

# A&I

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**Gaucher's disease**

orphan**a**nesthesia

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

**SUPPLEMENT NR. 7 | 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](http://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](http://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](http://www.ai-online.info/Orphsuppl)  
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**Find a survey of the recommendations published until now on:**

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# orphan**a**nesthesia

## Anesthesia recommendations for **Gaucher's disease**

**Disease name:** Gaucher's disease

**ICD 10:** E75.2

**Synonyms:** sphingolipidosis (lysosomal storage disorder, deficiency of glucocerebrosidase)

**Disease summary:** Gaucher's disease is one of the most common lysosomal storage disorders with defects in the enzyme glucosylceramidase (glucocerebrosidase). The disease is caused by mutations in the GBA gene on chromosome 1 (autosomal recessive) and affects both sexes. The incidence is estimated to be 1:40,000 in Europe. Most commonly, the disease is differentiated into a non-neuronopathic form and a neuronopathic form. Thus, patients may present with symptoms early during childhood or later in life. Patients may develop organ damage with some manifestations in the coagulation system relevant to anesthesia, an impaired immune competence, reduced lung function and elevated pulmonary arterial pressure in adults [1,2]. A specific therapy with eliglustat may interact with perioperative medication and should be paused for 48 hours before surgery. Neuraxial anesthesia, regional-anesthesiologic nerve blocks and general anesthesia can be performed safely in these patients with individual risk assessment.

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Medicine is in progress



Perhaps new knowledge

Every patient is unique

Perhaps the diagnosis is wrong

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Find more information on the disease, its centers of reference and patient organizations on Orphanet: [www.orpha.net](http://www.orpha.net)

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### Emergency information

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<b>A</b>	<b>AIRWAY / ANESTHETIC TECHNIQUE</b>	All types of anesthesia possible with individual risk assessment. Prefer regional anesthesia in pulmonary comorbidity, reevaluate potential coagulopathy carefully.
<b>B</b>	<b>BLOOD PRODUCTS (COAGULATION)</b>	Standard coagulation tests (partial thromboplastin time, INR, blood count). If bleeding history, advanced tests are recommended (e.g., thromboelastography, coagulation factors).
<b>C</b>	<b>CIRCULATION</b>	No specific recommendation.
<b>D</b>	<b>DRUGS</b>	Significant interaction possible with <i>eliglustat</i> with CYP2D6 inhibitors inducing toxicity – stop <i>eliglustat</i> 48 hours before surgery. In emergency cases, check for drug interaction (5HT3-antagonists, beta-blockers, opioids, anti-infectives).
<b>E</b>	<b>EQUIPMENT</b>	No specific recommendation.

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### Typical surgery and procedures

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Patients with Gaucher's disease may present for all types of surgery. However, as the disease can lead to splenomegaly and was also described to be associated with femoral head necrosis and bone pathology, spleen extirpation and musculoskeletal surgery are typical for these patients.

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### Type of anesthesia

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The anesthesiologic relevant aspects in Gaucher's disease are related to

- Hematopoietic system with anemia, thrombocytopenia, impaired coagulation capacity and leucopenia with impaired immune competence,
- Impaired lung function,
- Impaired CNS function including epilepsy and dysphagia,
- Pulmonary arterial hypertension in adult patients, e.g., receiving enzyme replacement therapy.

Based on current knowledge, all types of anesthesia can be performed safely. The choice of anesthesia should depend on the specific manifestations of the individual patients. If possible, regional anesthesia should be preferred especially in patients with pulmonary complications but should be evaluated carefully against the background of potential coagulopathy. In general, anesthesia, volatile and intravenous hypnotics can be administered. In patients with a history of coagulopathy or abnormal laboratory coagulation tests, neuraxial anesthesia should be avoided.

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### Necessary additional preoperative testing (beside standard care)

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Standard preoperative measures to evaluate patients and an interdisciplinary case review (e.g., involving surgeon, anesthesiologist and hematologist) will allow individualized decision-making for additional diagnostic testing. This should also focus on a detailed patient history of clinical signs of bleeding (e.g., gum bleeding, heavy menstruation bleeding, spontaneous hematoma) and obtaining blood tests. Standard coagulation tests (partial thromboplastin time, INR, blood count) can be expanded to identify impaired hemostasis like thrombocytopeny (thromboelastography) or deficiency of coagulation factors [3].

Furthermore, the pulmonary and cardiovascular capacity should be evaluated to assess metabolic equivalents and signs for cardiac decompensation (as described in guidelines for evaluation of adults before non-cardiac surgery). In specific cases, ECG and transthoracic sonography should be performed to determine ejection fraction, valvular defects (like tricuspid insufficiency) and elevated pulmonary-arterial pressure. Specific attention should also be paid to the medication intake of patients as some patients may be treated with eliglustat (Cerdelga®) which may interact with drugs during anesthesia.

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### Particular preparation for airway management

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There is currently no evidence of specific preparations necessary in these patients.

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### Particular preparation for transfusion or administration of blood products

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Standard preparation for scheduled surgery is advisable. However, in patients with clinical evidence of coagulopathy or those with thrombocytopenia, preoperative measures are to be discussed interdisciplinary with surgeons, anesthesiologists and patients. For patients with an increased risk of bleeding complications, blood products should be prepared.

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### Particular preparation for anticoagulation

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Gaucher's disease seems not to be associated with thrombotic events but has some risks for increased bleeding. In patients with clinical evidence of coagulopathy or those with thrombocytopenia, an individualized risk assessment is necessary. If there is an increased risk for perioperative thrombotic events, anticoagulation treatment is indicated as standard of care.

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### Particular precautions for positioning, transportation and mobilization

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Gaucher's disease is associated with musculoskeletal deformities that will be evaluated in pre-operative assessment. Based on that, individual measures are to be planned. However, there are no specific points to consider in these patients according to current evidence.

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### Interactions of chronic disease and anesthesia medications

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Some patients will be treated with specific agents approved for Gaucher's disease. Specific caution is necessary in patients receiving eliglustat (Cerdelga®), a drug acting as highly specific inhibitor of enzymes for glucocerebrosidase syntheses [6,7]. This substrate-reducing therapy may interfere with perioperative drug therapy. Eliglustat is metabolized strongly by hepatic CYP2D6 enzymes and to a lesser extent by CYP3A4. Thus, all inhibitors of these pathways may increase the toxicity of eliglustat and should be avoided. Indeed, not only CYP2D6 inhibitors but also inducers or substrates of cytochrome P450 (CYP) enzymes may affect the metabolism of the drug. For concomitant treatment, data bases for drug interaction may be a source of further information as are clinical pharmacologists.

For perioperative use, this interaction may affect the following drugs (examples):

- 5HT3-antagonists (ondansetron),
- Beta blockers,
- Opioids like codeine and tramadol,
- Anti-infectives (rifampicin, erythromycin, voriconazole, posaconazole, clarithromycin, ciprofloxacin, etc.).

Interaction with eliglustat may cause significant adverse events, with long-QT-syndrome and sequelae like Torsade-de-pointes-tachycardia.

Due to the circulating half-life of eliglustat of 7-9 hours, the drug should be stopped about 48 hours before surgery.

There are no specific anesthetic implications for patients treated with enzyme replacement therapy.

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### **Anesthetic procedure**

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Gaucher's disease does not alter standard anesthetic procedures. In patients with coagulopathy, hemostasis should be optimized preoperatively.

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### **Particular or additional monitoring**

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The choice of monitoring and additional measures perioperatively should be adapted according to the patient's individual conditions.

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### **Possible complications**

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Perioperative bleeding complications should be anticipated. In patients with concomitant use of eliglustat, the risk for cardiac events may be increased. In patients with Gaucher's disease, there is an increased risk of postoperative infections, thus, antibiotic prophylaxis should be administered.

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### **Postoperative care**

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The choice of postoperative care should be adapted to the patient's individual condition. In patients with a high risk of bleeding, postoperative pain management should evaluate the choice of drugs used including increased risk related to NSAIDs use.

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### **Disease-related acute problems and effect on anesthesia and recovery**

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There are no specific points to consider.

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### **Ambulatory anesthesia**

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The choice between ambulatory versus in-hospital care should be adapted to the patient's individual conditions.

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### **Obstetrical anesthesia**

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The choice of anesthesia in obstetric surgery and to support birth follows an individual risk assessment. Due to potential interactions with the coagulation system and with physiologic changes in pregnancy, blood test results should be available to assess the current status of platelet count and coagulation parameters. There is some evidence in literature that neuraxial anesthesia like epidural anesthesia or spinal anesthesia are possible modalities during birth. However, the risk of perioperative bleeding could be increased [4,5].

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