Aktiv Druck & Verlag GmbH | ISSN 0170 - 5334 I 02330

www.ai-online.info

61. Jahrgang | September 2020

A&I

ANÄSTHESIOLOGIE & INTENSIVMEDIZIN

Offizielles Organ: Deutsche Gesellschaft für Anästhesiologie und Intensivmedizin e.V. (DGAI)
Berufsverband Deutscher Anästhesisten e.V. (BDA)
Deutsche Akademie für Anästhesiologische Fortbildung e.V. (DAAF)
Organ: Deutsche Interdisziplinäre Vereinigung für Intensiv- und Notfallmedizin e.V. (DIVI)

Infantile neuroaxonal dystrophy

Insulinoma

orphanaesthesia

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

SUPPLEMENT NR. 19 | 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 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:

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

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

DGAI
Deutsche Gesellschaft für Anästhesiologie & Intensivmedizin
www.dgai.de

A&I
ANÄSTHESIOLOGIE & INTENSIVMEDIZIN
www.ai-online.info

www.orphananesthesia.eu

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

orphananesthesia

Anaesthesia recommendations for

Infantile neuroaxonal dystrophy

Disease name: Infantile neuroaxonal dystrophy

ICD 10: G23.0

Synonyms: INAD, NBIA2, Phospholipase A2-associated neurodegeneration (PLAN), Seitelberger Disease, Neurodegeneration with brain iron accumulation A

Disease summary: Infantile neuroaxonal dystrophy (INAD) is a neurodegenerative disorder associated with a mutation in the PLA2G6 gene. It is the second most common type of neurodegeneration with brain iron accumulation (NBIA) mainly in the globus pallidus, caudate nucleus and substantia nigra after pantothenate kinase-associated neurodegeneration (PKAN, formerly Hallervorden-Spatz disease). INAD is inherited in an autosomal recessive fashion. PLA2G6 encodes a calcium-independent phospholipase and has been associated with infantile neuroaxonal dystrophy (INAD), atypical neuroaxonal dystrophy (NAD) and dystonia-parkinsonism. PLA2G6 is protective of mitochondrial health. It is also important for membrane homoeostasis and calcium signalling. INAD is characterised histologically by axonal spheroid bodies. Phenotypically, INAD is characterised by psychomotor regression with early onset between six months and three years of age. Hypotonia occurs early with severe weakness that may be replaced by spasticity. Many INAD patients also experience progressive dementia. Death usually occurs before the age of 10 years due to respiratory complications. Patients may present for gastrostomy tube and tracheostomy placement due to bulbar dysfunction and some may require surgical correction of scoliosis to improve respiratory status. The main anaesthetic concern for INAD patients is their poor preoperative respiratory status due to poor airway clearance and respiratory mechanics that often make postoperative intubation necessary.

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

▶ **Citation:** Sinskey J, Holzman RS: Infantile neuroaxonal dystrophy. Anästh Intensivmed 2020;61:S343–S348. DOI: 10.19224/ai2020.S343

Typical surgery

Gastrostomy, tracheostomy, scoliosis repair. Additional reported procedures include oral rehabilitation, deep brain stimulation, debridements and pallidothalamotomy.

Type of anaesthesia

INAD patients can safely undergo general anaesthesia. Although many patients are not cooperative, preoperative sedation should be approached cautiously, if at all, because of limited respiratory reserves and increased susceptibility to respiratory depression.

Unlike patients with muscular dystrophy, these patients can safely receive inhaled anaesthetic agents without risk of rhabdomyolysis. Most INAD patients are immobilised and succinylcholine should be avoided due to the risk of hyperkalaemia and cardiac arrest.

Necessary additional pre-operative testing (beside standard care)

Patients with INAD have progressive weakness that can lead to cardiopulmonary compromise.

Preoperative electrocardiogram and echocardiogram should be obtained as patients with advanced disease may develop pulmonary hypertension and cardiac dysfunction.

The pulmonary status can be assessed with a chest radiograph and arterial blood gas analysis. A pulmonary function test may be helpful if the patient can cooperate.

Particular preparation for airway management

Patients may be difficult to intubate due to joint changes or muscle contractures that can limit cervical spine mobility and mouth opening even with muscle relaxation. Trismus and torticollis have been reported in patients with PKAN and spinal muscular atrophy (SMA) although muscular relaxation has been reported with the induction of anaesthesia as well as the administration of dexmedetomidine in PKAN.

Patients are at increased risk of aspiration due to bulbar dysfunction. Some patients may already have a tracheostomy in place at the time of surgery.

Particular preparation for transfusion or administration of blood products

Patients may require blood transfusions with more invasive procedures. Patients with neuromuscular scoliosis presenting for scoliosis repair have been shown to have higher blood loss compared to patients with idiopathic scoliosis.

© Anästh Intensivmed 2020;61:S343–S348 Aktiv Druck & Verlag GmbH

Particular preparation for anticoagulation

Not reported.

Particular precautions for positioning, transportation and mobilisation

Patients with advanced disease may develop spastic tetraplegia and optic atrophy. Extra care should be taken during positioning of the patient for surgery.

Interactions of chronic disease and anaesthesia medications

Not reported. Patients may be on medications to treat spasticity and seizures. Patients treated with dopaminergic blockers such as phenothiazines, butyrophenones and metoclopramide may experience exacerbation of their movement disorders. Patients taking anticonvulsants may have alterations in their biotransformation of medications used during anaesthesia.

Anaesthetic procedure

Avoid succinylcholine due to the risk of hyperkalaemic cardiac arrest. Inhaled anaesthetics, propofol and opiates may be safely used. Rapid sequence induction with non-depolarising neuromuscular blocking agents can be considered, remembering that a priming dose or a larger dose of relaxant will enhance the onset of neuromuscular blockade, but may result in residual blockade and a prolonged duration of effect.

Patients may be more sensitive to non-depolarising neuromuscular blocking agents. Consider intubation without neuromuscular blockade, given that patients often have profound weakness.

Particular or additional monitoring

If a neuromuscular blocking agent is used, baseline values should be obtained with a peripheral nerve stimulator prior to administration.

Patients undergoing high risk-surgery or those with moderate-severe cardiorespiratory dysfunction would benefit from the placement of an arterial catheter for close haemodynamic monitoring and frequent arterial blood gas analysis. Clinically acceptable normothermia should be ensured.

Possible complications

Patients may have a hyperkalaemic cardiac arrest after administration of succinylcholine.

Many patients have pre-existing respiratory dysfunction and may require postoperative intubation.

© Anästh Intensivmed 2020;61:S343–S348 Aktiv Druck & Verlag GmbH

Post-operative care

Patients should be closely monitored postoperatively; this will typically require an intensive care unit admission in many, if not most, circumstances. Many patients will require post-operative intubation and mechanical ventilatory support. Even if patients are extubated in the operating room, they have a high risk of respiratory failure following extubation. If patients require postoperative mechanical support, they may be extubated to a non-invasive ventilation modality (e.g., bi-level positive airway pressure, BIPAP). Non-invasive positive pressure ventilation can help prevent hypoventilation and atelectasis and decrease the work of breathing while chest physiotherapy can improve secretion clearance.

Disease-related acute problems and effect on anaesthesia and recovery

Patients may have severe weakness which may be confused with residual neuromuscular blockade if a non-depolarising neuromuscular blocking agent has been administered.

Ambulatory anaesthesia

INAD patients are not candidates for ambulatory surgery due to the severity of the disease.

Obstetrical anaesthesia

Patients have an extremely short life expectancy (less than 10 years of age) and are unlikely to reach childbearing age.

© Anästh Intensivmed 2020;61:S343–S348 Aktiv Druck & Verlag GmbH

References

1. Schneider SA. Neurodegeneration with Brain Iron Accumulation. Curr Neurol Neurosci Rep 2016;16:9
2. Morgan NV, et al. PLA2G6, encoding a phospholipase A2, is mutated in neurodegenerative disorders with high brain iron. Nat Genet 2006;38:752–754
3. Gregory A, Kurian MA, Maher ER, et al. PLA2G6-Associated Neurodegeneration. In: Pagon RA, Adam MP, Ardinger HH, et al. (eds.): GeneReviews®. Seattle (WA): University of Washington, Seattle 1993-2017; 2008 Jun 19. https://www-ncbi-nlm-nih-gov.ucsf.idm.oclc.org/books/NBK1675/ (Accessed on: Jan 20, 2020).
4. Sinskey JL, Holzman RS. Perioperative considerations in infantile neuroaxonal dystrophy. Paediatr Anaesth 2017;27:322–324
5. Hinkelbein J, Kalenka A, Alb M. Anesthesia for patients with pantothenate-kinase-associated neurodegeneration (Hallervorden-Spatz disease) – a literature review. Acta Neuropsychiatr 2006;18:168–172
6. Roy RC, McLain S, Wise A, Shaffner LD. Anesthetic management of a patient with Hallervorden-Spatz disease. Anesthesiology 1983;58:382–384
7. Keegan MT, Flick RP, Matsumoto JY, Davis DH, Lanier WL. Anesthetic management for two-stage computer-assisted, stereotactic thalamotomy in a child with Hallervorden-Spatz disease. J Neurosurg Anesthesiol 2000;12:107–111
8. Justesen CR, Penn RD, Kroin JS, Egel RT. Stereotactic pallidotomy in a child with Hallervorden-Spatz disease. J Neurosurg 1999;90:551–554
9. Islander, G. Anesthesia and spinal muscle atrophy. Paediatr Anaesth 2013;23:804–816
10. Shapiro F, Sethna N. Blood loss in pediatric spine surgery. Eur Spine J 2004;13,1:S6–S17
11. Hogarth P. Neurodegeneration with Brain Iron Accumulation: Diagnosis and Management. J Mov Disord 2015;8:1–13
12. Madhusudhana Rao B, Radhakrishnan M. Dexmedetomidine for a patient with Hallervorden-Spatz syndrome during magnetic resonance imaging: a case report. J Anesth 2013;27:963–964
13. Mata-Pérez K, Teja-Ángeles E, Durán-Gutiérrez LA. Oral rehabilitation under general anesthesia in a boy with Hallervorden-Spatz syndrome. Report of a case. Acta Pediatr Mex 2012;33:227–231
14. Koc D, Imer P, Byri Y, Seker A. Anesthetic management for deep brain stimulation in a patient with pantothenate kinase-associated neurodegeneration Pediatr Anesth Crit Care J 2014; 2:122–125
15. Sinha R, Biyani G, Bhattacharjee S. Anaesthetic management of a child with panthothenate kinase-associated neurodegeneration. Indian J Anaesth 2015;59:43–46
16. Keegan MT, Flick RP, Matsumoto JY, Davis DH, Lanier WL. Anesthetic management for two-stage computer-assisted, stereotactic thalamotomy in a child with Hallervorden-Spatz Disease. J Neurosurg Anesthesiol 2000;12:107–111
17. Balas I, Kovacs N, Hollody K. Staged bilateral stereotactic pallidothalamotomy for life-threatening dystonia in a child with Hallervorden-Spatz disease. Mov Disord 2006;21:82–85.

© Anästh Intensivmed 2020;61:S343–S348 Aktiv Druck & Verlag GmbH

Date last modified: **January 2020**

This recommendation was prepared by:

Authors

Jina Sinskey, Anaesthesiologist, Department of Anesthesia and Perioperative Care, University of California San Francisco, San Francisco, CA, USA
jina.sinskey@ucsf.edu

Robert S. Holzman, Anaesthesiologist, Department of Perioperative Anesthesia, Children's Hospital Boston, USA

Disclosure The authors have no financial or other competing interest to disclose. This recommendation was unfunded.

This recommendation was reviewed by:

Reviewers

Victor Baum, Anaesthesiologist, George Washington University School of Medicine, Washington, USA
victorbaum1@comcast.net

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

Disclosure The reviewers have no financial or other competing interest to disclose.

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

© Anästh Intensivmed 2020;61:S343–S348 Aktiv Druck & Verlag GmbH

Herausgeber

DGAI
Deutsche Gesellschaft für Anästhesiologie und Intensivmedizin e.V.
Präsident: Prof. Dr. R. Rossaint, Aachen

BDA
Berufsverband Deutscher Anästhesisten e.V.
Präsident: Prof. Dr. G. Geldner, Ludwigsburg

DAAF
Deutsche Akademie für Anästhesiologische Fortbildung e.V.
Präsident: Prof. Dr. H. Bürkle, Freiburg

Schriftleitung
Präsident/in der Herausgeberverbände
Gesamtschriftleiter/Editor-in-Chief:
Prof. Dr. Dr. Kai Zacharowski, Frankfurt
Stellvertretender Gesamtschriftleiter/Deputy Editor:
Prof. Dr. T. Volk, Homburg/Saar
CME-Schriftleiter/CME-Editor:
Prof. Dr. W. Zink, Ludwigshafen

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

Verlag & Druckerei

Aktiv Druck & Verlag GmbH
An der Lohwiese 36 |
97500 Ebelsbach | Deutschland
www.aktiv-druck.de

Geschäftsführung
Wolfgang Schröder | Jan Schröder | Nadja Schwarz
Tel.: 09522 943560 | Fax: 09522 943567
E-Mail: info@aktiv-druck.de

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

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

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

Titelbild
Gestaltung: Klaus Steigner
Paumgartnerstraße 28 | 90429 Nürnberg
E-Mail: mazyblue@klaus-steigner.de
www.klaus-steigner.de

Erscheinungsweise 2020
Der 61. Jahrgang erscheint jeweils zum Monatsanfang, Heft 7/8 als Doppelausgabe.

Bezugspreise (inkl. Versandkosten):

- **Einzelhefte** 30,- €
- **Jahresabonnement:**

Europa (ohne Schweiz) (inkl. 7 % MwSt.)	258,- €
Schweiz	266,- €
Rest der Welt	241,- €

Mitarbeiter aus Pflege, Labor, Studenten und Auszubildende (bei Vorlage eines entsprechenden Nachweises)

Europa (ohne Schweiz) (inkl. 7 % MwSt.)	94,- €
Schweiz	90,- €
Rest der Welt	94,- €

Für Mitglieder der DGAI und/oder des BDA ist der Bezug der Zeitschrift im Mitgliedsbeitrag enthalten.

Die Beiträge aus der A&I finden Sie online unter: **www.ai-online.info**

Indexed in **Current Contents®/Clinical Medicine, EMBASE/Excerpta Medica; Medical Documentation Service; Research Alert; Sci Search; SUBIS Current Awareness in Biomedicine; VINITI: Russian Academy of Science.**

Wichtiger Hinweis
Für Angaben über Dosierungsanweisungen und Applikationsformen kann vom Verlag und den Herausgebern keine Gewähr übernommen werden. Derartige Angaben müssen vom jeweiligen Anwender im Einzelfall anhand anderer Literaturstellen auf ihre Richtigkeit überprüft werden. Gleiches gilt für berufs- und verbandspolitische Stellungnahmen und Empfehlungen.

Allein aus Gründen der besseren Lesbarkeit wird auf die gleichzeitige Verwendung männlicher, weiblicher und weiterer Sprachformen verzichtet. Sämtliche Personenbezeichnungen gelten für alle Geschlechterformen. Dies impliziert keinesfalls eine Benachteiligung der jeweils anderen Geschlechter, sondern ist als geschlechtsneutral zu verstehen.

CONTACT US

Please do not hesitate to contact us. We will be glad to answer and provide further information to you at any time.

.....
Name

.....
First Name

.....
Department / Hospital

.....
Place

.....
Telephone

.....
E-Mail

.....
Date / Signature

Please contact me for further information ○

I would like to participate in the project ○

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