

# A successful anesthetic approach in a patient with Schwartz–Jampel syndrome

### ABSTRACT

Schwartz–Jampel syndrome (SJS) is a rare genetic condition that is characterized by several musculoskeletal abnormalities, such as myotonia, joint contractures, and facial dysmorphisms. Patients with this syndrome can present an anesthetic challenge, due to an increased risk of developing malignant hyperthermia (MH) and the possibility of encountering a difficult airway. Several precautions must be taken when general anesthesia is required, such as the avoidance of potential triggers for MH, continuous core temperature, and end-tidal CO<sub>2</sub> monitoring, assuring that dantrolene is readily available. It is also fundamental to prepare for a possible difficult airway, guaranteeing that difficult airway devices are available. We describe the anesthetic management of a 14-year-old boy diagnosed with SJS who was scheduled for multiple dental extractions and was successfully anesthetized with our approach.

**Key words:** Difficult airway; malignant hyperthermia; Schwartz–Jampel syndrome

### Introduction

Schwartz–Jampel syndrome (SJS) is a rare autosomal recessive disease first described in 1962.<sup>[1]</sup> The prevalence of SJS is <1:1,000,000 and about 100 cases have been reported worldwide.<sup>[2]</sup> Patients develop myotonia, with continuous tensing of the skeletal muscles,<sup>[3]</sup> causing stiffness that interferes with eating, sitting, and walking. Contraction of the facial muscles leads to a “mask-like” expression with blepharophimosis and pursed lips. Patients also present with micrognathia, microstomia, jaw muscle rigidity, and thermoregulatory disturbance, with an increased risk of malignant hyperthermia (MH) and having a difficult airway.<sup>[3-8]</sup> We report the anesthetic management of a youngster with SJS proposed to multiple dental extractions and alveoloplasty.

### Case Report

A 14-year-old male, American Society of Anesthesiologists (ASA) status III, with SJS and epilepsy, was scheduled for multiple dental extractions and alveoloplasty under general anesthesia.

He had no history of previous anesthetics or known allergies.

His usual medication was carbamazepine 200 mg BID for his epilepsy, which was under control. Family history revealed that his mother and brother were diagnosed with the same condition.

Physical examination showed a patient with a weight of 35 kg, a height of 130 cm, and a short stature compared with other children with the same age. He had an unusual facies, muscle

This is an open access article distributed under the terms of the Creative Commons Attribution-NonCommercial-ShareAlike 3.0 License, which allows others to remix, tweak, and build upon the work non-commercially, as long as the author is credited and the new creations are licensed under the identical terms.

**For reprints contact:** reprints@medknow.com

**How to cite this article:** de Oliveira Camacho FC, Lopes Amaral TM, de Barros Mourão JI. A successful anesthetic approach in a patient with Schwartz–Jampel syndrome. Saudi J Anaesth 2018;12:128-30.

Access this article online	
<b>Website:</b> www.saudija.org	<b>Quick Response Code</b> 
<b>DOI:</b> 10.4103/sja.SJA_393_17	

**FERNANDO CALADO DE OLIVEIRA CAMACHO, TÂNIA MARINA LOPES AMARAL, JOANA IRENE DE BARROS MOURÃO**

Department of Anesthesiology, São João Hospital Centre, Porto, Portugal

**Address for correspondence:** Dr. Fernando Calado de Oliveira Camacho, Department of Anesthesiology, São João Hospital Centre, Alameda Professor Hernâni Monteiro, 4200-319 - Porto, Portugal. E-mail: fcamacho00@gmail.com

stiffness, and rigidity. Vital signs and remaining physical examination were normal.

The patient was scheduled as the first one of the day. Before his arrival to the operating theater, the circuit of the anesthetic workstation was flushed and we guaranteed that dantrolene and cooling devices were available. Difficult airway devices were readily accessible should any problem with the endotracheal intubation arise.

Before induction, a peripheral venous catheter was placed. No premedication was administered. He was monitored under the ASA standards plus core temperature measurement and bispectral index monitoring (BIS). After 3 min of preoxygenation with oxygen (O<sub>2</sub>) at 100%, induction of anesthesia was accomplished with a propofol bolus at a 2 mg/kg dosage. We had no difficulties with mask ventilation. Direct laryngoscopy was attempted. His Cormack–Lehane grade was III, and we performed two attempts of tracheal intubation with a Macintosh Blade No. 3 with no success. We decided to interrupt laryngoscopy and ventilate the child with the facial mask. Afterward, a new attempt with a C-MAC® videolaryngoscope was performed. We visualized 60% of the glottis, but inserting the endotracheal tube was impossible. We decided to introduce a wireframe laryngeal mask (LM) that allowed an adequate ventilation. After discussing the case with the surgical team, we carried out the procedure using the frame LM for airway (LMA) management with pressure-controlled ventilation targeted to 4–6 mL/kg and a respiratory frequency adjusted to achieve an end-tidal CO<sub>2</sub> (EtCO<sub>2</sub>) of 35–40 mmHg. Maintenance of anesthesia was accomplished with a propofol infusion at 5 mg/kg/h and local anesthetic infiltration. Fifteen minutes before the end, paracetamol 425 mg intravenously was administered to provide postoperative analgesia. The procedure lasted 2 h and underwent without any clinical events. The patient remained hemodynamically stable, with no significant fluctuations in blood pressure, pulse, or peripheral O<sub>2</sub> saturation. His core temperature remained within the normal range. Blood loss was minimal. When the surgery ended, we stopped the propofol infusion. As soon as he regained consciousness, airway reflexes, and spontaneous breathing, with adequate lung volumes per minute, the LMA was removed. The patient was transferred to the postanesthetic care unit, with normal vital signs and painless, where he remained for 24 h under observation. In the next day, he was discharged home without any anesthetic or surgical complications.

## Discussion

SJS is a rare condition, with about 100 cases described in the medical literature.<sup>[2]</sup> It is characterized by a noticeable

clinical heterogeneity, and it can be divided into three types: IA, IB, and II.<sup>[6,8,9]</sup>

There are various issues concerning the anesthetic approach in SJS. The first one is the possibility of encountering a difficult airway. It is fundamental to assure a careful difficult airway plan, guaranteeing that the appropriate devices are available. The second issue that may arise is the possibility of a thermoregulatory dysfunction, with the patient at risk of developing MH. Therefore, we avoided using any potential triggers, such as volatile anesthetics and depolarizing neuromuscular blockers (succinylcholine).<sup>[10]</sup> We guaranteed that no volatile anesthetics were administered by flushing the workstation according to the manufacturer's recommendations. Core temperature and EtCO<sub>2</sub> monitoring are essential to evaluate for any signs of increased metabolism, and we assured that such information was available throughout the perioperative period. Dantrolene is the primary drug to use should any suspicion of MH arise,<sup>[10]</sup> so we made sure that it was readily available at the operating theater. Finally, we took additional precautions with the ventilation in this case, as these patients can have skeletal deformities, muscle stiffness, and weakness that can impair the lung function. They may have limited chest wall expansion, with reduced vital capacity and decreased chest wall compliance, all of which can compromise the adequacy of the ventilation.<sup>[3,5,9]</sup>

As a result of the potential risks of anesthetizing and operating on a patient with SJS, it is necessary to keep a close vigilance for at least 24 h to be certain that no postanesthetic complications arise. We secured the postoperative care in an intermediate care unit.

Our patient was safely anesthetized with our anesthetic approach. The authors intend to underline that it is essential to have a detailed knowledge of the anesthetic concerns and complications that can develop throughout the perioperative period, and how to prevent and treat them, as this is a rare syndrome that most anesthesiologists will not encounter during their career. This case shows that an LM could also be an alternative for a difficult airway in SJS.

## Financial support and sponsorship

Nil.

## Conflicts of interest

There are no conflicts of interest.

## References

1. Schwartz O, Jampel RS. Congenital blepharophimosis associated with a unique generalized myopathy. *Arch Ophthalmol* 1962;68:52-7.

2. Chandra SR, Issac TG, Gayathri N, Shivaram S. Schwartz–Jampel syndrome. *J Pediatr Neurosci* 2015;10:169-71.
3. Berardinelli A, Ferrari Ginevra O, Lanzi G. The Schwartz-Jampel syndrome. A minireview. *Basic Appl Myol* 1997;7:363-7.
4. Ray S, Rubin AP. Anaesthesia in a child with Schwartz-Jampel syndrome. *Anaesthesia* 1994;49:600-2.
5. Ho NC, Sandusky S, Madike V, Francomano CA, Dalakas MC. Clinico-pathogenetic findings and management of chondrodystrophic myotonia (Schwartz-Jampel syndrome): A case report. *BMC Neurol* 2003;3:3.
6. Basiri K, Fatehi F, Katirji B. The Schwartz–Jampel syndrome: Case report and review of literature. *Adv Biomed Res* 2015;4:163.
7. Mukaihara K, Godai K, Yamada T, Hasegawa-Moriyama M, Kanmura Y. Successful airway management using a MultiViewScope handle with a stylet scope in a patient with Schwartz-Jampel syndrome. *JA Clin Rep* 2016;2:36.
8. Abdel-Aziz M, Azab NA. A case of Schwartz-Jampel syndrome with cleft palate. *Int J Pediatr Otorhinolaryngol* 2009;73:1601-3.
9. Ewees BE, El-Fawy DM. Anesthesia for herniotomy in Schwartz-Jampel syndrome. *Ain Shams J Anaesthesiol* 2015;8:455-7.
10. Safety Committee of Japanese Society of Anesthesiologists. JSA guideline for the management of malignant hyperthermia crisis 2016. *J Anesth* 2017;31:307-17.