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Ellis-van Creveld syndrome

orphan**anesthesia**

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

SUPPLEMENT NR. 20 | 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.

Bisher in A&I publizierte
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Find a survey of the recommendations published until now on:

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orphan**a**nesthesia

Anesthesia recommendations for Ellis-van Creveld syndrome

Disease name: Ellis-van Creveld syndrome (EVC)

ICD 10: Q77.6

ORPHAcode: 289

Synonyms: Chondroectodermal dysplasia, Mesodermic dysplasia

Disease summary: Ellis-van Creveld syndrome is a rare autosomal recessive genetic disorder with an unknown exact prevalence. Approximately 150 cases have been reported in the literature over the last 50 years, with a higher incidence notably in the Amish population. Clinically, the syndrome is characterized by a tetrad of features: short stature with disproportionately short limbs, postaxial polydactyly (extra fingers and/or toes), ectodermal dysplasia manifesting as dysplastic nails and dental anomalies, and a high prevalence of congenital heart defects, primarily atrial septal abnormalities such as common atrium [1-5].

The disease is caused by mutations in the EVC, EVC2, and less frequently DYNC2L1 or other genes. These genes play critical roles in the structure and function of the primary cilium, a cellular organelle essential for signal transduction during embryonic development of cartilage and ectodermal tissues. Recent molecular genetic studies have reinforced the classification of EVC as a ciliopathy and pave the way for potential targeted therapeutic interventions [1].

A hallmark of the syndrome is a narrow chest due to shortened ribs, which results in reduced thoracic compliance and often severe postnatal respiratory distress. This respiratory impairment is a major source of morbidity and frequently demands intensive neonatal and pediatric respiratory support.

In addition to the cardinal skeletal and cardiac manifestations [6-8], systemic features such as mild developmental delay and distinctive oral abnormalities (including lip and palate malformations, absent or malformed teeth) are described [9,10]. Current treatment remains supportive and multidisciplinary. Cardiac surgery to correct congenital defects – usually undertaken in childhood – is standard care. Pulmonary management, including mechanical ventilation when needed, is life-saving. Advances in surgical techniques and intensive care over recent years have improved long-term survival rates, especially in patients with significant cardiopulmonary complications [5-7].

During childhood and adulthood, patients undergo frequently surgical procedures due to cardiac abnormalities [2,3], dental problems as well as orthopedic disabilities. Life expectancy is impaired mainly due to the severity of respiratory distress as well as congenital heart disease in EVC patients [1-5]. Perioperative medicine (e.g., surgical procedures, anaesthesia) should be performed after careful evaluation of the patient and their specific clinical features.

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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

Emergency information

A	AIRWAY / ANESTHETIC TECHNIQUE	Higher risk of difficult airway status. Use smaller ET or laryngeal mask than usual. Higher risk for failure in spinal & epidural anesthesia.
B	BLOOD PRODUCTS (COAGULATION)	No disease-specific precautions needed.
C	CIRCULATION	High incidences of congenital heart disease & anomalies High incidences of restrictive lung disease.
D	DRUGS	No MH risk, no specific disease-specific risk when using drugs.
E	EQUIPMENT	Preparation for respiratory failure in patients with restrictive lung disorder. Prolonged PACU stay recommended, higher risk for pulmonary complications.

Typical surgery and procedures

In neonatal patients:

Cardiosurgical procedures for correction of cardiac abnormalities (mainly atrial septation defects, frequently with multiple coexisting malformations like hypoplastic left ventricle, VSD were reported) [6-8,11,12].

In childhood and adulthood:

Cardiosurgical procedures for correction of cardiac abnormalities (mainly atrial septation defects, common atrium, AV septal defects. Sometimes with coexisting cardiac malformations) [1,6,11].

Dental surgery due to hypoplastic or absent teeth, orthopedic surgery (due to bone deformities like progressive valgus deformity, profound contractures, lateral patellar subluxation). Amputation of supernumerary fingers / toes (due to medical / anthropological reasons) [7,9,10,13-18].

Type of anesthesia

A general recommendation regarding an ideal anesthetic approach cannot be given, as both general anesthesia and regional anesthesia techniques might present potential problems in

specific EVC patients. An individualized perioperative plan undertaken by an experienced anesthesiologist taking into account the surgical needs and the patient's needs is the optimal technique.

General anesthesia might be difficult due to the respiratory and cardiac status of the patient with EVC. Airway management should be performed after careful evaluation of difficulties for potential intubation or mask ventilation which are mainly unrelated to the Ellis-van Creveld syndrome.

Neuraxial regional anesthesia techniques might be difficult due to skeletal abnormalities like lumbar lordosis or (but only in few patients) scoliosis. There is however no specific contraindication against neuraxial blockade in patients with EVC. One case report describes problems with an obstructed epidural catheter in a young man after uncomplicated placement. The obstruction was linked with some spinal abnormalities including short pedicles and a "very narrow bony canal" [7]. Catheter removal was only possible after maximal back flexion.

Peripheral regional anesthesia techniques should be performable in most patients and be beneficial especially in patients with reduced cardiac and pulmonary function. Due to deformities in the extremities a landmark-guided approach is discouraged for peripheral nerve blocks. An ultrasound examination will help to find the relevant structures for nerve blocks.

Necessary additional preoperative testing (beside standard care)

A thorough evaluation of the patient's history should focus on cardiac problems (corrected or uncorrected cardiac abnormalities), respiratory status (stable/unstable respiratory dysfunction). Specific laboratory results are usually not helpful in preoperative evaluation if no specific questions arise from medical history or clinical examination (e.g., potential bleeding disorder in the anamnesis unrelated to EVC).

Transthoracic echocardiography is recommended in all patients (due to the high prevalence of cardiac abnormalities), in stable patients who underwent regular consultations with cardiology, previous echocardiography results may be helpful to tailor an optimal anesthetic approach (general anesthesia vs. regional anesthesia).

Particular preparation for airway management

A standardized approach for airway examination and detection of airway challenges is recommended. The particular preparation for airway management should be based on the examination results. Small-diameter equipment is recommended in many patients according to the short stature. Orofacial features in EVC patients are regularly of no particular issue to airway management. However, positioning and fixation of a laryngeal mask might be challenging in some patients [15,18,19].

Particular preparation for transfusion or administration of blood products

No specific recommendations are given. No typical bleeding disorders were reported for EVC patients.

Particular preparation for anticoagulation

In patients with cardiac abnormalities and/or cardiac surgery, anticoagulation should be restarted early after the operation if anticoagulation is needed e.g., due to atrial fibrillation, in presence of mechanical valves.

Particular precautions for positioning, transportation and mobilization

Due to existing contractures in many EVC patients, careful patient positioning should be performed for surgery [19].

Interactions of chronic disease and anesthesia medications

Not reported.

Anesthetic procedure

Preoperative evaluation: see details above.

Premedication: might be performed weighing the benefits and risks in individual patients.

Prophylaxis for endocarditis: should be performed on patients with congestive heart disease according to current international guidelines and / or after discussion with the responsible cardiologist [19].

Patient positioning & monitoring: avoid overextension of limbs with contractures. Size of the blood pressure cuff should be properly selected to avoid measurement error.

IV line: placement might be difficult due to small vein calibers.

Anesthesia: Induction of anesthesia should be performed after consideration of all patient-specific risk. Using small-sized endotracheal tubes adjusted to the patient's height seems appropriate [19-21]. Ventilation should be performed carefully with adequately low tidal volumes and properly adjusted ventilator settings to reduce baro-/volutrauma in these patients with thoracic deformities and lower chest wall compliance [18]. There are no absolute or known relative contraindications for anesthesia-related drugs in patients with EVC. There is no specific risk for malignant hyperthermia.

Total intravenous or balanced anesthesia using volatile anesthetics can be performed safely.

Regional anesthesia can be performed as described above. However, single case reports describe difficulties in performing neuraxial anesthesia [22,23].

Particular or additional monitoring

Not reported.

Possible complications

In patients with cardiac abnormalities, current guidelines for prophylaxis for endocarditis should be followed [10]. No specific complications were reported regarding complications after surgery in patients with EVC syndrome. Two single-center reports described an unexpected high mortality in EVC patients after cardiac surgery [2,3,5]. However, the mortality after surgery of these defects is usually lower than 1% in the general population. Further studies are needed to evaluate this topic adequately. Respiratory complications due to severe restrictive lung function were reported in case reports [24].

Lack of larger patient populations results in uncertainty of specific recommendations to reduce postoperative morbidity or mortality in non-cardiac surgery.

Postoperative care

Postoperative care should be based upon the patients pre-existing conditions as well as the surgical or interventional procedure. Respiratory and cardiac function should be monitored in an appropriately extended stay in PACU, IMC or ICU before transfer to the normal ward or discharge at home is acceptable.

Disease-related acute problems and effect on anesthesia and recovery

Deterioration of pre-existing respiratory or cardiac impairment.

Ambulatory anesthesia

Specific recommendations for or against ambulatory anesthesia cannot be given as no published literature exists regarding this topic. Cardiac dysfunction and / or respiratory dysfunction might be a relevant factor for not performing ambulatory anesthesia in some patients.

Obstetrical anesthesia

Patients with EVC are fertile, thus an obstetrical anesthetist might meet women with EVC. In general, neuraxial as well as general anesthesia might be performed in this patient population. Severe complications were not reported. However, the lack of reports of obstetrical anesthesia should result in proper shared decision making regarding the selection of anesthesia techniques for specific women. A single case report describes a patient undergoing cesarean section after unsuccessful spinal anesthesia and performance of uncomplicated general anaesthesia [25].

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

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