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# A&I

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## Kartagener syndrome

## Kennedy disease

orphananesthesia

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

SUPPLEMENT NR. 10 | 2021

## 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.

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## 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.

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orphananesthesia

# Anaesthesia recommendations for

# Kennedy disease

**Disease name:** Kennedy disease

**ICD 10:** G 12.1

**Synonyms:** Spinal and bulbar muscular atrophy, X-linked spinal and bulbar muscular atrophy, bulbospinal muscular atrophy

**Disease summary:** Kennedy disease (KD) is an adult-onset, X-linked recessive trinucleotide, polyglutamine (poly-G) disorder, caused by expansion of a polymorphic CAG tandem-repeat in exon 1 of the androgen-receptor (AR) gene on chromosome Xq11-12 [1]. The exact mechanism resulting in neuronal degeneration and loss is currently unknown; however, the severity of disease does appear to increase with increasing number of trinucleotide repeats.

The estimated world-wide incidence of KD is approximately one in 40,000 males. Because of the X-linked transmission, KD almost exclusively affects males but is transmitted by female carriers [2].

Patients usually present between 30 and 50 years of age with amyotrophic weakness and wasting of the facial, bulbar and proximal limb muscles. There are occasionally sensory and/or endocrine disturbances, such as androgen resistance, gynaecomastia, elevated testosterone or progesterone, and reduced fertility [3-5]. Specific initial symptoms may include tremor, muscle cramping, fatigue, and slurred speech. As the disease progresses, patients may develop difficulty chewing and swallowing, tongue wasting, dysarthria, dysphonia, and impaired mobility. Severe cases may develop respiratory compromise and are at risk for aspiration. In addition, patients may suffer from spontaneous, self-limited laryngospasm. Some features of KD resemble the early signs of amyotrophic lateral sclerosis (ALS), which can lead to misdiagnosis. A formal diagnosis of KD is confirmed by genetic testing confirming >35 CAG trinucleotide repeats in the AR gene.

A number of studies are under way examining the efficacy of various pharmacologic treatments. Currently, there is no known effective treatment for Kennedy's disease.

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

Muscle biopsy, tracheostomy, orthopaedic/trauma, abdominal.

## Type of anaesthesia

It is recommended that KD patients with severe amyotrophic weakness avoid general anaesthesia and choose local anaesthesia.

Patients with moderate to mild KD can opt for general anaesthesia. There have been reports of the use of propofol, fentanyl, and remifentanil. However, when using these drugs, it is necessary to monitor the respiration sufficiently. There are reports using inhalational anaesthesia (sevoflurane, isoflurane), but no reports using desflurane.

In KD patients, there is always a possibility that the use of muscle relaxants can prolong the action of muscle relaxants or enhance the action of muscle relaxants. When using a muscle relaxant, it is necessary to observe the degree of muscle relaxation using a muscle relaxation monitor.

Sugammadex is useful for antagonising muscle relaxants.

Succinylcholine administration should be avoided because of possible hyperkalaemia.

## Necessary additional pre-operative testing (beside standard care)

Depending on the severity of symptoms, pulmonary function testing including lung volumes and blood gas analysis may be done to evaluate the extent of pulmonary involvement [6,7].

The creatine kinase level is usually elevated (and can be used as a screening tool) [6,7].

In patients with severe weakness and reduced mobility, it may be prudent to evaluate for lower extremity thrombosis [7].

## Particular preparation for airway management

Providers should be prepared for potential laryngospasm and complications of respiratory muscle compromise. Patients may be at risk for aspiration and post-operative respiratory failure.

## Particular preparation for transfusion or administration of blood products

Not reported.



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### Particular preparation for anticoagulation

There is no evidence to support a need for anticoagulation.

### Particular precautions for positioning, transportation and mobilisation

Positioning should be carefully performed to optimise the safety of each individual patient.

### Interactions of chronic disease and anaesthesia medications

Not reported.

### Anaesthetic procedure

Reports of anaesthesia in patients with KD are few. To date, there is one case report of epidural anaesthesia and a small case series in which six patients received a total of 13 general anaesthetics. Currently, there is no definitive recommendation for either general or regional anaesthesia. Individual patient symptoms, risk factors, and surgical type should be considered when choosing the appropriate type of anaesthesia. Specific risks may include laryngospasm, possible hyperkalaemia with succinylcholine administration, increased sensitivity to non-depolarising neuromuscular blocking drugs, aspiration, and post-operative respiratory failure.

For induction of anaesthesia, propofol and thiopental were used without complication in previous reports [6,7]. Rapid sequence induction or awake intubation may be necessary, depending on the severity of bulbar symptoms or a previous history of aspiration. Patients with KD have no alterations in opioid requirements versus unaffected individuals [6,7]. However, it is necessary to pay close attention to respiratory depression.

Inhalation anaesthesia using either sevoflurane or isoflurane has been performed without complication. There are no reports of desflurane use. Since KD patients are at risk of developing laryngospasm, the use of drugs as e.g. desflurane with airway irritants should be reconsidered prior to use in KD patients. Nitrous oxide has also be used without complication in previous reports [6].

Extreme caution is required for the use of neuromuscular blockade.

Although the use of non-depolarising neuromuscular blockade is acceptable, close monitoring for degree of relaxation is required. Patients with KD have decreased levels of acetylcholine and may have an increased sensitivity to non-depolarising neuromuscular blocking drugs as well as a potential for residual blockade following reversal with anticholinesterase medications. The use of sugammadex for reversal of neuromuscular blockade may be preferred; however, there are few reports of its use in KD [7]. Although no events were reported in patients who received succinylcholine, it is likely that it should be avoided in patients with KD due to risk of hyperkalaemia [6].



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## Particular or additional monitoring

Monitoring of neuromuscular blockade is required [7].

## Possible complications

1. The acute onset of laryngospasm [8,9]
2. Increased sensitivity to non-depolarizing muscle relaxants
3. Hyperkalaemia with the use of succinylcholine [10,11]
4. Hyper-CK level
5. Post-operative respiratory failure or aspiration

## Post-operative care

The degree of post-operative monitoring depends on the surgical procedure and the pre-operative condition of the patient. Intensive care is not mandatory. However, attention should be paid to post-operative laryngospasm, aspiration, and prolongation of neuromuscular blockade. Therefore, close observation with a saturation monitor is recommended in post-operative periods, particularly in patients with greater severity of disease.

## Disease-related acute problems and effect on anaesthesia and recovery

Not reported.

## Ambulatory anaesthesia

Ambulatory anaesthesia will be acceptable if the symptoms of Kennedy syndrome are mild and surgery is at low risk. However, ambulatory anaesthesia should be carefully considered in patients with more advanced disease due to a greater risk of post-operative complications.

## Obstetrical anaesthesia

Female carriers of the mutation are usually clinically unaffected, with rare, mild symptoms. [12,13] Clinical manifestations in female carriers include hyper-CK-aemia, fasciculations, minimal weakness, or muscle cramps [14].



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