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

www.ai-online.info

64. Jahrgang | Dezember 2023

A&I

ANÄSTHESIOLOGIE & INTENSIVMEDIZIN

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

Congenital cataracts, facial dysmorphism and neuropathy syndrome

Hermansky-Pudlak syndrome

orphanaesthesia

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

SUPPLEMENT NR. 15 | 2023

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.

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

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

orphananesthesia

Anaesthesia recommendations for

Hermansky-Pudlak syndrome

Disease name: Hermansky-Pudlak syndrome

ICD 10: E70.3

Synonyms: /

Disease summary: Hermansky-Pudlak syndrome (HPS) is a rare autosomal recessive disease characterised by abnormal intracellular vesicle formation and trafficking [7,17]. It was first described in 1959 [11]. There are a few hot spots of HPS in many regions of the world [6]. Generally, there is a lack of data regarding the disease's real frequency. Its prevalence is estimated with 1 in 500,000 to 1 in 1,000,000 [14,21]. For unknown reasons, , an unusual high incidence of up to 1:1,800 persons has been identified in the northwestern part of Puerto Rico [6].

There are 11 known genetically distinct subtypes of HPS (HPS-1 to HPS-11) [6,7]. Mutations result in defects in intracellular protein trafficking and in the biogenesis of lysosomes as well as the biogenesis of lysosome-related organelles (BLOCs), such as melanosomes and platelet-dense granules [6,23].

Oculocutaneous albinism (OCA) is characterised by various degrees of hypopigmentation of hair, skin retina and iris. A decline in visual acuity may be accompanied by retinal hypopigmentation, horizontal nystagmus, photophobia, and strabismus [2,6,21,23]. HPS-2 patients may also result in immunodeficiency from severe neutropenia, often treated with G-CSF [10].

In addition, HPS-1 and HPS-4 are at high risk to develop pulmonary fibrosis in adults as well as HPS-2 in children. Fibrosis is supposed to be promoted by lysosomal accumulation of ceroid-lipofuscin in alveolar macrophages, disruption of type-II pneumocytes and a consecutive inflammation cascade [1,7,23,28]. In adults, interstitial, irreversible and progressive lung diseases usually develop in the third decade of life.

Due to a dysfunction in platelet aggregation in HPS, anamnesis often reveals a history of bruising, nose bleeding, menorrhagia or (unexpected) prolonged bleeding after dental extractions or other surgical procedures [6,20,22,23]. However, the severity of the bleeding tendency may widely vary between patients with HPS [5]. In the absence of dense bodies, the bleeding time may be prolonged despite a regular number of platelets and inconspicuous coagulation factors [5,12,18,29].

Moreover, in about 15% of HPS (type 1–4) a patient's phenotypic presentation may include granulomatous colitis with crampy abdominal pain, weight loss, malabsorption, and frequent watery, bloody diarrhoea [6,9,12,23]. Cardiomyopathy and renal glomerular dysfunction secondary to ceroid deposition have been reported as rare complications of HPS [12].

▶ **Citation:** Gaik C, Wiesmann T: Hermansky-Pudlak syndrome. Anästh Intensivmed 2023;64:S454–S464.
DOI: 10.19224/ai2023.S454

The diagnosis of the disease is based on clinical features including the typical triad of OCA accompanied by a bleeding disorder due to platelet dysfunction [6,11]. The latter is demonstrated by th absence of platelet dense bodies (δ granules) on whole-mount electron microscopy of platelets, which represents the “gold standard” test for platelet abnormality in HPS [6,7,28].

Molecular genetic analysis is recommended to determine the specific subtypes of HPS in order to plan follow-up procedures and better estimate the prognosis in affected patients [6]. However, detailed diagnostics are complex and genetic testing and confirmation are expensive and usually only available on a research basis [2]. Furthermore, the diagnosis of an intestinal lung disease is established with a high-resolution computed tomography of the chest (HRCT), which also provides good radiologic monitoring of disease status and progression [1]. Alung biopsy is not necessary for diagnosis and may merely provoke bleeding complications [6,28].

There are currently no definitive therapeutic or even preventative approaches for HPS [6]. Therefore, therapy is solely symptomatic.

The prognosis of HPS usually depends on the progression of lung fibrosis and in patients with HPS-2 on the immunological defect. The course of pulmonary fibrosis is variable but universally progressive and a major cause of mortality in adults, especially in HPS-1 [7,8,9]. Generally, because of absent efficacy, a corticosteroid or immunosuppressive therapy is not recommended in HPS patients with pulmonary fibrosis [28]. Administration of pirfenidone as a new therapy approach is assumed to slow down the progression of fibrosis, but its use is controversial and not yet generally recommended [8,15]. Patients with HPS-1, HPS-2 and HPS-4 may be referred for lung transplant evaluation to prolong life expectancy [6,7]. Finally, lung transplantation is the only curative therapy for pulmonary fibrosis now [17]. However, the treatment of a Crohn’s disease-like gastrointestinal disorder in HPS has been reported to respond well to immunosuppressive therapy [9]. Unfortunately, the average life expectancy of patients with HPS is 40–50 years (especially in cases of pulmonary impairment) [28].

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

© Anästh Intensivmed 2023;64:S454–S464 Aktiv Druck & Verlag GmbH

Emergency information

A	**AIRWAY / ANAESTHETIC TECHNIQUE**	No definite recommendation for either GA or RA, but GA may be preferred (TIVA / balanced) – however, be aware of difficult mask ventilation and rapid desaturation because of restrictive pulmonary dysfunction (high risk for pulmonary fibrosis of variable course in HPS-1, -2, -4) – (modified) RSI in case of gastrointestinal affection – perform lung-protective ventilation – avoid neuraxial RA (bleeding complications!) or only use it with extreme caution.
B	**BLOOD PRODUCTS (COAGULATION)**	Increased bleeding tendency due to qualitative platelet disorder (platelet number and routine haematology tests may be normal!) – prematching of RBCs and 24h-availability of haematologic / coagulation monitoring recommended – consider perioperative haematologic consultation – inform your local transfusion specialist in advance for sufficient storage of blood products, especially platelets! – consider perioperative DDAVP, PPSB, TXA, rFVIIa or platelet transfusion and cell saving strategies to prevent (life-threatening!) bleeding complications.
C	**CIRCULATION**	Tno specific recommendations
D	**DRUGS**	No risk for MH – consider malabsorption of oral medication (gastrointestinal affection) – avoid NSAIDs (bleeding complications)
E	**EQUIPMENT**	Ambulatory anaesthesia not recommended – patient positioning / mobilisation with caution (easy bruising), strive for optimal padding – use neuromuscular monitoring and ultrasound for vessel canulation / peripheral RA – be aware of perioperative respiratory complications due to fibrotic lung disease (up to VV-ECMO necessity) and delayed bleeding complications – help might be necessary due to visual impairment

Typical surgery

Lung transplantation [7,24], video-assisted thoracic surgery (VATS) [14,15,16], bronchoscopies for routine surveillance or taking transbronchial biopsies, any kind of lung biopsy (even if not routinely recommended for diagnosis – see above) [7].

Intestinal surgery as a last resort in case of inflammatory bowel disease (resembling Crohn's colitis clinically) [6,23].

Ophthalmologic surgery like frequently necessary strabismus correction [3].

Type of anaesthesia

Despite reports of successful performance in pregnant women with HPS, spinal or peridural anaesthesia should be avoided or only be used with extreme caution in affected patients due to the risk of bleeding complications [2,19,21,25,27]. Besides, there are reports of local infiltration of anaesthetics for dental surgery without complications [18]. No reports were made for the performance of ultrasound-guided peripheral nerve blocks. There is no definite recommendation for either general or regional anaesthesia. Basically, general anaesthesia may be preferred in patients with HPS due to varying bleeding tendency in these patients. Any neuraxial anaesthesia (e.g., spinal or epidural) should be decided individually for each patient and surgery by weighing the risks and benefits of these procedures. Peripheral regional anaesthesia might be performed in patients with bleeding disorders with adequate ultrasound guidance and approaches with low risks for vessel puncture.

General anaesthesia can be performed as balanced anaesthesia with volatile anaesthetics or as total intravenous anaesthesia (TIVA). Monitoring of neuromuscular block is advised before emergence of the anaesthesia, especially in patients with known pulmonary impairment due to HPS.

Using ultrasound visualisation of correct wire localisation within the blood vessel for placement of central venous line in V. jugularis interna / externa or V. subclavia is strictly recommended to avoid vascular damage (e.g., dissection) accidental pneumothorax or lung injury with respect to widespread pulmonary impairment in these patients.

Adequate prophylaxis of postoperative nausea and vomiting is recommended as usual.

Intraoperative patient positioning should focus on optimal padding of the patient to avoid easy bruising in patients with known bleeding tendency.

Necessary additional preoperative testing (beside standard care)

As usual, the clinical condition and (pulmonary) status of patients with HPS is most important pre-operative (i.e., exercise intolerance via walking test for 6 minutes or other exercise testing). A chest X-ray may reveal extensive bilateral interstitial disease in patients with pulmonary involvement. Computed tomography can be performed when X-ray results remain unclear [1,6,17]. Spirometry may quantify the current degree of the pulmonary impairment.

Moreover, a thorough anamnesis should focus on bleeding and haematological complications. Evaluating platelet count and coagulation parameters may be performed preoperatively. Nevertheless, one should be aware that despite affected platelets in HPS this is a qualitative platelet dysfunction and that the total number of platelets as well as routine haematology tests (i.e., prothrombin time, activated partial thromboplastin time, von Willebrand factor) are usually within normal ranges [6,7,17]. There are numerous reports of a typically prolonged bleeding time, an impaired secondary platelet aggregation and a reduction in platelet nucleotides associated with an increased ATP/ADP ratio [19,21,27]. Highly specialised laboratory tests with screening platelet aggregation and ATP secretion in

© Anästh Intensivmed 2023;64:S454–S464 Aktiv Druck & Verlag GmbH

response to various platelet stimulatory agents is probably not available everywhere [13]. However, an abnormal platelet aggregation is neither pathognomonic for HPS nor does the finding of a normal aggregation give definite proof of its absence [23]. Whenever practical and possible, it is recommended to perform most surgical procedures in a hospital with 24 hours of postoperative haematologic and coagulation monitoring [23].

With respect to minimise intraoperative fluid requirement in cases of pulmonary impairment and as a prophylaxis against bleeding, there are reports of preventive platelet administration and / or perioperative intravenous infusion of desmopressin (DDAVP), whereby the clinical response to DDAVP may be variable [7,14,17,18,20,21,27]. Moreover, there is one case report about the successful use of tranexamic acid as well as recombinant activated factor VII (rFVIIa) to prevent bleeding in a patient with HPS [5].

Like other patients with lung diseases, patients with pulmonary fibrosis due to HPS should be provided with influenza immunisation and pneumococcal vaccination, in the absence of contraindications [6,23].

Particular preparation for airway management

As far as known, there are no anatomic peculiarities due to HPS itself. Nevertheless, a standardised approach for airway examination and detection of airway challenges is recommended. A thorough preparation for airway management should be based on the examination results.

However, mask ventilation may be difficult due to the patient's decreased pulmonary compliance [14]. Due to typically low pulmonary reserves in patients with HPS, one should be aware of a high risk of rapid desaturation in case of complications in airway management [14].

HPS may also affect the gastrointestinal tract by a granulomatous colitis with inflammatory bowel disease [6,9]. In case of associated gastrointestinal reflux, a rapid sequence induction may be considered. Due to the above mentioned pulmonary morbidities, a modified RSI approach might be better than a classic RSI approach.

Particular preparation for transfusion or administration of blood products

Due to a disease-associated bleeding tendency, prematching of RBCs is advised for patients with HPS. Inform your local tranfusion specialist in advance for storage of sufficient numbers of blood products, especially platelets. Depending on the type of surgery, acute bleeding may occur and an aggressive haemostatic therapy is advisable i.e. with platelet transfusion, prothrombin complex concentrate, desmopressin or tranexamic acid [7]. Furthermore, cell saving strategies might be advisable.

There is a lack of data without any general recommendation in the management for bleeding in patients with HPS and individualised therapeutic plans for management of potential peri-operative bleeding are appropriate for affected patients [7]. With respect to a perspectively transplantation a restrictive transfusion approach may help to avoid alloimmunisation. For the same reason, only HLA-matched leucodepleted platelet concentrates should be used whenever possible, although random platelets may be needed in a critical emergency [7,29].

© Anästh Intensivmed 2023;64:S454–S464 Aktiv Druck & Verlag GmbH

Particular preparation for anticoagulation

Prophylactic or therapeutic anticoagulation should be performed only after weighing the risks and benefits of possible thromboembolic and bleeding complications. A thorough evaluation of the paient's bleeding history is better than laboratory tests to identify patients at risk for bleeding. Regarding the functional platelet defect in HPS, a comprehensive haematologic consultation is recommended to assess the bleeding risk that may require utilising extracorporeal membrane oxygenation (ECMO) in affected patients [7,24].

Particular precautions for positioning, transportation and mobilisation

Be aware of easy bruising in patients with known bleeding tendency during positioning, transport or mobilisation.

Interactions of chronic disease and anaesthesia medications

Not reported.

Anaesthetic procedure

Preoperative evaluation: see details above.

Premedication might be applied after weighing the benefits and risks in individual patients. Patients with granulomatous colitis may have malabsorption of oral medications – premedication may be given intravenously to these patients [16]. Sedative premedication should be avoided in pulmonary compromised patients.

There are no specific recommendations when it comes to patient positioning. Thorough padding is helpful to avoid bruises in patients prone to related complications.

IV line: no specific recommendations for peripheral IV line. Using ultrasound visualisation of correct wire localisation within the blood vessel for placement of central venous line into the V. jugularis interna/externa or V. subclavia is strictly recommended to avoid an accidental pneumothorax or lung injury considering the widespread pulmonary impairment in these patients.

Invasive blood pressure measurement: facilitates frequent arterial blood gas analysis, especially in cases of pulmonary impairment.

(Mechanical) ventilation: A restrictive pulmonary dysfunction is characteristic in patients with HPS; moreover, ventilation should be performed in a way which is lung-protective and includes adequate low tidal volumes to avoid baro-/volutrauma [14,17]. Single-lung ventilation during lung surgery may be challenging due to recurring hypoxaemia with a need for intermittent bilateral lung ventilation [16].

Anaesthesia: Total intravenous or balanced anaesthesia using volatile anaesthetics can be performed safely. There are no absolute or known relative contraindications for anaesthesia-related drugs just because of the disease HPS. There is no specific risk of malignant hyperthermia.

© Anästh Intensivmed 2023;64:S454–S464 Aktiv Druck & Verlag GmbH

Local/regional anaesthesia can be performed as described above. With respect to a potentially increased bleeding risk, regional anaesthesia should be primarily considered in patients severely affected by pulmonary problems in order to reduce the patient's work of breathing [14]. Despite reports of successful performance in patients with HPS, neuraxial (spinal or peridural) anaesthesia should be avoided or only performed with extreme caution in affected patients with a positive bleeding anamnesis due to bleeding complications [2,19,21].

Particular or additional monitoring

Extended haemodynamic monitoring may help to optimise intraoperative fluid management, especially in case of any significant blood loss that requires differentiated and careful transfusion and fluid administration. Hypervolaemia may be detrimental in patients with severe pulmonary dysfunction [14].

Possible complications

Severe respiratory failure / hypoxaemia.

Life-threatening bleeding complications.

Postoperative care

Postoperative care should focus on respiratory failure due to fibrotic lung disease which might complicate postoperative care [14]. Respiratory function should be monitored during an appropriately extended stay in PACU, IMC or ICU before transfer to the normal ward (or discharge at home) is acceptable.

Furthermore, the development of bleeding and haematoma at the operation site should be followed closely.

Providing adequate analgesia is an important challenge in the anaesthetic management of patients with HPS because of its value and influence on respiratory function. Analgesic medications such as nonselective nonsteroidal antiinflammatory drugs (NSAIDs) should be avoided due to potential bleeding complications [6,14,23,28]. Weighing the risks (thromboembolic events) and benefits (sufficient analgesia) selective COX-2 NSAIDs may be considered as alternatives [23]. There are no HPS-specific contraindications for opioid agents [23]. In case of pulmonary impairment one should be aware of impaired spontaneous breathing and consecutive respiratory acidosis [14].

A thorough dermatologic assessment and aggressive skin and eye protection are essential in the pre-transplant and post-transplant periods, given the increased risk of squamous cell carcinomas of the skin, basal cell carcinomas and melanomas due to hypopigmentation and perioperative immunosuppressive therapy [6,7,17,23].

© Anästh Intensivmed 2023;64:S454–S464 Aktiv Druck & Verlag GmbH

Disease-related acute problems and effect on anaesthesia and recovery

Emergency-like situations are life-threatening bleeding complications, a recurrent/acute pneumothorax, acute exacerbations of pulmonary fibrosis all the way to severe respiratory failure and the necessity of a veno-venous ECMO (often as a bridging therapy for final lung transplantation) [7,24,26].

Differential diagnostics should focus on idiopathic pulmonary fibrosis (IPF), nonspecific interstitial pneumonia (NSIP), pulmonary fibrosis due to a variety of other causes, Chediak-Higashi syndrome, Griscelli syndrome [6,27].

Ambulatory anaesthesia

We strongly encourage to operate patients with HPS in centres where haematologists/ haemostaseologists and ICU facilities are available 24 hours a day due to the risk of bleeding/respiratory complications in the postoperative setting. A careful follow-up after discharge is also prudent to check for any delayed bleeding [17].

Obstetrical anaesthesia

Patients with HPS are fertile, thus the obstetrical anaesthetist might face women with HPS for labour analgesia. Pulmonary manifestation, frequently occurring in the women's fertile age, may aggravate pregnancy and labour.

Besides, bleeding complications are the main problems during delivery. Despite reports of successful performance in pregnant women with HPS, spinal or peridural anaesthesia should be avoided or only be used with extreme caution in affected patients due to bleeding complications [2,19,21,25,27]. Therefore, anamnesis should focus on former episodes of epistaxis, easy bruising, gastrointestinal bleeding, prolonged bleeding during menstruation or tooth extraction as well as prolonged post-operative bleeding or even post-partum haemorrhage [2,4]. The mode of delivery must be decided individually [27]. Many authors recommend a vaginal birth in the aim to minimise bleeding complications [2]. Nevertheless, even in vaginal delivery bleeding may complicate post-partum period and moreover, there is a report of uneventful caesarean section in a woman with HPS receiving i.a. preventive platelets [19]. Referring to these issues, decision making and preventive administration of i.e., tranexamic acid, DDAVP or platelet transfusion may be facilitated with consideration of haematologists/haemostaseologists, especially in view of frequently normal test results for i.e. platelet count, prothrombin time and partial thromboplastin time in patients with HPS [2,4,25,27,29].

Women with HPS must be considered to be at risk of serious bleeding during labour. Therefore, an early predefined interdisciplinary schedule is recommended for safe and preconceived action especially in emergency situations involving perioperative pregnant women with HPS .

In summary, pregnant women with HPS should be supervised in specialised centres following a multidisciplinary approach. A well-considered peripartum decision-making and management is indispensable in the setting of rare HPS. A multidisciplinary team including obstetricians, anaesthesiologists and haematologists/haemostaseologists should discuss an individual patient-specific approach for an optimal and maximum safe peri-partum management [2].

© Anästh Intensivmed 2023;64:S454–S464 Aktiv Druck & Verlag GmbH

References

1. Avila NA, Brantly M, Premkumar A, Huizing M, Dqyer A, Gahl WA. Hermansky-Pudlak Syndrome: Radiography and CT of the Chest Compared with Pulmonary Function Tests and Genetic Studies. Am J Roentgenol 2002;179:887–892
2. Bachmann C, Abele H, Wallwiener D, Kagan KO. Neonatal and maternal risk in Hermansky-Pudlak syndrome : peripartum management – brief report and review of literature. Arch Gynecol Obstet 2014;289:1193–1195
3. Bagheri A, Abdollahi A. Hermansky-Pudlak Syndrome; a Case Report. J Ophthalmic Vision Res 2010;5:269–272
4. Beesley RD, Robinson RD, Stewart TL. Two Successful Vaginal Births after Caesarean Section in a Patient with Hermansky-Pudlak Syndrome Who Was Treated with 1-Deamino-8-Arginine-Vasopression during Labor. Mil Med 2008;173;10:1048
5. Del Pozo Pozo AI, Jiménez-Yuste V, Villar A, Quintana M, Hernández-Navarro F. Successful thyroidectomy in a patient with Hermansky-Pudlak syndrome treated with recombinant factor VII and platelet concentrates. Blood Coagul Fibrinolysis 2002;13:551–553
6. El-Chemaly S, Young LR. Hermansky-Pudlak Syndrome. Clin Chest Med 2016;37:505–511
7. El-Chemaly S, O'Brien KJ, Nathan SD, Weinhouse GL, Goldberg HJ, Connors JM, et al. Clinical management and outcomes of patients with Hermansky-Pudlak syndrome pulmonary fibrosis evaluated for lung transplantation. PLoS ONE 2018;13:e0194193
8. Furuhashi K, Enomoto N, Fujisawa T, Hashimoto D, Inui N, Nakamura Y, et al. Hermansky-Pudlak Syndrome with Nonspecific Interstitial Pneumonia. Intern Med J 2014;53:449–453
9. Girot P, Le Berre C, De Maissin A, Freyssinet M, Trang-Poisson C, Bourreille A. Crohn's-like acute severe colitis associated with Hermansky-Pudlak syndrome: A case report. World J Gastroenterol 2019;25:1031–1036
10. Hengst M, Naehrlich L, Mahavadi P, Grosse-Onnebrink J, Terheggen-Lagro S, Skanke LH, et al. Hermansky-Pudlak syndrome type 2 manifests with fibrosing lung disease early in childhood. Orphanet J Rare Dis 2018;13:42
11. Hermansky F, Pudlak P. Albinism Associated With Hemorrhagic Diathesis and Unusual Pigmented Reticular Cells in the Bone Marrow: Report of Two Cases With Histochemical Studies. Blood 1959;14:162–169
12. Huizing M, Gahl WA. Disorders of Vesicles of Lysosomal Lineage: The Hermansky-Pudlak Syndromes. Curr Mol Med 2002;2:451–467
13. Hurford MT, Sebastiano C. Hermansky-Pudlak Syndrome: Report of a Case and Review of Literature. Int J Clin Exp Pathol 2008;1:550–554
14. Ideno S, Hatori E, Takeda J, Morisaki H. Anesthetic management of a patient with Herman-Pudlak syndrome undergoing video-assisted bullectomy. J Clin Anesth 2015;27:243–246
15. Kanazu M, Arai T, Sugimoto C, Kitaichi M, Akira M, Abe Y, et al. An Intractable Case of Hermansky-Pudlak Syndrome. Intern Med 2014;53:2629–2634
16. Kaseda K, Goto T, Emoto K, Hayashi Y. Surgery for secondary pneumothorax caused by Hermansky-Pudlak syndrome. Eur Respir J 2014;44:1715–1717
17. Lederer DJ, Kawut SM, Sonett JR, Vakiani E, Seward SL, White JG, et al. Successful Bilateral Lung Transplantation for Pulmonary Fibrosis Associated With Hermansky-Pudlak Syndrome. J Heart Lung Transplant 2005;24:1697–1699
18. Minkin P, Bertetti R, Liindsey S, Bovino B. Management of Tooth Extraction in a Patient With a Rare Bleeding Disorder Associated with Hermansyk-Pudlak Syndrome: A Case Report. J Oral Maxillofac Surg 2015;73:219–223
19. Nisal M, Pavord S, Oppenheimer CA, Francis S, Khare M. Hermansky-Pudlak syndrome: Management of a rare bleeding disorder in a twin pregnancy. J Obstet Gynaecol 2012;32:185–186

20. Ozgur M, Yilmaz B. Unexpected intra-operative bleeding due to Hermansky-Pudlak Syndrome. Indian J Anaesth 2015;59:393–394
21. Poddar RK, Coley S, Pavord S. Hermansky-Pudlak syndrome in a pregnant patient. Br J Anaesth 2004;93:740–742
22. Rivera-Concepción J, Acevedo-Canabal J, Burés A, Vargas G, Cadilla C, Izquierdo NJ. Bleeding Assessment in female patients with Hermansky-Pudlak syndrome – A case series. Eur J Haematol 2019;102:432–436
23. Seward SL, Gahl WA. Hermansky-Pudlak Syndrome: Health Care Throughout Life. Pediatrics 2013;132:153–160
24. Sim W, Kim SY, Han J, Rim TH, Lee JG, Paik HC, et al. Extracorporeal Membrane Oxygenation Bridge to Lung Transplantation in a Patient with Hermansky-Pudlak Syndrome and Progressive Pulmonary Fibrosis. Acute Crit Care 2019;34:95–98
25. Spencer J, Rosengren S. Hermansky-Pudlak Syndrome in Pregnancy. Am J Perinatol 2009; 26:617–620
26. Umei N, Ichiba S, Chida S. Successful use of veno-venous extracorporeal membrane oxygenation as a bridge to lung T transplantation in a patient with pulmonary fibrosis. Gen Thorac Cardiovasc Surg 2017;65:478–480
27. Van Avermaete F, Muys J, Jacquemyn Y. Management of Hermansky-Pudlak syndrome in pregnancy and review of literature. BMJ Case Rep 2016 Nov 17;2016:bcr2016217719. DOI: 10.1136/bcr-2016-217719
28. Vicary GW, Vergne Y, Santiago-Cornier A, Young LR, Roman J. Pulmonary Fibrosis in Hermansky-Pudlak Syndrome. Ann Am Thorac Soc 2016;13:1839–1846
29. Yusuf L, Srivasavi D, Ciantar E. Hermansky-Pudlak syndrome in pregnancy: A case report. Obstet Med 2016;9:171–173.

Date last modified: **February 2022**

This recommendation was prepared by:

Authors

Christine Gaik, Anaesthesiologist, Universitätsklinikum Marburg, Germany
gaikc@med.uni-marburg.de

Thomas Wiesmann, Anaesthesiologist, Diakonie-Klinikum Schwäbisch Hall, Germany
thomas.wiesmann@diakoneo.de

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

This recommendation was reviewed by:

Reviewers

Matthias Griese, Paediatrician, Department of Paediatric Pneumology, University-Clinic Munich, Germany
matthias.griese@med.uni-muenchen.de

Barbara Zieger, Paediatrician, Clinic for Paediatric Haematology and Oncology, University-Clinic Freiburg, Germany
barbara.zieger@uniklinik-freiburg.de

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 specialists instead.

© Anästh Intensivmed 2023;64:S454–S464 Aktiv Druck & Verlag GmbH

Herausgeber

DGAI
Deutsche Gesellschaft für Anästhesiologie und Intensivmedizin e. V.
Präsident: Prof. Dr. B. Pannen, Düsseldorf

BDA
Berufsverband Deutscher Anästhesistinnen und Anästhesisten e. V.
Präsidentin: Prof. Dr. G. Beck, Mannheim

Schriftleitung
Präsident/in der Herausgeberverbände
Gesamtschriftleiter/Editor-in-Chief:
Prof. Dr. Dr. Kai Zacharowski, ML FRCA FESAIC, 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:
Neuwieder Straße 9 | 90411 Nürnberg | Deutschland | Tel.: 0911 9337812
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 Müller | Robert Kux
Tel.: 09522 943570 | Fax: 09522 943577
E-Mail: anzeigen@aktiv-druck.de

Verlagsrepräsentanz
Jürgen Distler
Neuwieder Straße 9 | 90411 Nürnberg
Tel.: 0171 9432534
E-Mail: jdistler@bda-ev.de

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

Titelbild
Dipl.-Designerin Monique Minde, Nürnberg

Erscheinungsweise 2023
Der 64. 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.

Allgemeine Geschäfts- und Lieferbedingungen
Die allgemeinen Geschäfts- und Lieferbedingungen entnehmen Sie bitte dem Impressum auf www.ai-online.info

Indexed in **CINAHL; Current Contents®/ Clinical Medicine, EBSCO; EMBASE/ Excerpta Medica; Medical Documentation Service; Research Alert; Sci Search; Scopus; 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.

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

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
Neuwieder Straße 9 | 90411 Nuremberg | Germany
Tel.: +49-911-933780
Email: info@orphananesthesia.eu