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Case report

A Maxillofacial Surgical Approach to Pediatric Schwartz-Jampel Syndrome: A Study of Jordanian Children

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Abstract
Schwartz-Jampel syndrome (SJS) is a rare neuromuscular disorder with autosomal recessive inheritance, characterized by defective muscle stiffness (myotonia) and cartilage development. The clinical manifestations of this syndrome include permanent generalized myotonia and distinctive facial features such as blepharophimosis, a puckered chin, pursed lips, and a fixed facial expression. Skeletal abnormalities include short stature, skeletal dysplasia, and joint contractures. Objective: We report four Jordanian children with SJS, who were referred to the maxillofacial surgery section for the management of mentalis muscle overactivity after carbamazepine medication and physiotherapy were not successful. The patients underwent Botulinum toxin A injections in the mentalis muscle under general anesthesia. One patient also underwent additional surgical intervention in the form of mentalis muscle myotomy. The laryngeal mask airway technique was used for general anesthesia, and precautions for malignant hyperthermia were taken. The patients were reviewed for up to eight months with satisfactory results. Mentalis muscle Botulinum toxin A injections for pediatric SJS patients under general anesthesia have satisfactory outcomes in controlling the muscle overactivity. The procedure can be performed safely with modified anesthetic techniques, provided precautions are taken for malignant hyperthermia.
Keywords: *Schwartz-Jampel syndrome, Botulinum toxin A, Myotonia, Mentalis muscle overactivity, Case report.*

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Introduction

Schwartz-Jampel syndrome (SJS) is a rare disorder that affects fewer than one in a million people. It is characterized by myotonia (prolonged muscle contraction) and chondrodysplasia that cause bone deformities. This syndrome is caused by mutations in the heparan sulfate proteoglycan 2 (HSPG2) gene, which encodes the perlecan protein regulating muscle contraction and chondrogenesis [1-6]. Perlecan is a large proteoglycan deposited in matrices of muscles, cartilage, and bone marrow [4].

Myotonia is considered the main morbidity in SJS since it interferes with eating, speech, sitting, and other physical activities. Sustained facial muscle contractions lead to the development of a mask-like face and limited mouth opening. [1,2,4-6].

Mentalis muscle overactivity can result in mandibular retrognathia with skeletal class II malocclusion, in addition to crowding of the lower anterior teeth. [1,6] which might necessitate future major surgical correction (e.g., distraction osteogenesis of the mandible) to address this skeletal discrepancy if left untreated. This study reports four cases of SJS pediatric patients with mentalis muscle overactivity being treated with Botulinum toxin A injections with good outcomes.

Case Report

Four patients diagnosed at Al-Bashir Hospital in Amman, Jordan, with SJS were referred between the years 2019 and 2023 from the pediatric neurology section to the maxillofacial surgery section for evaluation of the mentalis muscle overactivity. All patients were of Middle Eastern ethnicity and descendants of unrelated families. The biographic data of the patients are summarized in Table 1.

Table 1. The biographic data of the patients.

Case	Gender	Age (years)	Procedure performed
1	Male	17	Botulinum toxin A injection
2	Male	12	Botulinum toxin A injection
3	Male	15	Botulinum toxin A injection
4	Female	11	Botulinum toxin A injection and Myotomy

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General examination showed that the patients exhibited an abnormal gait and relatively short stature. Additionally, there was evident general myotonia, facial muscle hyperactivity, allergic rhinitis, and breathing difficulty due to neck scoliosis, retrognathia, and restricted mouth opening.
The patients' medical history revealed that, in an attempt to control the motor overactivity, the patients were on a trial medication of carbamazepine (Tegretol®) 15mg/kg daily in divided doses prescribed by the neurologists to manage the myotonia. As a result, the patients developed mood changes with no muscular improvement; subsequently, intensive physiotherapy was initiated, but with no satisfactory result.
Maxillofacial and radiographic examination (orthopantogram and skull lateral view) revealed common characteristics of an underdeveloped face, limited mouth opening, and retrognathic mandibles and potential for impacted teeth (Figure 1-A, B, C). The mentalis muscle overactivity and pursed lips were evident clinically. All patients were scheduled for surgery under general anesthesia and were admitted on the same operation day for observation of the possible development of malignant hyperthermia.

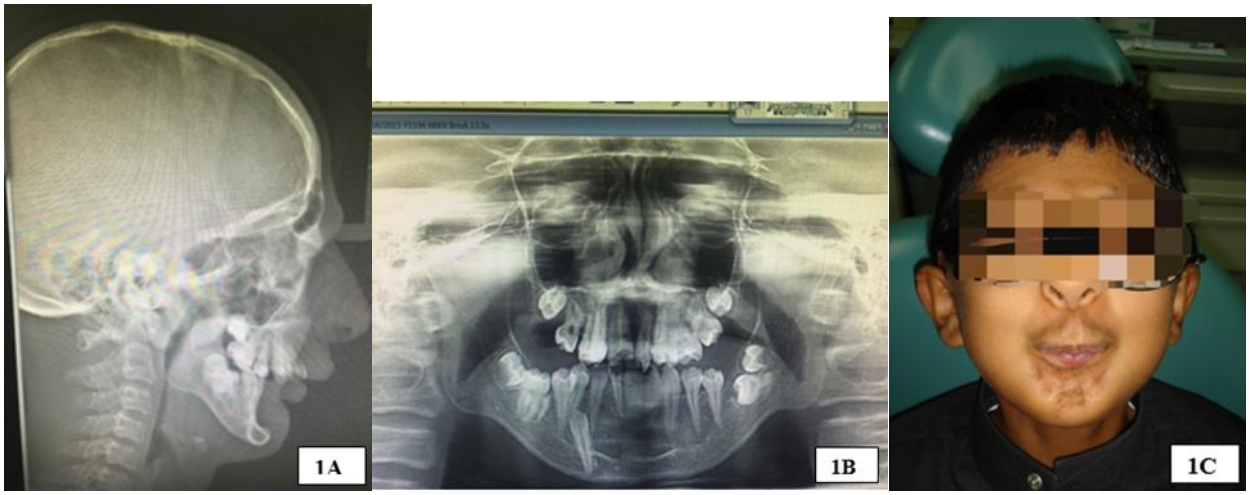

Figure 1: A: Lateral cephalometric view of one of the patients, B: panoramic view, C: facial features before treatment.

The patients were seated in a reverse-Trendelenburg position on the operating table, and the mentalis muscle was marked with a sterile surgical marking pen (Figure 2). Botulinum toxin A injection (Botox®, Botulinum toxin type A, Allergan Inc., Irvine, California, USA) was administered to all four patients under general anesthesia. One vial of Botox® (containing 100 units of Botulinum toxin A) was diluted with 4 mL of 0.9% normal saline, resulting in a concentration of 25 units per mL. Using an insulin syringe (1 mL volume/30-gauge needle), a total of 12.5 units (0.5 mL) was injected and distributed equally bilaterally in the lower portion of the mentalis muscle to avoid asymmetric chin expressions. Care was taken to avoid injecting into the upper portion of the mentalis muscle, as this would paralyze the neighboring orbicularis oris muscle, leading to drooling afterward.

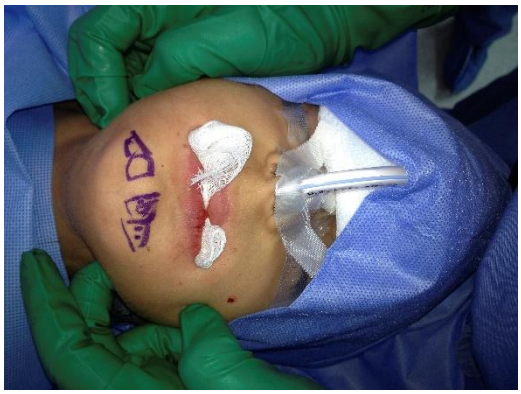

Figure 2: Marking the injection sites of Botulinum toxin A.

Intraoperative and immediate postoperative recovery were uneventful, and the patients remained stable. Postoperative instructions were given to avoid massaging the area, and appropriate analgesics were prescribed (paracetamol 10 mg/kg/dose, 4 times daily as needed for 2 days). The patients were followed up

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in the surgical ward and found to be stable, hence, they were discharged the following day. Regular follow-ups in the outpatient clinic for 5 to 8 months were conducted, and the results were satisfactory with no mentalis muscle overactivity. However, patients unfortunately failed to maintain long-term follow-up.
Three cases responded well to the Botulinum toxin A injection in the chin with marked improvement in the pursed-lip sign (Figure 3-A & B). The fourth case was unresponsive to Botulinum toxin A injection; therefore, surgical intervention in the form of mentalis muscle myotomy was performed to weaken the target overactive muscle.

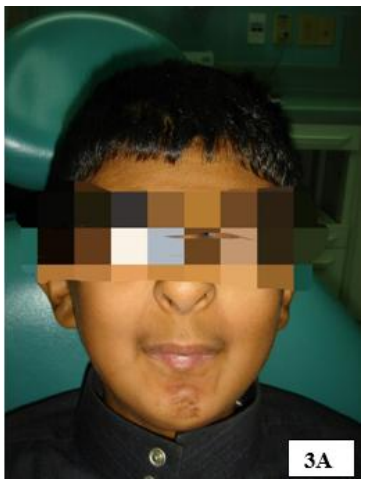

Figure 3-A & B: Photos after mentalis Botulinum toxin A injection.

The patient was nasally intubated, and a pharyngeal pack was inserted. One ampoule (1.8 mL) of local anesthesia (2% lidocaine with 1:100,000 epinephrine) was infiltrated at the operative sites. The incision was performed intraorally, to avoid skin scarring. A small vertical incision was made about 10 mm below the mucogingival junction at the canine area bilaterally. An anteriorly placed incision conserves both attached and unattached oral tissue for closure. The classical long horizontal incision was avoided, since it may result in more untoward fibrosis-related morbidity later on. Once through the mucosa, the mental nerve branches are unlikely to be encountered in this site; however, if encountered, they should be gently protected and reflected away from the surgical field. The dissection was continued sharply, and the underlying symphyseal bone was exposed in a sub-periosteal manner. By inserting a suitable periosteal elevator, tunneling was performed to detach the inferior portion of the mentalis muscle from its bony attachment, resulting in a favorable outcome of soft tissue drape ptosis. No dissection was required superiorly to preserve the superior portion of the mentalis muscle. Soft tissue closure was accomplished in a one-layer fashion with the re-approximation of only the mucosa using resorbable sutures (Coated VICRYL RAPIDE 4/0). No extraoral dressing on the chin was required, as this prevented the redraping of the overlying soft tissue. At the end of the procedure, the pharyngeal pack was removed.
A definitive conclusion regarding the long-term effects on the improvement of lower anterior teeth crowding and skeletal class II malocclusion could not be drawn, as jaw growth had almost entirely ceased by the time the patients presented for treatment.

Discussion

In 1962, Oscar Schwartz and his colleague Robert Jampel described two cases with blepharophimosis, facial dysmorphism features, generalized myopathy, and joint deformities. Since then, the term Schwartz-Jampel syndrome (also known as myotonic chondrodystrophy syndrome) has been used [7]. Up to the year 2022, only 150 cases of SJS had been reported in the literature worldwide [8].
The skeletal abnormalities include short stature, scoliosis, skeletal dysplasia, and joint contractures. [9] The considerable muscle weakness, in addition to contracture formation, limb stiffness, and skeletal abnormalities, causes the abnormal waddling gait. [1,9,10]. Patients with SJS have distinctive facial features including ptosis, a fixed facial expression, pursed lips, a puckered chin, microstomia, limited mouth opening, microretrognathia, bimaxillary hypoplasia, and a high arched palate [1,2,6,10]. Dental findings include teeth crowding, posterior crossbite, and missing, impacted, or supernumerary teeth [1].
In SJS patients, myotonia is most prominent in the facial muscles, where the contracture of the perioral muscles results in pursed lips and micrognathia [11]. Upon reviewing the literature, many SJS patients exhibit airway issues, including pediatric obstructive sleep apnea [1], laryngeal hypoplasia, dyspnea, which can complicate the endotracheal intubation if needed [10]. These airway difficulties, along with the risk of malignant hyperthermia, complicate anaesthetic management and necessitate a careful approach. [2,9,10]

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Managing children with SJS requires a multidisciplinary team approach, including a neurologist, geneticist, physical therapist, orthopedic surgeon, ophthalmologist, psychologist, occupational therapist, and maxillofacial surgeon [2,11].

Many studies have shown that myotonia in SJS cases may respond to sodium channel blockers like carbamazepine and procainamide, with better outcomes when treatment is initiated early in childhood. [2,6,11]. However, carbamazepine therapy was not effective in controlling the myotonia in our patient group. Physical therapy is crucial to prevent contracture formation and fixed skeletal deformities. Non-pharmacologic treatments such as warming, massage, and stretching have also been used in conjunction with medications [2,12].

Botulinum toxin A is a neurotoxin that effectively treats conditions involving excessive muscle activity by temporarily paralyzing the affected muscles through blocking the release of acetylcholine from the motor nerve endings [13]. It is widely considered the primary therapy for facial muscle dystonia. [5] Botulinum toxin A injection into the mentalis muscle is typically helpful [2,9,13]. Although rarely, surgical intervention in the form of mentalis muscle myotomy is needed. This procedure was performed on our patients under general anesthesia in accordance with our hospital protocols and the guidance of the anesthesiology team, due to the potential pain associated with multiple injections. In addition, pediatric and pre-adolescent patients, especially those diagnosed with SJS, may experience significant anxiety and fear, which can hinder their ability to cooperate and communicate effectively. Additionally, these procedures typically involve precise site site-specific, symmetrical multiple injections in sensitive, complex anatomical areas near the oral cavity. The administration of general anesthesia would promote patient immobility and reduce discomfort during the procedure, thereby optimizing the overall outcomes.

Clinical results of Botulinum toxin A injections into the mentalis muscle vary between studies. Aburahma *et al.* [9] reported limited cosmetic and functional improvement in lip pursing and mouth opening in two out of four patients, whereas Bandeira *et al.* [5] reported a significant clinical improvement in all their patients. The latter study also noted variability in results with high and low doses of Botulinum toxin A. Determining the appropriate dose and frequency of Botulinum toxin A injections remains debatable due to individual patient differences, the severity of myotonia, and the location of the muscle involvement. Apparently, dosage intervals should be individually tailored to achieve symptom control while minimizing side effects [2,5,9].

Although there is no evidence that surgical operations might induce anesthetic risk for SJS patients, we believe that overnight recovery for post-surgical interventions performed under general anesthesia is a wise choice. Furthermore, consultation with the primary neurologist is essential to discuss the precautions that need to be addressed. Despite the controversy among studies about the occurrence of malignant hyperthermia [2,9,10,14], precautions for malignant hyperthermia were considered intraoperatively and postoperatively by closely monitoring vital signs during the one-day hospital stay following surgery.

Reviewing the literature revealed only a few reports of maxillofacial surgical interventions, particularly the use of Botulinum toxin A muscular injection in patients with SJS, with variable outcomes [2,5,9,13,15]. This report can add to the knowledge and emphasize the usefulness of this procedure in the management of SJS.

Conclusion

Overall, mentalis muscle Botulinum toxin A injection under general anesthesia for SJS patients appears to be an effective option within the comprehensive treatment plan for this rare disease.

Due to the rarity of this syndrome, a standardized treatment plan is still lacking and is awaiting consensus among surgeons.

Declaration

The authors declare no conflicts of interest, of any kind, with this case report.

Ethical Approval

Written informed consent has been obtained from the patient's parents/guardians for the publication.

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