

## Anaesthesia recommendations for patients suffering from

### Segawa's dystonia

**Disease name:** Segawa's dystonia

**ICD 10:** G24.8

**Synonyms:** Segawa's disease, dopamine-responsive dystonia (DRD), hereditary progressive dystonia with diurnal fluctuation, DYT5a dystonia, GTP cyclohydrolase 1-deficient dopa-responsive dystonia

Segawa's disease (dopa-responsive dystonia, DRD) is an autosomal dominant hereditary syndrome, which was first described by Masaya Segawa and colleagues in 1970. It is caused by a mutation of the GCH 1 gene on 14q22.1-q22.2, which causes a biochemical defect in the synthesis of tetrahydrobiopterin. Due to the absence of co-factors for phenylalanine hydroxylase (PAH), the production of tyrosine is disrupted. Tyrosine is a substrate for the biological synthesis of dopamine, among others. In case of Segawa's disease, the resultant lack of dopamine in particular affects the basal ganglia. The reported prevalence of Segawa's disease is approximately 0.5/106, but it is likely to be underdiagnosed. Typical symptoms are progressive dystonia with diurnal fluctuation, frequently of the lower extremities (with a unilateral or bilatral inner rotation of a single or both feet), and primarily rapid exhaustion. In most cases, the condition responds well to low doses of L-dopa (20-300 mg) with frequently complete remission of symptoms. Even after long-term treatment, no side effects (ON-OFF phenomena, freezing) have been observed in most patients.

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Medicine in progress



Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

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

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### **Typical surgery**

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There is no curative surgery involved. However, associated surgeries may include orthopaedic procedures and caesarean section.

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### **Type of anaesthesia**

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There is no definite recommendation for either general or regional anaesthesia.

Regional or local anaesthesia can be performed without complications. There are several reports of caesarean section under spinal anaesthesia with 0,5% heavy bupivacain and fentanyl, epidural anaesthesia with 2% lidocaine and general anaesthesia with fentanyl, atracurium and isoflurane with nitrous oxide. In addition, there is a case report of general anaesthesia with sufentanil and propofol.

Succinylcholine should be avoided in wheelchair-bound patients.

L-Dopa treatment should strictly be continued, and side effects should be considered and monitored. Time without dopamine treatment should be kept to the absolutely required minimum.

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### **Necessary additional diagnostic procedures (preoperative)**

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If muscular weakness is present and regional anaesthesia is planned, neurological consultation is helpful for legal reasons.

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### **Particular preparation for airway management**

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The disease does not directly affect the airway. The preoperative assessment should be conducted according to the usual criteria.

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

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None reported, not expected.

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### **Particular preparation for anticoagulation**

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None reported. Note a possible immobilization in a wheelchair.

### **Particular precautions for positioning, transport or mobilisation**

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No specific precautions.

### **Probable interaction between anaesthetic agents and patient's long-term medication**

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Teratogenic side effects of L-dopamine are a subject of discussion.

Analogous to the treatment of Parkinson's disease, possible side effects of dopamine should be respected.

### **Anaesthesiologic procedure**

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Avoid succinylcholine in case of immobilization because of the risk of hyperkalemic cardiac arrest and rhabdomyolysis.

Opiates, propofol and local anaesthetics have been used without complication.

There are several reports of caesarean section under spinal anaesthesia with 0,5% heavy bupivacain and fentanyl, epidural anaesthesia with 2% lidocaine and general anaesthesia with fentanyl, atracurium and isoflurane with nitrous oxide.

Any kind of stress could amplify the symptoms and should be prevented by anxiolytic medication.

### **Particular or additional monitoring**

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Monitoring of the neuromuscular blockade is strictly recommended if any neuromuscular blocking agent is used. The temperature should be monitored as usual.

### **Possible complications**

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The delayed intake of dopamine could cause an amplification of the symptoms, as well as any kind of stress. A sufficient premedication is recommended to avoid stress.

### **Postoperative care**

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L-Dopamine should consistently be given on time. Due to lack of experience with this exceptionally rare disease, the patient should be monitored at intensive or intermediate care unit. Stress should be strictly prevented with benzodiazepines.

## **Information about emergency-like situations / Differential diagnostics**

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*caused by the illness to give a tool to distinguish between a side effect of the anaesthetic procedure and a manifestation of the disease*

Disease triggered emergency-like situations are not common in DRD.

### **Ambulatory anaesthesia**

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Given the lack of experience and due to legal reasons, a postoperative monitoring is important and excludes ambulatory anaesthesia in most cases.

### **Obstetrical anaesthesia**

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There are cases of exacerbation of symptoms caused by discontinuation of L-dopamine therapy (teratogenic side effects are discussed).

There are several reports of caesarean section under spinal anaesthesia with 0,5% heavy bupivacain and fentanyl, epidural anaesthesia with 2% lidocaine and general anaesthesia with fentanyl, atracurium and isoflurane with nitrous oxide.

## Literature and internet links

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*Please note that this guideline has not been reviewed by an anaesthesiologist but by two disease experts instead.*

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