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Dyskeratosis congenita

orphan**anesthesia**

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

SUPPLEMENT NR. 12 | 2026

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 Patientinnen und Patienten mit seltenen Erkrankungen. Damit will OrphanAnesthesia einen wichtigen Beitrag zur Erhöhung der Patientensicherheit leisten.

Patientinnen und 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 Anästhesistinnen und Anästhesisten damit keine Erfahrungen gesammelt haben, sodass 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 eine Anästhesistin bzw. ein Anästhesist sowie eine weitere Krankheitsexpertin bzw. ein weiterer Krankheitsexperte (z. B. Pädiaterin bzw. Pädiater oder Neurologin bzw. Neurologe) beteiligt sind. Das Projekt ist international ausgerichtet, sodass 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.

OrphanAnesthesia –

a 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 recommendations 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.

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Projektleitung

**Dr. med. Christine Gaik,
DESAIC, FESAIC**

Oberärztin
Klinik für Anästhesie und
Intensivtherapie
Universitätsklinikum Gießen
und Marburg GmbH,
Standort Marburg
Baldingerstraße
35033 Marburg, Deutschland
E-Mail:
gaikc@med.uni-marburg.de

**Priv.-Doz. Dr. med.
Philipp Gude, MHBA**

Geschäftsführender Oberarzt
Klinik für Anästhesiologie und
Intensivmedizin
Universitätsklinikum
St. Elisabeth-Hospital
Bleichstraße 15
44787 Bochum, Deutschland
E-Mail:
philipp.gude@ruhr-uni-bochum.de

orphan**a**nesthesia

Anesthesia recommendations for

Dyskeratosis congenita

Disease name: Dyskeratosis congenita

ICD 10: Q82.8

Synonyms: Zinsser-Engman-Cole syndrome, Hoyeraal-Hreidarsson syndrome, Revesz syndrome, DC, DKC

Disease summary:

Dyskeratosis congenita (DC) is a rare disease of abnormal telomere biology, leading to hematopoietic failure among other heterogeneous multisystem manifestations.

There are multiple forms of the condition, with varying patterns of inheritance. There is a 3:1 male:female predilection, and an incidence of approximately 1:1 000 000. The most common gene mutation (*DKC1*) in the X-linked form of DC results in impaired telomere maintenance. Other disease genes have been implicated, with both autosomal dominant and recessive inheritance, including: *TERC*, *TINF2*, *ACD*, *RTEL1*, *TERT*, *CTC1*, *NHP2*, *NOP10*, *PARN*, *WRAP53*, *NAF1*, *SNT1*, *POT1*, and *ZCCHC8*. (1-5) As all of these genes are important in telomere maintenance, DC is now regarded as principally a disorder of defective telomere maintenance.

DC classically presents with the clinical triad of dysplastic nails, lacy reticular pigmentation of the upper chest/neck, and oral leukoplakia (white plaques) (2). Bone marrow failure is common (80%). DC is also associated with pulmonary fibrosis, pulmonary arteriovenous malformations, poor dentition, esophageal stenosis, cirrhosis, hepatopulmonary syndrome, vascular ectasias, urethral stenosis, peripheral neuropathy, immunodeficiency, and accelerated aging. Patients with DC have an elevated risk for leukemia as well as squamous cell cancers of the head and neck, skin, or anogenital regions (6). The treatment for DC is individualized according to symptoms. Hematopoietic stem cell transplant is considered in cases of bone marrow failure or leukemia (7.)

Medicine is in progress



Perhaps new knowledge

Every patient is unique

Perhaps the diagnosis is wrong

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

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Emergency information

A	AIRWAY / ANESTHETIC TECHNIQUE	Possible difficult intubation: poor dentition, friable oral tissue, mandibular hypoplasia, oropharyngeal tumors. Thrombocytopenia and coagulopathy may rule out neuraxial anesthesia.
B	BLOOD PRODUCTS (COAGULATION)	Blood products should be leukodepleted, irradiated (if patient is lymphopenic), CMV-negative. Stem cell transplant candidates should have minimal necessary exposure to red blood cells or platelets. Avoid blood donated by relatives. Maximize blood conservation strategies.
C	CIRCULATION	Hypoxemia: pulmonary fibrosis, pulmonary arteriovenous malformations, ascites.
D	DRUGS	No specific contraindication to anesthesia medications. No association with malignant hyperthermia.
E	EQUIPMENT	Standard monitors should be used as appropriate (e.g., electrocardiogram, non-invasive blood pressure, pulse oximetry, capnography, gas analyzer). Intraoperative transesophageal echocardiography may be indicated for patients with pulmonary hypertension and right heart failure. Possible esophageal strictures or varices.

Typical surgery and procedures

Dental procedures, ophthalmologic procedures, bone marrow aspirate, liver biopsy, gastroscopy, colonoscopy, head and neck tumor resection. There are also case reports of organ transplants in this patient population (i.e., liver, lung) (8-10).

Type of anesthesia

There are very few published case reports of anesthetic management in DC (10-14). General anesthesia has been described for patients with DC. Balanced anesthetic maintenance with volatiles, opioids, and/or propofol is effective.

Given the high propensity for bone marrow failure in this patient population, thrombocytopenia and coagulopathy must be ruled out prior to providing neuraxial anesthesia or performing regional anesthesia techniques with high risk for bleeding. Furthermore, due to the telomere defect, wound healing and tissue repair are not completely normal. Careful surgical technique is important for DC patients undergoing any operative procedure, and they often require a longer recovery time compared to non-DC patients.

Necessary additional pre-operative testing (beside standard care)

Neurologic examination and documentation of pre-existing peripheral neuropathies is recommended. A careful airway examination is also essential.

Full blood count (including platelet count), coagulation profile, type and screen should be obtained, as should standard liver and renal function tests.

Patients with poor exercise tolerance, known pulmonary fibrosis, or pulmonary arteriovenous malformations may warrant pulmonary function testing, room air arterial blood gas, and/or an echocardiographic study.

A preoperative hematology consult will guide perioperative medication and blood product management.

Particular preparation for airway management

Patients with DC may be at risk for difficult intubation. The underlying telomere defect commonly results in poor dentition and friable oral tissue. Oropharyngeal tumors may distort the airway anatomy. Mandibular hypoplasia has also been reported in DC.

Pulmonary disease is a common (20%) complication of DC (15). With pulmonary fibrosis, poor lung compliance and gas exchange may further complicate airway management and ventilation, as may concomitant pulmonary hypertension, shunting, and right heart failure. Pulmonary arteriovenous malformations also lead to hypoxemia in patients with DC. Liver dysfunction resulting in ascites may further limit safe apnea time.

A thorough airway examination is indicated. Adjuncts should be immediately available for safe tracheal intubation (e.g., supraglottic devices, video laryngoscope, flexible bronchoscope). Apneic oxygen may be advisable for patients with pulmonary complications and apnea intolerance. Efforts should be made to avoid traumatizing the oral mucosa, which may be prone to bleeding.

Particular preparation for transfusion or administration of blood products

Neutropenia, anemia, thrombocytopenia, and pancytopenia are commonly seen in DC. Preoperative evaluation of full blood count is essential, with transfusion thresholds individualized since chronically anemic patients may be adapted to lower hemoglobin levels. The immunomodulatory effects of blood product transfusion should be carefully considered in these immunocompromised patients.

Blood products should be leukodepleted, irradiated (if patient is lymphopenic), and CMV-negative. Avoid blood donations by relatives if stem cell transplant is being considered, as this may increase the risk of future graft rejection. Indeed, stem cell transplant candidates should have the minimal necessary exposure to red cells or platelets (2,16).

Blood conservation strategies are likely beneficial for this patient population. Consider administering tranexamic acid to avoid fibrinolysis (17). Use of topical hemostatic agents have not been described but may be useful.

Pre-operative administration of granulocyte colony-stimulating factor (GCSF) for treatment of leukopenia has been described (14). However, in patients taking androgen therapy for DC, concurrent GCSF administration has been implicated in life-threatening splenic rupture (18).

Particular preparation for anticoagulation

The need for perioperative anticoagulation must be weighed against bleeding risks and should be addressed in a multidisciplinary fashion.

Particular precautions for positioning, transportation and mobilization

Not reported.

Interactions of chronic disease and anesthesia medications

Not reported. Liver failure may be associated with prolonged elimination of anesthetic medications, such as opioids, benzodiazepines, and neuromuscular blocking agents. In such instances, choosing medications that are eliminated independently of liver function may result in more predictable offset of medication effects.

Commonly used anesthetic agents may be administered to patients with pulmonary complications of dyskeratosis congenita. Most halogenated volatile anesthetics have broncho- dilatory properties, although desflurane may cause bronchial irritation. At higher concentrations, volatile anesthetics reverse hypoxic pulmonary vasoconstriction and may worsen intrapulmonary shunting. Nitrous oxide may increase pulmonary vascular resistance and worsen pulmonary hypertension; additionally, it may confer risk of air trapping and pneumothorax in patients with pulmonary honeycomb cysts or bullae. Avoidance of excessive sedation and respiratory depression is advisable.

Anesthetic procedure

General anesthesia has been performed successfully in DC. There are no specific restrictions on the types of medications for induction or maintenance, though the presence of significant liver dysfunction may support the selection of agents that do not depend on hepatic clearance. Careful planning is necessary to ensure safe airway management. For patients with pulmonary fibrosis, a restrictive approach to fluid management is preferred and prolonged hyperoxygenation should be avoided, in order to avoid exacerbating alveolar dysfunction (19). Any decision to pursue elective surgery under general anesthesia should be carefully weighed against the risk of pulmonary exacerbation.

No reported literature on regional anesthesia in patients with DC. Given the risk of pulmonary complications, regional anesthesia may often be preferable to general anesthesia (19). Neuraxial anesthesia may be unsafe in the context of severe thrombocytopenia. However, regional techniques with low risk of bleeding may be considered. Continuous regional anesthesia catheters may be helpful for postoperative analgesia, but this must be weighed against the risk of infection in immunocompromised patients.

Dental procedures using local anesthesia have been described (20).

Overall, there are very few reports describing anesthetic management of patients with DC (10-14,20).

Particular or additional monitoring

Standard monitors should be used as appropriate (e.g., electrocardiogram, non-invasive blood pressure, pulse oximetry, capnography, gas analyzer).

Intraoperative transesophageal echocardiography may be indicated for patients with pulmonary hypertension and right heart failure, but esophageal probe placement may be difficult or prone to complications if there are esophageal strictures or varices. Invasive arterial blood pressure transduction and stroke volume variation may also guide fluid management.

Urethral stenosis may hinder insertion of urinary catheters.

Intraoperative use of thromboelastography has not been described but is likely helpful for managing acute hemorrhage.

Possible complications

Difficult airway management is a significant concern, as are postoperative pulmonary complications. Patients with poor pulmonary function may require postoperative ventilatory support.

Hemorrhage and opportunistic infection are complications relating to bone marrow failure.

Meticulous attention to aseptic technique and administration of surgical antibiotic prophylaxis are essential given immunocompromise.

Post-operative care

Patients with pulmonary dysfunction may require prolonged post-operative ventilatory support in the intensive care unit and may benefit from non-invasive positive pressure ventilation after extubation.

Continued dosing of tranexamic acid and antibiotic prophylaxis until 48 hours post-operatively has been reported (17).

Disease-related acute problems and effect on anesthesia and recovery

Airway management and ventilation considerations as mentioned previously.

Ambulatory anesthesia

Minor procedures can be considered as day cases, particularly in patients with limited systemic manifestations of DC. Local and regional anesthetic techniques are preferred in patients with pulmonary compromise (19).

Obstetrical anesthesia

No reported literature on obstetrical anesthesia in patients with DC. Pregnancy may be associated with new pancytopenia or worsening of existing cytopenias (1).

There must be careful consideration of risks associated with neuraxial anesthesia (bleeding disorder) versus general anesthesia (difficult airway).

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Date last modified: October 2025

This recommendation was prepared by:

Authors

Mike Wong, Department of Anesthesia, Pain Management & Perioperative Medicine,
Dalhousie University, Saint John, New Brunswick, Canada
mike.wong@dal.ca

Chrison Wong, Department of Anesthesia, Pain Management & Perioperative Medicine,
Dalhousie University, Halifax, Nova Scotia, Canada

Bruce Macaulay, Department of Anesthesia, Pain Management & Perioperative Medicine,
Dalhousie University, Halifax, Nova Scotia, Canada

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This recommendation was reviewed by:

Reviewer 1

Inderjeet Dokal, Paediatrician, Centre for Genomics and Child Health Blizard Institute, Barts and The London School of Medicine and Dentistry, Queen Mary University of London, UK
i.dokal@qmul.ac.uk

Reviewer 2

Sharon A. Savage, Paediatrician and Paediatric Haematologist, Division of Cancer Epidemiology and Genetics, National Cancer Institute, Bethesda, USA

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Authors

Mike Wong, Department of Anesthesia, Pain Management & Perioperative Medicine,
Dalhousie University, Saint John, New Brunswick, Canada
mike.wong@dal.ca

Reviewer

Martin Jöhr, Former head of the Division of Pediatric Anaesthesia, Luzerne Hospital (LUKS), Adligenswil, Switzerland
joehrmartin@bluewin.ch

Editorial Review

Christine Gaik, Specialist in anaesthesia and intensive care medicine, University Hospital Marburg, Germany
gaikc@med.uni-marburg.de

Philipp Gude, Specialist in anaesthesia and intensive care medicine,
Ruhr University Bochum, Bochum, Germany
St. Elisabeth-Hospital, Bochum, Germany
philipp.gude@kklbo.de

Herausgeber



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Redaktion/Editorial Staff

Korrespondenzadresse:

Neuwieder Straße 9 | 90411 Nürnberg |

Deutschland | Tel.: 0911 9337812

E-Mail: redaktion@ai-online.info

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Geschäftsführung

Wolfgang Schröder | Jan Schröder |

Tel.: 09522 9435-0 | Fax: 09522 9435-67

E-Mail: info@aktiv-druck.de

Anzeigen | Vertrieb

Pia Müller | Tel.: 09522 9435-70

E-Mail: anzeigen@aktiv-druck.de

Verlagsrepräsentanz

Jürgen Distler

Neuwieder Straße 9 | 90411 Nürnberg

Tel.: 0171 9432534

E-Mail: jdistler@bda-ev.de

Herstellung | Gestaltung

Robert Kux | Tel.: 09522 9435-71

E-Mail: ai@aktiv-druck.de

Titelbild

Gestaltung: Roland Mehl

3D Arts GmbH

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E-Mail: rmehl@3darts.de

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German Society of Anaesthesiology and
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Tel.: +49-911-933780
Email: info@orphananesthesia.eu