orphanaesthesia

Anesthesia 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 [1].

Myotonia results in a fixed facial expression with blepharophimosis, microstomia, pursed lips and mask-like facies [2]. 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.

Medicine is in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnosis is wrong

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

A	**AIRWAY / ANESTHETIC TECHNIQUE**	Micrognathia, cervical kyphoscoliosis, and a limited mouth opening make tracheal intubation difficult in the patients with SJS. Regional anesthesia (if possible) is a recommended type of anesthesia because of the difficulties in airway management.
B	**BLOOD PRODUCTS (COAGULATION)**	Not reported.
C	**CIRCULATION**	Not reported.
D	**DRUGS**	Although scientific evidence denies the association between SJS and malignant hyperthermia, it might be prudent to avoid volatile anesthetics and succinylcholine because of a reported case of thermoregulatory dysfunction in a patient with SJS.
E	**EQUIPMENT**	No specific recommendations are given.

Typical surgery and procedures

Typical surgeries in the SJS patients include treatment for blepharophimosis, juvenile cataract, cleft palate, and joint contractures [3,4].

Type of anesthesia

Regional anesthesia (if possible) is a recommended type of anesthesia because of the difficulties in airway management [3,4]. Several reports have described successful anesthetic managements using caudal block. Neuraxial blocks except for caudal block might be difficult because of the skeletal abnormalities.

General anesthesia avoiding volatile anesthetics and succinylcholine is generally used in the literature due to the fear of malignant hyperthermia. However, there is no report showing that SJS is related to malignant hyperthermia [5].

Necessary additional pre-operative testing (beside standard care)

Since the patients with SJS have skeletal abnormalities including joint contractures [6]. The patients should be consulted to orthopedic doctors.

Particular preparation for airway management

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 [3,4].

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 pediatric patients. Optic stylets are reported to be suitable option for tracheal intubation in children with limited mouth opening [4]. Supraglottic airway devices are other options for the airway management in the patients with SJS [7].

Particular preparation for transfusion or administration of blood products

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

Particular preparation for anticoagulation

Not reported.

Particular precautions for positioning, transportation and mobilization

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

Interactions of chronic disease and anesthesia medications

Not reported.

Anesthetic procedure

Although scientific evidence denies the association between SJS and malignant hyperthermia, it might be prudent to avoid volatile anesthetics and succinylcholine because of a reported case of thermoregulatory dysfunction in a patient with SJS [5,8].

Particular or additional monitoring

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

Monitoring body temperature is recommended.

Possible complications

Thermoregulatory dysfunction during surgery has been reported in a patient with SJS [8].

Post-operative care

Respiratory monitors should be used postoperatively.

Disease-related acute problems and effect on anesthesia and recovery

Thermoregulatory dysfunction during surgery has been reported in a patient with SJS [8]. When malignant hyperthermia is suspected dantrolene should be given.

Ambulatory anesthesia

Not reported.

Obstetrical anesthesia

Not reported.

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Date last modified: **October 2025**

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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 The reviewers have no financial or other competing interest to disclose.

Update and revision (October 2025)

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