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3MC syndrome/Michels syndrome

Xeroderma pigmentosum

orphan**anesthesia**

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

SUPPLEMENT NR. 1 | 2020

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 OrphanAnesthesia 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.

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 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 Handlungsempfehlungen finden Sie unter:

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

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

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Projektleitung

Prof. Dr. Tino Münster, MHBA

Chefarzt
Klinik für Anästhesie und
operative Intensivmedizin
Krankenhaus Barmherzige
Brüder Regensburg
Prüfeninger Straße 86
93049 Regensburg,
Deutschland

Tel.: 0941 369-2350

E-Mail: Tino.Muenster@barmherzige-regensburg.de

orphananesthesia

Anaesthesia recommendations for Xeroderma pigmentosum

Disease name: Xeroderma pigmentosum (XP)

ICD 10: Q82.1

Synonyms: Kaposi disease, Ichthyosis; individuals suffering from this disease are often referred to as children of the night or moon people

Disease summary: Xeroderma pigmentosum (XP) is a rare autosomal recessive disorder due to the defect in the nucleotide repair genes resulting in inability to repair the damaged DNA caused by ultraviolet (UV) rays. Hence people suffering from this disease show extreme sensitivity to sunlight and UV radiation and are characterised by skin lesions mostly in the sun-exposed areas such as head, face and neck. It affects 1 in 250,000 people and has 1000-fold increased risk of developing cancers in sun-exposed areas. Even the incidence of internal malignancy is 10-20 times higher than in normal individuals. The disease may also be associated with progressive neurological degeneration. Most people require multiple surgeries for removal of the skin, ocular lesions and malignancies. The main anaesthetic concerns are difficult intravenous cannulations, difficult airway (bag-mask ventilation and intubation), genotoxic and progressive neurological deterioration of the patients with the use of volatile anaesthetic agents, increased sensitivity to opioids, benzodiazepines and muscle relaxants, and difficult extubation due to epiglottis subsidence.

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

Patients with XP undergo excisions of squamous cell carcinoma, basal cell carcinoma on the UV exposed areas (face, head, neck) along with skin grafts and skin flaps.

They also undergo eyeball enucleation, ectropion correction, eye tumour removal, and cataract surgery.

Type of anaesthesia

The lesions are usually present on the head, the face and the upper part of the body, so most of the patients require general anaesthesia.

Volatile agents should be avoided as these agents have been shown to facilitate the progression of the disease. Halothane has genotoxic effects and isoflurane, sevoflurane are associated with worsening of the neurological symptoms. So total intravenous anaesthesia should be the preferred anaesthetic technique.

Patients are sensitive to benzodiazepines and opioids. Thus premedication with benzodiazepines are better avoided and, if necessary, such drugs should be administered under monitoring.

Patients are more sensitive to muscle relaxants. So muscle relaxants should be totally avoided, if possible. If not, then at least a smaller dose of a shorter-acting muscle relaxant with strict neuromuscular blockade monitoring is recommended.

Whenever possible, if the surgical site permits, regional anaesthesia should be preferred over general anaesthesia.

Necessary additional pre-operative testing (beside standard care)

Genetic screening can be done to determine the type of XP (total 7 types, XPA to XPG) and also to identify the neurodegenerative type (concerns all types except for XPC and XPE).

Neurological assessment as well as CT/MRI of the brain in patients with neurological symptoms to rule out any causes other than XP.

Sensorineural hearing deficiency tests in patients with hearing loss.

Particular preparation for airway management

There are cases in which patients suffering from this disease are reported to have difficult airways. Multiple lesions on the face and facial disfiguring can cause difficult face mask ventilation, thus multiple sizes and types of face masks should be prepared accordingly.

In addition, there are reports of difficult intubations requiring stylet and bougie during intubation. Therefore, a difficult airway cart should be available during airway management. Awake fiberoptic intubation can be the choice of airway management in patients with an anticipated difficult airway.

We also advise to apply a gauze piece soaked with paraffin in case of ulcerative lesions to prevent bleeding and skin peeling resulting from the direct pressure of the mask.

We also suggest the awake extubation of patients with epiglottic dehiscence, as these patients are prone to post-operative stridor.

Particular preparation for transfusion or administration of blood products

No recommendations found.

Particular preparation for anticoagulation

No recommendations found.

Particular precautions for positioning, transportation and mobilisation

Disease progresses when the skin is exposed to UV rays. The patient should therefore wear protective clothing, UV-shielding films and sunscreens having high sun protective factor (SPF) in the operation theatre.

Covering all the parts of body with proper drapes before turning the operating-room lights on and during whole of the intraoperative period is advised. Avoidance of halogenated lights should be done as much as possible.

Bones may be fragile in these patients due to avoidance of sunlight leading to vitamin D deficiency, so proper padding of pressure points and gentle handling during transport is a must.

Use of eye ointments and proper closure of the eyes must be ensured in patients with conjunctival and corneal lesions.

Interactions of chronic disease and anaesthesia medications

The patients may be taking chemotherapeutic agents like 5-fluorouracil (5-FU) which causes myelosuppression. Nitrous oxide should therefore be avoided in such patients.

Patients with XP may be on steroid therapy, so steroid supplementation in the intraoperative and steroid maintenance in the postoperative period is advised.

Anaesthetic procedure

Due to multiple lesions on the skin, intravenous cannulation and fixation can be difficult.

For general anaesthesia, total intravenous anaesthesia (TIVA) is preferred. Halothane, isoflurane and sevoflurane have been shown to have genotoxic effects and also have caused

worsening of neurological symptoms. Therefore, these agents are better avoided. Propofol, ketamine or dexmedetomidine in combination with short-acting opioids are the preferred anaesthetic agents for TIVA.

Nitrous oxide should be avoided in patients taking 5-fluorouracil as both the drugs cause myelosuppression.

Since these patients are sensitive to the paralysing effects of muscle relaxants due to neuronal dysfunction and muscle atrophy, total avoidance or minimal use of shorter-acting agents with strict neuromuscular-block monitoring is advised. Intubation without the use of muscle relaxants can be facilitated by applying other anaesthetic adjuncts like dexmedetomidine.

In addition, dexmedetomidine also reduces the induction and maintenance dose of propofol, blunts the airway reflex during intubation and extubation, decreases the requirement of opioids and also helps to enhance the recovery. Ketamine can also be a good adjunct of TIVA.

Another feature is that these patients are very sensitive to opioids. Hence short-acting opioids such as fentanyl can be used for the intraoperative period. Multimodal analgesia with ketamine, paracetamol, ketorolac, dexmedetomidine and the infiltration of local anaesthetics decreases the requirement of opioids. The concomitant use of benzodiazepines and opioids has led to apnoea and desaturation indicating that these patients are very sensitive to this combination.

Eye ointment use in intraoperative periods prevents the drying of cornea and thus corneal ulcerations.

There are no contraindications for regional anaesthesia provided to the site of the surgical field and patient's choice.

Particular or additional monitoring

Standard ASA I and II monitoring is required for all patients and includes temperature, heart rate, ECG, blood pressure, peripheral oxygen saturation concentration and end-tidal carbon dioxide concentration.

A peripheral nerve stimulator to assess the neuromuscular blockade is recommended when muscle relaxants are used.

Bispectral index (BIS) monitoring is beneficial to ensure the depth of anaesthesia wherever applicable.

Possible complications

Progression of the disease and worsening of the neurological symptoms in the immediate and late post-operative period occur in patients especially when volatile agents such as halothane, isoflurane, sevoflurane are used.

Delayed recovery, agitation, abnormal movements and confusion are the immediate manifestations, whereas memory disorders, false recognition and reversible cognitive decline have been reported as late complications after the use of volatile agents.

Post-operative care

Avoid the exposure of the skin to UV lights in the exposed parts.

Monitor for the smooth recovery as drugs effects are more pronounced in such patients.

Disease-related acute problems and effect on anaesthesia and recovery

No recommendations found.

Ambulatory anaesthesia

No recommendations found.

Obstetrical anaesthesia

No recommendations found.

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This recommendation was prepared by:

Authors

Bashu Dev Parajuli, Lecturer, Department of Anaesthesiology, Tribhuvan University Teaching Hospital (TUTH,) Maharajgunj Medical Campus (MMC), Institute of Medicine(IOM), Maharajgunj, Kathmandu, Nepal
bashuparajuli2012@gmail.com

Megha Koirala, Lecturer, Department of Anaesthesiology, Tribhuvan University Teaching Hospital (TUTH,) Maharajgunj Medical Campus (MMC), Institute of Medicine(IOM), Maharajgunj, Kathmandu, Nepal

Basanta Ghimire, Registrar, Anaesthesiologist, Nepal Medcity Hospital, Nakhu, Kathmandu, Nepal

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This recommendation was reviewed by:

Reviewer 1

Rakhee Goyal, Anaesthesiologist, Department of Anaesthesia and Critical Care, Army Hospital (Research and Referral), New Delhi, India
rakheegoyalkumar@gmail.com

Reviewer 2

Carlos R Degrandi Oliveira, Anaesthesiologist, MD, Santos, Brazil
degrandi@gmail.com

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Please note that this recommendation has been reviewed not by an anaesthesiologist and some other disease expert but by two anaesthesiologists instead.

Herausgeber



DGAI

Deutsche Gesellschaft
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Carolin Sofia Kopp B.A.
Korrespondenzadresse: Roritzerstraße 27 |
90419 Nürnberg | Deutschland
Tel.: 0911 9337812 | Fax: 0911 3938195
E-Mail: anaesth.intensivmed@dgai-ev.de

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Nadja Schwarz
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E-Mail: info@aktiv-druck.de

Anzeigen | Vertrieb

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

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

Herstellung | Gestaltung

Pia Engelhardt | Stefanie Triebert
Tel.: 09522 943570 | Fax: 09522 943577
E-Mail: ai@aktiv-druck.de

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ADDRESS

German Society of Anaesthesiology and
Intensive Care Medicine
Ursula Homberg
Roritzerstrasse 27 | 90419 Nuremberg | Germany
Tel.: +49-911-9337828 | Fax: +49-911-3938195
Email: uhomberg@orphananesthesia.eu