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Dyke-Davidoff-Masson syndrome

Ellis-van Creveld syndrome

orphanaesthesia

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

SUPPLEMENT NR. 2 | 2022

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 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.

Find a survey of the recommendations published until now on:

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orphananesthesia

Anaesthesia recommendations for

Ellis-van Creveld syndrome

Disease name: Ellis-van Creveld syndrome

ICD 10: Q77.6

Synonyms: EVC, Chondroectodermal dysplasia, Mesodermal dysplasia

Disease summary: Ellis-van Creveld syndrome (EVC) is a rare, autosomal recessive disorder with unknown prevalence (about 150 cases were published in the last 50 years, higher prevalence in Amish communities). It is characterised by a tetrad of short stature, short limbs, postaxial polydactylism (supernumerary fingers and / or toes), ectodermal dysplasia and a high prevalence of congenital heart defects (mainly abnormalities of atrial septation) [1,2]. Mutations in the EVC, EVC2 and DYNC2LI1 genes are causative for this disease. Relevant clinical symptoms arise from the disorder of chondral and ectodermal tissues in the body. A narrow thorax due to the shortness of ribs may result in severe postnatal respiratory distress [1]. Dysplastic nails and teeth are further typical symptoms of the disorder. There is no cure for this disease and treatment is mostly symptomatic, involving treatment of respiratory distress due to the narrow chest and the combination with heart failure due to cardiac abnormalities [1]. Cardiac surgery is regularly performed in childhood for correction of the congenital heart defects. During childhood and adulthood, patients frequently undergo surgical procedures due to cardiac abnormalities [2,3], dental problems as well as orthopaedic disabilities. Life expectancy is impaired mainly due to the severity of respiratory distress as well as congenital heart disease in EVC patients [1-3,4,5]. Peri-operative medicine (e.g. surgical procedures, anaesthesia) should be performed after careful evaluation of the specific patient and her/his specific clinical features.

Medicine is 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

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Typical surgery

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) [1-3].

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) [1-3].

Dental surgery due to hypoplastic or absent teeth, orthopaedic surgery (due to bone deformities like progressive valgus deformity, profound contractures, lateral patellar subluxation) [6]. Amputation of supernumerary fingers / toes (due to medical / anthropological reasons).

Type of anaesthesia

A general recommendation regarding an ideal anaesthetic approach cannot be given, as both general anaesthesia and regional anaesthesia techniques might present potential problems in specific EVC patients. An individual approach with the surgeon as well as the patient and the competent anaesthetist will evaluate the optimal technique.

General anaesthesia might be difficult due to the respiratory and cardiac status of the patient with EVC. Thorough preoperative airway evaluation should be performed. Ellis-van-Creveld is not linked with higher risks for difficult airway status but overall data for this topic is limited.

Neuraxial regional anaesthesia techniques might be difficult due to skeletal abnormalities like lumbar lordosis or (but only in few patients) scoliosis. Therefore, there is no specific contraindication against neuraxial blockades in patients with EVC which is only based by the disease itself. 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 anaesthesia techniques should be performable in most patients. Due to deformities in the extremities, landmark-guided approach is discouraged for peripheral nerve blocks. An ultrasound examination will help to find the relevant structures for nerve blocks.

Necessary additional pre-operative 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 will usually not be helpful in pre-operative evaluation if no specific questions arise from anamnesis or clinical examination (e.g. potential bleeding disorder in the anamnesis unrelated to EVC).

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Transthoracic echocardiography is recommended in all patients (due to the high prevalence of cardiac abnormalities).

Particular preparation for airway management

A standardised approach for airway examination and detection of airway challenges is recommended. A 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. Orodental features in EVC patients are usually no particular issue regarding airway management [8,9]. However, positioning and fixation of a laryngeal mask might be challenging in some patients.

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 operation if anticoagulation is needed, e.g. due to atrial fibrillation, in presence of mechanical valves.

Particular precautions for positioning, transportation and mobilisation

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

Interactions of chronic disease and anaesthesia medications

Not reported.

Anaesthetic procedure

Preoperative evaluation: see details above.

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

Prophylaxis of endocarditis: should be performed on patients with congestive heart disease according to current international guidelines and/or after discussion with the responsible cardiologist [10].

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Patient positioning and monitoring: avoid hyperextension in limbs with contractures. Size of the blood pressure cuff should be properly selected to avoid severe measurement error.

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

Anaesthesia: Induction of anaesthesia should be performed under consideration of patient-specific risk factors unrelated to EVC. 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 [11]. There are no absolute or known relative contraindications for anaesthesia-related drugs just because of the disease EVC. There is no specific risk for malignant hyperthermia.

Total intravenous or balanced anaesthesia using volatile anaesthetics can be performed safely.

Regional anaesthesia can be performed as described above.

Particular or additional monitoring

Not reported.

Possible complications

In patients with cardiac abnormalities, current guidelines for endocarditis prophylaxis should be followed [10]. No specific complications were reported regarding complications after surgery in patients with EVC syndrome. However, two single-centre 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 a general population. Further studies are needed to evaluate this topic adequately.

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

Post-operative care

Post-operative care should be based upon the patient's 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 to home is acceptable.

Disease-related acute problems and effect on anaesthesia and recovery

Deterioration of pre-existing respiratory or cardiac impairment.

Ambulatory anaesthesia

Specific recommendations for or against ambulatory anaesthesia 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 anaesthesia in some patients.

Obstetrical anaesthesia

Patients with EVC are fertile, thus the obstetrical anaesthetist might face women with EVC for labour analgesia. In general, neuraxial as well as general anaesthesia might be performed in this patient population. Severe complications were not reported. However, the lack of reports of obstetrical anaesthesia should result in proper shared decision-making regarding the selection of anaesthesia techniques for specific women. A single case report describes a patient undergoing caesarean section after unsuccessful spinal anaesthesia and performance of an uncomplicated general anaesthesia [11].

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References

1. Baujat G, Le Merrer M. Ellis-van Creveld syndrome. Orphanet J Rare Dis 2007;2:27
2. O'Connor MJ, Collins RT. Ellis-van Creveld syndrome and congenital heart defects: presentation of an additional 32 cases. Pediatr Cardiol 2012;33:491; discussion 491–492
3. Hills CB, Kochilas L, Schimmenti LA, Moller JH. Ellis-van Creveld syndrome and congenital heart defects: presentation of an additional 32 cases. Pediatr Cardiol 2011;32:977–982
4. Pérez-Andreu J, Ray VG, Arribas JM, Sánchez SJ. Ellis-van Creveld syndrome in adulthood: extending the clinical spectrum. Singapore Med J 2015;56:e110-111
5. O'Connor MJ, Rider NL, Thomas Collins R, Hanna BD, Holmes Morton D, Strauss KA. Contemporary management of congenital malformations of the heart in infants with Ellis-van Creveld syndrome: a report of nine cases. Cardiol Young 2011;21:145–152
6. Weiner DS, Jonah D, Leighley B, Dicintio MS, Holmes Morton D, Kopits S. Orthopaedic manifestations of chondroectodermal dysplasia: the Ellis-van Creveld syndrome. J Child Orthop 2013;7:465–476
7. Hopman G, Waaijer A, Van Tuijl I. A rare problem with an epidural catheter in a patient with Ellis-van Creveld syndrome. Paediatr Anaesth 2009;19:812–813
8. Tuna EB, Koruyucu M, Kürklü E, Çifter M, Gençay K, Seymen F, et al. Oral and craniofacial manifestations of Ellis-van Creveld syndrome: Case series. J Craniomaxillofac Surg 2016; 44:919–924
9. Shaik S, Raviraj J, Dirasantchu S, Venkata SS. Ellis-van Creveld syndrome with unusual oral and dental findings: A rare clinical entity. Dent Res J (Isfahan) 2016;13:193–197
10. Habib G, Lancellotti P, Antunes MJ, Bongiorni MG, Casalta J-P, Del Zotti F, et al. 2015 ESC Guidelines for the management of infective endocarditis: The Task Force for the Management of Infective Endocarditis of the European Society of Cardiology (ESC). Endorsed by: European Association for Cardio-Thoracic Surgery (EACTS), the European Association of Nuclear Medicine (EANM). Eur. Heart J 2015;3075–3128
11. Fernández-Meré LA, Alvarez-Blanco M, Jorge-García J, Martínez-Suárez MA. [Anesthesia in a patient with Ellis-van Creveld syndrome]. Rev Esp Anestesiol Reanim 2010;57:528–531.

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

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