orphanaesthesia

Anesthesia recommendations for

Segawa's dystonia

Disease name: Segawa's dystonia

ICD 10: G24.1, G24.8

ORPHAcode: 98808, 101150

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

Brief disease summary: Segawa's disease (dopa-responsive dystonia, DRD) is an autosomal dominant hereditary syndrome, which was first described by Masaya Segawa and colleagues in 1970 [1]. It is caused by a mutation of the GCH 1 gene on 14q22.1-q22.2 resulting in a biochemical defect in the synthesis of tetrahydrobiopterin [2]. Tetrahydrobiopterin is an essential co-factor for tyrosine hydroxylase which participates in dopamine synthesis. Reduced dopamine levels in the basal ganglia lead to dystonia symptoms that resemble Parkinson's disease. Phenotypic variability is wide, and autosomal recessive forms of the disease can occur. The reported prevalence of Segawa's disease is approximately 0.5 per million, with a female to male ratio of 2.5:1 [3].

Segawa's dystonia is characterized by a triad of symptoms:

a) progressive dystonia of childhood onset, more pronounced in the lower extremities

b) diurnal fluctuation (symptoms worsening throughout the day and markedly improving after sleep)

c) complete or nearly complete alleviation of symptoms with administration of low-dose oral levodopa [4-6].

Clinically, patients may present with gait disturbances, foot deformities, abnormal body habitus, and postural tremor involving all extremities. Cervical dystonia (torticollis) and bulbar involvement have been described [5]. Generally, there is no cognitive or intellectual impairment. In most cases, the condition responds well to low doses of levodopa (20-300 mg) often resulting in complete remission of symptoms. Treatment is lifelong, and life expectancy is not reduced.

Diagnosis may be incorrect; if uncertainty exists, the diagnosis should be re-evaluated.

Every patient is unique; individual circumstances must always guide clinical care.

Medicine is in progress; new clinical knowledge may not be yet reflected in this recommendation.

Recommendations are not rules or laws; they provide a framework to support clinical decision-making. Although this recommendation has passed a structured review process, it does not meet the formal criteria of a guideline.

Translations may not always reflect the most recent updates of the English version.

Find more information on the disease, its centers of reference and patient organizations on Orphanet: www.orpha.net

Emergency information

A	**AIRWAY / ANESTHETIC TECHNIQUE**	In cases of torticollis and dysphagia, with bulbar muscle involvement, the possibility of a difficult airway and difficult bag-mask ventilation should be anticipated, and preparation for advanced airway management should be considered.
B	**BLOOD PRODUCTS (COAGULATION)**	Not applicable, note the possibility of anticoagulation prophylaxis in cases of chronic immobilization.
C	**CIRCULATION**	Chronic therapy with levodopa is associated with hypovolemia, orthostatic hypotension, cardiac irritability and autonomic instability. Prefer direct acting vasopressors (e.g., phenylephrine).
D	**DRUGS**	No association with malignant hyperthermia. Cautious use of dopamine-blocking agents, which include phenothiazines (e.g., prochlorperazine), butyrophenones (e.g., droperidol, haloperidol), and metoclopramide, due to a theoretical risk of triggering dystonia. Propofol and ondansetron should be titrated carefully, as both agents have been associated with dystonic episodes. Avoid ketamine. Avoid halothane. Cautious use of succinylcholine (possible risk of hyperkalemia, especially in patients with chronic immobilization).
E	**EQUIPMENT**	Continuous monitoring of the neuromuscular junction Consider postoperative monitoring in an intermediate or intensive care unit.

Additional disease information

Not applicable.

Typical surgery and procedures

There are various case reports on patients with Segawa's dystonia undergoing orthopedic procedures [5,6], obstetric procedures [7-9], skin grafting [5,10], imaging techniques under sedation [5], electroconvulsive therapy [11], breast surgery [8] and hiatal hernia surgery [12] involving both adult and pediatric patients.

Type of anesthesia

Both general and regional anesthesia are feasible. Data regarding patients with Segawa syndrome undergoing surgical procedures can only be derived from case reports. There are case reports on both pediatric patients [5,6,12] and adults [5,8-11].

Regarding preoperative medication, levodopa should be continued until morning of surgery and resumed immediately after surgery as it has a short half-life (60-90 minutes) [6].

Patients with Segawa syndrome may present with cervical dystonia (torticollis), abnormal body habitus, hypersalivation, oromandibular dystonia and dysphagia [5,12]. These features, together with bulbar symptoms, increase the risk for aspiration and peri- and postoperative respiratory complications. This risk is increased in cases of concomitant scoliosis that predisposes to restrictive lung disease [10,12].

General anesthesia can be administered by total intravenous technique (TIVA) [6] or by inhalation [5,6,8-10,12].

Propofol administration either at induction or as part of TIVA needs careful titration (slow incremental dosing) [10], as it has been associated with dystonic episodes [6,12]. The majority of inhalational anesthetics (sevoflurane, isoflurane and desflurane) can be used safely, with exception of halothane. Inhalational anesthetics inhibit synaptic reuptake of dopamine leading to increased extracellular dopamine concentrations in patients with chronic levodopa use. Especially halothane, which sensitizes myocardium to catecholamines, can be arrhythmogenic.

There seem to be no special side effects associated with opioid use. Ketamine should be avoided, as on all patients on chronic levodopa therapy [9].

Succinylcholine should be avoided especially in wheelchair-bound patients, as it can precipitate hyperkalemia. Non-depolarizing muscle relaxants have been safely used. Both reversal with neostigmine-atropine [9], neostigmine-glycopyrrolate [12] and sugammadex [6] have been described without adverse reactions. As with various neuromuscular disorders, muscle relaxant reversal should be performed and patients should be extubated after achieving a Train-of-Four (TOF) ratio of 0.9, ensuring adequate tidal volume and regular respiration rate.

Most cases with regional anesthesia involve parturients, so guidelines considering regional techniques will be found on the section regarding obstetric anesthesia.

Necessary additional preoperative testing (beside standard care)

A baseline neurological consultation is preferable, especially if the patient is not under treatment with levodopa. If muscular weakness is present and regional anesthesia is planned, prior neurological consultation is helpful.

Particular preparation for airway management

Patients with Segawa syndrome can present with cervical dystonia (torticollis), abnormal body habitus, hypersalivation, oromandibular dystonia and dysphagia [5,12]. These features, together with bulbar symptoms, increase the risk for aspiration and peri- and postoperative

respiratory complications. This risk is increased in cases of concomitant scoliosis that predisposes to restrictive lung disease [10,12]. Preparation for difficult airway placement and difficult bag mask ventilation should be considered in cases with cervical, oromandibular and bulbar involvement.

Particular preparation for transfusion or administration of blood products

There is no evidence to demonstrate any specific issues related to blood product administration.

Particular preparation for anticoagulation

There is no evidence for any particular anticoagulation regime. In case of immobilization in a wheelchair, thromboprophylaxis should be discussed.

Particular precautions for positioning, transportation and mobilization

Abnormal body habitus and flexed posture in untreated patients can hinder positioning and transport [5,10]. Dystonia may involve neck (spasmodic torticollis) or face and jaw (oromandibular dystonia), which can hinder positioning for airway management [10].

Interactions of chronic disease and anesthesia medications

Teratogenic side effects of levodopa, especially during pregnancy, are a subject of discussion [7].

Chronic levodopa therapy is associated with hypovolemia, hypotension and autonomic instability [13,14]. Anesthetic agents should be administered slowly and incrementally. Fluid co-loading may be necessary. When required, sympathomimetic drugs are to be used very cautiously, as they can cause an acute rise in blood pressure. Direct-acting agents (phenylephrine) should be preferred over indirect-acting agents (ephedrine).

Drugs antagonizing dopamine within the basal ganglia such as phenothiazines, butyrophenones, benzamide and metoclopramide should be avoided.

As with propofol, ondansetron has been implicated in dystonic episodes and should therefore be given incrementally or substituted with dexamethasone.

Anesthetic procedure

Avoid succinylcholine in patients with prolonged immobilization because of the risk of hyperkalemia. Non-depolarizing muscle relaxants and reversal agents have been used without complications.

Opioids, propofol and local anesthetics have been used without complication.

Avoid ketamine and halothane.

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

For obstetric anesthesia see below on the relevant section.

Particular or additional monitoring

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

The indication for continuous arterial blood pressure measurement should be set generously, as cardiovascular instability may be more pronounced.

Possible complications

The delayed intake of dopamine could cause an amplification of the symptoms, as well as any kind of stress. Premedication is recommended to avoid stress. Bulbar involvement and dysphagia can complicate extubation and postoperative ventilation.

Postoperative care

Levodopa (L-dopa) should consistently be administered on time. Due to lack of experience with this exceptionally rare disease, the patient should be monitored in an intensive care unit or intermediate care unit [8]. Stress should be strictly prevented.

Disease-related acute problems and effect on anesthesia and recovery

Emergency-like situations, triggered by the disease, are not common in DRD.

Dysphagia in patients not receiving medication (i.e., levodopa) could lead to aspiration pneumonia.

Ambulatory anesthesia

Given the lack of experience and the possibility of bulbar muscle involvement with concomitant dysphagia, hypersalivation and cervical dystonia [5], postoperative monitoring is important and excludes ambulatory anesthesia in most cases [8].

Anesthetic involvement in patients with a history of Segawa's dystonia should be scheduled in the second trimester to allow for early planning [7].

There are several case reports of cesarean sections on parturients either under spinal anesthesia with 0.5% heavy bupivacaine and sufentanil [8], epidural anesthesia with 2% lidocaine [9], with chloroprocaine 3% [15], combined spinal epidural anesthesia (CSE) with 0.5% heavy bupivacaine and fentanyl, or general anesthesia with fentanyl, atracurium and isoflurane with nitrous oxide [9]. There is also a case report describing labor epidural analgesia with fentanyl [5].

As data can be obtained only through previously reported cases, both regional and general anesthesia for obstetric procedures appear to be feasible. In accordance with international guidelines, regional anesthesia remains the gold standard for patients with Segawa's dystonia undergoing cesarean section, as for the remaining population, because it is safer and associated with less maternal and neonatal morbidity than general anesthesia (Level 1 evidence) [16].

There is a lack of consensus regarding the continuation of levodopa in pregnancy, as it has been associated with potential effects on normal fetal development. Discontinuation of levodopa therapy can lead to symptom recurrence and exacerbation [7]. Two case reports describe patients with Segawa syndrome who continued levodopa therapy during pregnancy [8,9], whereas another report describes a patient who chose to discontinue treatment [7]. The decision on whether to discontinue therapy or not should be made after thorough explanation of benefits and potential risks by both neurologists and obstetricians on a case-by-case basis.

The effect of pregnancy on Segawa's dystonia (e.g., the possibility of exacerbation) is not known.

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

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