

A&I

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Xeroderma pigmentosum

orphan**anesthesia**

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

SUPPLEMENT NR. 16 | 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
Handlungsempfehlungen finden
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orphan**a**nesthesia

Anesthesia 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 disease caused an inability to repair DNA pyrimidine dimers caused by ultraviolet (UV) exposure. It was first described in 1874 by Hebra and Kaposi, clinically manifesting as hypersensitivity to sunlight, freckles, and skin cancer, with neurological manifestations described later in 1883. The incidence is highest in Japan at 45 per million, 1 per million in the United States, and 2.3 per million in Western Europe. Both sexes are affected equally by XP [1].

According to Lehmann, there are 8 subtypes of XP:

XP type: XP-A

Gene/locus: XPA / 9q22.3

Clinical presentation: 25% of the patients with XP. Most serious form of the disease. Patients present a diverse range of symptoms, including the development of numerous skin cancers at an early age and serious neurological abnormalities.

XP type: XP-B

Gene/locus: XPB (ERCC3) / 2q21

Clinical presentation: Least frequent of the XP types. Increased sensitivity to UV light. Patients may develop numerous skin cancers at an early age, some mild neurological abnormalities and they can present characteristics of Cockayne syndrome.

XP type: XP-C

Gene/locus: XPC / 3p25

Clinical presentation: 25% of the patients with XP the most frequent form in the Caucasian population. Considered as the classical form of XP. It is the variant with the highest capability to repair DNA even it shows increased sensitivity to UV exposure. Patients may develop severely atypical and dense lentigines at exposed areas as well as ocular anomalies but no neurological abnormalities.

XP type: XP-D

Gene/locus: XPD (ERCC2) / 19q13.2-q13.3

Clinical presentation: 15% of the patients with XP. It presents with increased sensitivity to UV light. Patients may develop numerous skin cancers at an early age. They may present in

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some cases severe neurological defects with progressive degeneration such as sensorineural deafness, ataxia and mental retardation.

XP type: XP-E

Gene/locus: XPE (DDB2) / 11p11-p12; 11q12-q13

Clinical presentation: Low frequency. The least aggressive form of XP. It is limited to skin problems. Patients present the development of cutaneous cancers at a later age, and they do not present neurological symptoms.

XP type: XP-F

Gene/locus: XPF (ERCC4) / 16p13.3

Clinical presentation: Low frequency. A less aggressive form of XP, most of the cases in the Japanese population. Development of cutaneous cancers at a later age with no neurological nor ocular symptoms.

XP type: XP-G

Gene/locus: XPG (ERCC5) / 13q32-q33

Clinical presentation: Low frequency but aggressive with increased sensitivity to UV light. Development of numerous skin cancers at an early age and neurological abnormalities in some cases.

XP type: XP-variant

Gene/locus: XPG (ERCC5) / 13q32-q33

Clinical presentation: Low frequency but aggressive with increased sensitivity to UV light. Development of numerous skin cancers at an early age and neurological abnormalities in some cases.

Most patients (especially the A, B, G and G subtypes) present within the first few years of life. People suffering from XP show extreme sensitivity to UV radiation with skin lesions mostly in the sun exposed areas such as head, face, and neck. Cutaneous symptoms include excessive freckling, hyper- and hypopigmentation, poikiloderma, skin atrophy and skin aging. Ocular manifestations include photophobia, keratitis, and ectropion [3]. Cutaneous and ocular neoplasia also occur at a very high rate. The median age for first non-melanoma skin cancer is nine, the median age for development of first malignant melanoma is 22 [4]. Most patients require multiple surgeries for removal of skin and ocular lesions, and malignancies. About 20-30% of the patients may also have progressive neurologic degeneration characterized by premature neuronal death, progressive mental retardation, hyper- or hyporeflexia, hearing loss, motor deficits, cognitive dysfunction, and cerebellar dysfunction [3,5,6].

Major anesthetic concerns include difficult intravenous cannulations, difficult bag-mask ventilation and intubation, neurological deterioration with volatile agents, increased sensitivity to opioids, benzodiazepines, and muscle relaxants, and difficult extubation.

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	Anticipate difficult bag mask ventilation as well as intubation, awake/asleep fiberoptic intubation depending on patient cooperation, supraglottic airway can be safe – opt for neuraxial or peripheral regional anesthesia whenever possible. Peripheral venous access may be difficult. OR lights should always be dimmed or have a protective film.
B	BLOOD PRODUCTS (COAGULATION)	May be anemic, arrange blood products according to surgery.
C	CIRCULATION	Standard monitoring is usually sufficient.
D	DRUGS	No added risk of MH – avoid volatile anesthetics as they can worsen neurological symptoms – avoid nitrous oxide as it can worsen myelosuppression – increased sensitivity to benzodiazepines, opioids, and muscle relaxants – TIVA with propofol, ketamine, dexmedetomidine is safe.
E	EQUIPMENT	Ultrasound for venous cannulation – fiberoptic bronchoscope, videolaryngoscope, McCoy blade for airway management – train of four monitoring. Protect skin and eyes from UV (ideally no exposure $>10\mu\text{W}/\text{cm}^2$. If an excessive source of UV does need to be used, place a special protective film (Dermagard®).

Typical surgery and procedures

Patients with XP can undergo multiple excisions of squamous cell carcinoma, basal cell carcinoma or melanoma of the UV exposed areas along with skin graft and flaps.

Ocular surgeries like enucleation, ectropion correction, removal of ophthalmic tumors, and cataract surgeries are also common.

Type of anesthesia

Lesions present in the head, neck, or trunk require general anesthesia. Volatile agents have been shown to facilitate disease progression, and hence, are usually avoided. Volatile anesthetics could have genotoxic properties [7]. While surgeries have been done under sevoflurane anesthesia without neurological complications in patients with normal preoperative neurological evaluation, there have been reports that sevoflurane could increase progression of neurological symptoms [8–10]. TIVA could be the preferred anesthetic technique when general anesthesia is required. Nitrous oxide is safe if the patient is not under medications that can cause myelosuppression [11–14].

Regional anesthesia should be the preferred choice when the necessary surgery allows it as it avoids risks related to anesthetic agents and difficult airway.

Premedication with benzodiazepine is avoided when possible, owing to their increased sensitivity.[15] If at all necessary, they should be administered under continuous monitoring. Similarly, opioid sensitivity is another concern.[15] Small doses of short acting opioids should be used when necessary. Opioid sparing analgesia regimen using regional analgesia techniques and multimodal analgesia can be helpful.

XP patients have increased sensitivity to neuromuscular blocking agents [8,16]. They should be avoided whenever possible. If they are necessary, monitored doses of short acting neuromuscular blocking agents should be used with proper neuromuscular monitoring.

Necessary additional preoperative testing (beside standard care)

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

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

Sensorineural hearing deficiency tests in patients having hearing loss.

Particular preparation for airway management

There are cases reported to have difficult airway in patients suffering from this disease [8,10,14,15,17–19]. Multiple lesions on the face and facial disfiguring can cause difficult face mask ventilation, thus multiple size and types of face masks should be prepared accordingly.

Also, there are reports of difficult intubation requiring stylet and bougie during intubation. So difficult airway cart should be available during airway management. Awake fiberoptic intubation can be the choice of airway management in patients with anticipated difficult airway provided the patient is cooperative enough.

We also advise keeping gauze pieces soaked with paraffin or petrolatum in cases of ulcerative lesions to prevent bleeding and peeling of skin from direct pressure of the mask.

We also suggest awake extubation in patients with epiglottic dehiscence, which are reported to cause postoperative stridor.

Particular preparation for transfusion or administration of blood products

Patients with XP can have anemia due to chronic inflammation, superinfection of ulcerative tumors as well as malnutrition. Proper nutrition for elective procedures can help decrease blood transfusion. Arrangement of adequate blood products is necessary depending upon the surgical procedure.

Some patients may also have Fanconi anemia [20]. These patients may require platelet transfusion in addition to red blood cells.

Particular preparation for anticoagulation

No recommendations found.

Particular precautions for positioning, transportation and mobilization

Disease progresses when the skin is exposed to the UV rays. So, patients should wear protective clothing, UV shielding films and sunscreens having high sun protective factors (SPF > 50%) in the operation room.

All potential sources of UV should be identified with a handheld UV detector and a maximal exposure of $10\mu\text{W}/\text{cm}^2$ is tolerated. If an excessive source of UV does need to be used, place a special protective film (e.g., Dermagard®) or UV lens filter for a camera between the source and the film. The lights of the OR and corridors should be dimmed, and the patient's body should be covered with drapes.

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. Halogenated lights should be avoided as far as possible [21,22].

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. Patients with XP can also have peripheral neuropathy and be at a higher risk of position related nerve injury. Preoperative documentation and perioperative precaution is essential.

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

Interactions of chronic disease and anesthesia medications

The patients may be taking chemotherapeutic agents like 5-Fluorouracil (5-FU) which causes myelosuppression. High doses of nitrous oxide have been shown to cause myelosuppression as well. As these patients usually require multiple surgeries, refraining from use of nitrous oxide may be prudent [23].

Anesthetic procedure

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

For general anesthesia, total intravenous anesthesia (TIVA) is preferred. Halothane, isoflurane, and sevoflurane have shown to have genotoxic effect and have caused worsening of neurological symptoms. So, these agents are best avoided. Propofol, ketamine or dexmedetomidine with short acting opioids are the preferred anesthetic 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 neuromuscular blocking agents due to neuronal dysfunction and muscle atrophy, total avoidance or minimal use of shorter acting agents like mivacurium with strict neuromuscular block monitoring is advised. Intubation without the use of muscle relaxants, can be facilitated using other anesthetic adjuncts like dexmedetomidine.

Besides, 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.

Airway management is usually challenging. Equipment for difficult airway management and front of neck access should be ready in all cases. Bag mask ventilation itself can be difficult owing facial deformity and multiple ulcerations. Endotracheal intubation can also be difficult. Awake fiberoptic intubation can be the safest option if the patient is cooperative.

Also, these patients are very sensitive to opioids. So short acting opioids such as remifentanyl can be used for the intraoperative period. Use of multimodal analgesia with ketamine, paracetamol, ketorolac, dexmedetomidine, infiltration of local anesthetics, and use of regional analgesia decreases the requirement of opioids. Concomitant use of benzodiazepines and opioids has led to apnea and desaturation indicating that these patients are very sensitive to this combination.[15]

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

There is no contraindications for regional anesthesia provided by 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 which include temperature, heart rate, ECG, blood pressure, peripheral oxygen saturation concentration and end-tidal carbon dioxide concentration.

Train of four monitoring to assess the neuromuscular block is recommended to be used when neuromuscular blocking agents are used.

Bispectral Index (BIS) monitoring is beneficial to ensure the depth of anesthesia wherever applicable, especially with TIVA.

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, confusion are the immediate manifestations whereas memory disorders, false recognition and reversible cognitive decline have been reported as the late complications after the use of volatile agents.[9]

Prolonged effects of opioids, benzodiazepines, and muscle relaxants can cause postoperative respiratory complications.

Postoperative care

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

Monitor for the smooth recovery as drugs effect are more pronounced in such patient.

Disease-related acute problems and effect on anesthesia and recovery

No recommendations found.

Ambulatory anesthesia

No recommendations found.

Obstetrical anesthesia

No recommendations found.

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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.

Update and revision (2025)

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