and drug doses should be modified accordingly. Baseline complete haemogramme and biochemistry profile should also be done.

## Particular or additional monitoring

Depending on the surgery and expected blood loss, arterial blood pressure monitoring can be used and adequate intravenous access should be obtained.



© Anästh Intensivmed 2018;59:S494-S499 Aktiv Druck & Verlag GmbH

## Possible complications

Extubation should be performed carefully and only when the patient is fully awake. Doing so will reduce the need for reintubation especially if the surgery involved fusion of the cervical spine or if airway oedema is expected post procedure, situations where reintubation could be even more difficult.

## Postoperative care

The degree of postoperative monitoring will depend on the surgical procedure, the preoperative condition of the patient and the intraoperative course. When postoperative ventilation is needed, avoid prolonged ventilation and attempt to wean rapidly. Extubation should be attempted at the earliest possibility and tracheostomy should not be chosen as an easy alternative.

## 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 disease*

Morquio syndrome (mucopolysaccharidosis type IVA): This disorder has similar clinical and radiologic findings but urinary mucopolysaccharide secretion is normal in SMC. Patients with this syndrome usually have corneal clouding, deafness and cardiac anomalies. Morquio syndrome is caused by a deficiency of N-acetyl-galactose-6 sulfatase enzyme. In both conditions, hypoplastic odontoid can be present.

Hurler syndrome (MPS type I): Occurs due to deficiency of α-L-iduronidase. Individuals with this condition also excrete dermatan sulphate and heparin sulphate in their urine.

Spondyloepimetaphyseal dysplasias (SEMDs): There is absence of the vertebral double hump with central constriction. In SEMDs, some vertebras show anterior pointing with narrow disc spaces, premature degenerative changes and no odontoid hypoplasia.

Achondroplasia: The vertebras are short and flat, pedicles are short and the spinal canal is narrow.

Metatropic dysplasia (MD): This condition causes general shortening in all long and short tubular bones. The lace-like appearance of iliac crests in SMC is a significant sign for distinguishing between MD and SMC.

## Ambulatory anaesthesia

Not reported.

## Obstetrical anaesthesia

Not reported.

**Literature and internet links**

1. Mona SA, Samia AT, Ekram F, et al. Dyggve–Melchior–Clausen syndrome: clinical, genetic, and radiological study of 15 Egyptian patients from nine unrelated families. J Child Orthop 2009;3:451-458
2. Cohn DH, Ehtesham N, Krakow D, et al. Mental retardation and abnormal skeletal development (Dyggve-Melchior-Clausen dysplasia) due to mutations in a novel evolutionarily conserved gene. Am J Hum Genet 2003;72:419-428
3. Nakamura K, Kurokawa T, Nagano A, Nakamura S, Taniguchi K, Hamazaki M. Dyggve-Melchior-Clausen syndrome without mental retardation (Smith-McCort dysplasia): morphological findings in the growth plate of the iliac crest. Am J Med Genet 1997;72:11-17
4. Bayrak IK, Nural MS, Diren HB. Dyggve-Melchior-Clausen syndrome without mental retardation (Smith- McCort dysplasia). Diagn Interv Radiol 2005;11:163-165
5. Kandziora F, Neumann L, Schnake KJ, et al. Atlantoaxial instability in Dyggve-Melchior-Clausen syndrome: Case report and review of the literature. J Neurosurg: Spine 2002; 96:112-117
6. Paupe V, Gilbert T, Merrer ML, et al. Recent advances in Dyggve-Melchior-Clausen syndrome. Moll Genet Metab 2004;83:51-59
7. Eguchi M, Kadota Y, Yoshida Y, Masuda M, Masuyama T, Kammura Y. Anesthetic management of a patient with Dyggve-Melchior-Clausen syndrome. Masui 2001;50:1116-1117
8. Schorr S, Legum C, Ochshorn M, Hirsch M, Moses S, Lasch EE, El- Masri M. The Dyggve-Melchior-Clausen Syndrome. Am J Roentgenol 1977;128:107-113
9. Dupuis N, Lebon S, Kumar M, Drunat S, Graul-Neumann LM, Gressens P, El Ghouzzi V. A novel RAB33B mutation in Smith-McCort dysplasia. Hum Mutat 2013;34:283-286.

**Last date of modification: February 2015**

*These guidelines have been prepared by:*

**Authors**
**Sonia Bansal,** Anaesthesiologist, National Institute of Mental Health and Neurosciences, Bangalore, India
rashibpgi@yahoo.co.in

**GS Umamaheswara Rao,** Anaesthesiologist, National Institute of Mental Health and Neurosciences, Bangalore, India

**Peer revision 1**
**Serge Dalmas**, Anaesthesiologist, Hôpital Jeanne de Flandre, Lille, France

**Eric Nectoux,** Département de Chirurgie et Orthopédie de l'Enfant, Hôpital Jeanne de Flandre, Lille, France
Eric.nectoux@chru-lille.fr

**Peer revision 2**
**David Weaver**, Department of Medical and Molecular Genetics, Indiana University School of Medicine, Indianapolis, USA
dweaver@iupui.edu



© Anästh Intensivmed 2018;59:S494-S499 Aktiv Druck & Verlag GmbH

## Herausgeber

**DGAI**
Deutsche Gesellschaft für Anästhesiologie und Intensivmedizin e.V.
Präsident: Prof. Dr. B. Zwißler, München

**BDA**
Berufsverband Deutscher Anästhesisten e.V.
Präsident: Prof. Dr. G. Geldner, Ludwigsburg

**DAAF**
Deutsche Akademie für Anästhesiologische Fortbildung e.V.
Präsident: Prof. Dr. F. Wappler, Köln



**Redaktion**
Carolin Sofia Kopp B.A. &
Dipl.-Sozw. Holger Sorgatz

Korrespondenzadresse: Roritzerstraße 27 | 90419 Nürnberg | Deutschland
Tel.: 0911 9337812 | Fax: 0911 3938195
E-Mail: anaesth.intensivmed@dgai-ev.de

## Verlag & Druckerei

**Aktiv Druck & Verlag GmbH**
An der Lohwiese 36 |
97500 Ebelsbach | Deutschland
www.aktiv-druck.de

**Geschäftsführung**
Wolfgang Schröder | Jan Schröder | Nadja Schwarz
Tel.: 09522 943560 | Fax: 09522 943567
E-Mail: info@aktiv-druck.de

**Anzeigen | Vertrieb**
Pia Engelhardt
Tel.: 09522 943570 | Fax: 09522 943577
E-Mail: anzeigen@aktiv-druck.de

**Verlagsrepräsentanz**
Jürgen Distler
Roritzerstraße 27, 90419 Nürnberg
Tel.: 0171 9432534 | Fax: 0911 3938195
E-Mail: jdistler@bda-ev.de

**Herstellung | Gestaltung**
Manfred Wuttke | Stefanie Triebert
Tel.: 09522 943571 | Fax: 09522 943577
E-Mail: ai@aktiv-druck.de

**Titelbild**
Dipl.-Designerin Monique Minde, Nürnberg

**Erscheinungsweise 2018**
Der 59. Jahrgang erscheint jeweils zum Monatsanfang, Heft 7/8 als Doppelausgabe.

**Bezugspreise** (inkl. Versandkosten):

- **Einzelhefte** 30,- €
- **Jahresabonnement:**
  Europa (ohne Schweiz) (inkl. 7 % MwSt.) 258,- €
  Schweiz 266,- €
  Rest der Welt 241,- €

**Mitarbeiter aus Pflege, Labor, Studenten und Auszubildende** (bei Vorlage eines entsprechenden Nachweises)
Europa (ohne Schweiz) (inkl. 7 % MwSt.) 94,- €
Schweiz 90,- €
Rest der Welt 94,- €

**Für Mitglieder der DGAI und/oder des BDA ist der Bezug der Zeitschrift im Mitgliedsbeitrag enthalten.**

**Allgemeine Geschäfts- und Lieferbedingungen**
Die allgemeinen Geschäfts- und Lieferbedingungen entnehmen Sie bitte dem Impressum auf www.ai-online.info

Indexed in **Current Contents®/Clinical Medicine, EMBASE/Excerpta Medica; Medical Documentation Service; Research Alert; Sci Search; SUBIS Current Awareness in Biomedicine; VINITI: Russian Academy of Science.**



**Wichtiger Hinweis**
Für Angaben über Dosierungsanweisungen und Applikationsformen kann vom Verlag und den Herausgebern keine Gewähr übernommen werden. Derartige Angaben müssen vom jeweiligen Anwender im Einzelfall anhand anderer Literaturstellen auf ihre Richtigkeit überprüft werden. Gleiches gilt für berufs- und verbandspolitische Stellungnahmen und Empfehlungen.

Online-Ausgabe der A&I ab April 2017 open access: **www.ai-online.info**

# CONTACT US

Please do not hesitate to contact us. We will be glad to answer and provide further information to you at any time.

.....
Name

.....
First Name

.....
Department / Hospital

.....
Place

.....
Telephone

.....
E-Mail

.....
Date / Signature

Please contact me for further information ○

I would like to participate in the project ○

## ADDRESS

German Society of Anaesthesiology and
Intensive Care Medicine
Nina Schnabel
Roritzerstrasse 27 | 90419 Nuremberg | Germany
Tel.: +49-911-9337822 | Fax: +49-911-3938195
Email: nschnabel@orphananesthesia.eu