

## Anaesthesia recommendations for **Schwartz-Jampel syndrome**

**Disease name:** Schwartz–Jampel syndrome

**ICD 10:** G71.13

**Synonyms:** chondrodystrophic myotonia, myotonic chondrodystrophy

**Disease summary:** Schwartz–Jampel syndrome (SJS) is a rare disorder that is characterized by myotonia and skeletal abnormalities. SJS is caused by mutations in the HSPG2 gene encoding protein perlecan.

Myotonia results in a fixed facial expression with blepharophimosis, microstomia, pursed lips and mask-like faces. The typical skeletal and other abnormalities include short stature, kyphoscoliosis, joint contractures, and micrognathia. Tracheal intubation is difficult in these patients because they have micrognathia, cervical kyphoscoliosis, and a limited mouth opening.

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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 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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Typical surgeries in the SJS patients include treatment for blepharophimosis, juvenile cataract, cleft palate, and joint contractures.

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### **Type of anaesthesia**

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Regional anaesthesia (if possible) is a recommended type of anaesthesia because of the difficulties in airway management. Several reports have described successful anaesthetic managements using caudal block. Neuraxial blocks except for caudal block might be difficult because of the skeletal abnormalities.

General anaesthesia avoiding volatile anaesthetics and succinylcholine is generally used in the literature due to the fear of malignant hyperthermia. There is, however, only one case report describing intraoperative high temperature of a 17 months old girl that underwent general anaesthesia, using nitrous oxide and a nondepolarizing neuromuscular blocking drug. Recent evidence shows that the myotonias are not related to malignant hyperthermia except for hypokalaemic periodic paralysis.

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

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Since the patients with SJS have skeletal abnormalities including joint contractures, an orthopaedist should be consulted.

These patients may have skeletal deformities of chest and back in the form of small chest cavity or kyphoscoliosis. So a preoperative lung function test may be helpful in these patients.

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

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Micrognathia, cervical kyphoscoliosis, and a limited mouth opening make tracheal intubation difficult in the patients with SJS. Because of the difficulties, careful examinations of the airways are crucial in the patients with SJS.

Appropriate difficult airway equipment should be prepared in the operating room. Specific skills are required when managing difficult airway associated with limited mouth opening in paediatric patients. Optic stylets are reported to be suitable option for tracheal intubation in children with limited mouth opening. Supraglottic airway devices are other options for the airway management in the patients with SJS.

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

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Not reported. The general rules for perioperative blood management may be applied.

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### **Particular preparation for anticoagulation**

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Not reported.

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### **Particular precautions for positioning, transportation and mobilisation**

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Proper positioning with particular attention to joints is essential because of skeletal abnormalities including joint contractures

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### **Interactions of chronic disease and anaesthesia medications**

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Not reported.

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### **Anaesthetic procedure**

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Although scientific evidence denies the association between SJS and malignant hyperthermia, it might be prudent to avoid volatile anaesthetics and succinylcholine because of a reported case of thermoregulatory dysfunction in a patient with SJS.

There is one report about higher doses of muscle relaxants.

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### **Particular or additional monitoring**

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Monitoring of the neuromuscular blockade is recommended when neuromuscular blocking agents are used.

Monitoring body temperature is recommended.

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### **Possible complications**

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Thermoregulatory dysfunction during surgery has been reported in a patient with SJS.

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### **Post-operative care**

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Respiratory monitors should be used postoperatively. In some cases, intensive care medicine is necessary.

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

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Thermoregulatory dysfunction during surgery has been reported in a patient with SJS.

**Ambulatory anaesthesia**

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Not reported.

**Obstetrical anaesthesia**

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Not reported.

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**Date last modified: March 2018**

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**Disclosure(s)** The author has no financial or other competing interest to disclose. This recommendation was unfunded.

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**Disclosure(s)** The reviewers have no financial or other competing interest to disclose.

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