Case Report

Orbicularis Oculi Myectomy as a Treatment for Blepharospasm in a Case of Schwartz Jampel Syndrome

Bahram Eshraghi[1], MD; Mahla Shadravan[2], MD; Elham Aalami[2], MD; Elias Khalili Pour[2], MD

[1]Eye Research Center, Farabi Eye Hospital, Tehran University of Medical Sciences, Tehran, Iran
[2]Department of Ophthalmic Plastic and Reconstructive Surgery, Eye Research Center, Farabi Eye Hospital, Tehran University of Medical Sciences, Tehran, Iran

Abstract

Purpose: To describe a patient with Schwartz Jampel vel Aberfeld syndrome (SJS) who underwent orbicularis oculi myectomy as a treatment for blepharospasm.
Case Report: A 4-year-old child with SJS did not respond to an injection of a single dose of botulinum toxin after one month, so orbicularis myectomy was then performed under general anesthesia. During the procedure, orbicularis vermiform movements were a useful guide for the extent of myectomy that the patient needed. He responded very well to this procedure and experienced significant relief of blepharospasm documented in follow-up visits for up to 6 months.
Conclusion: Blepharospasm in patients with SJS can be treated with orbicularis oculi myectomy as a good functional method with faster and durable response in comparison to botulinum toxin injection.

Keywords: Blepharospasm; Orbicularis Oculi Myectomy; Schwartz Jampel Syndrome

J Ophthalmic Vis Res 2016; 11 (3): 329-332.

INTRODUCTION

Schwartz Jampel vel Aberfeld syndrome (SJS) is a rare congenital disorder with autosomal recessive inheritance in which the affected patients have myotonic myopathy, osteoarticular abnormalities, skeletal dysplasia, and ocular manifestations. This syndrome has 2 clinical types, Type I and Type II.

Type I has 2 subtypes: IA and IB. Type IA is the classic and most common form, which is diagnosed in childhood with myotonia and bone dysplasia that are not severe.[1,2] Strong contractions of the orbicularis oculi muscle causes maldevelopment of the eyelids. The other manifestations are joint contractures and generalized muscular hypertrophy.[3] Type IB manifests at birth with more severe bone dysplasia and maldevelopment of the vertebral column, but myotonia may develop later.[4] The long-term survival rate with this subtype is good. Type II is the most severe type and is recognized at birth, and the mortality rate in this group is higher due to the severity of symptoms.[2]

Blepharospasm is one of the most bothersome symptoms that needs to be treated as soon as possible, especially in children. Multiple methods of management for blepharospasm of this syndrome have been introduced, such as botulinum toxin injection into the orbicularis muscle and orbicularis oculi muscle myectomy. Intolerance to anesthetic drugs and secondary complications such as malignant

Correspondence to:
Elias Khalili Pour, MD. Eye Research Center, Farabi Eye Hospital, Qazvin Square, Tehran 13366, Iran.
E-mail: ekhalilipour@gmail.com

Received: 07-09-2014 Accepted: 08-02-2015

Access this article online

Quick Response Code:

Website: www.jovr.org

DOI: 10.4103/2008-322X.188401

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: Eshraghi B, Shadravan M, Aalami E, Pour EK. Orbicularis oculi myectomy as a treatment for blepharospasm in a case of Schwartz Jampel syndrome. J Ophthalmic Vis Res 2016;11:329-32.

hyperthermia place these patients in a high-risk group for general anesthesia.

In this report, we present a case with SJS, treated with orbicularis myectomy under general anesthesia to relieve blepharospasm, who was followed for 6 months.

CASE REPORT

A 4-year-old boy was referred to the Ophthalmic Plastic and Reconstructive Surgery Department of Farabi Eye Hospital, Tehran University of Medical Sciences, Tehran, Iran, because of severe blepharospasm and secondary functional blindness. He had facial dysmorphism at birth and showed developmental delay. The patient was unable to walk until he was 2.5 years old and had delayed speech development as well as a hypertrichotic and mask-like face [Figures 1 and 2]. His blepharospasm began in infancy and increased progressively.

Considering these clinical aspects, SJS was the suspected diagnosis, and we referred him to the neurology department for confirmation of SJS. A neurologist diagnosed him with SJS Type IB. Due to severe blepharospasm, cycloplegic refraction was impossible and the vertical palpebral fissure in the right and left eyes were 2 and 1 mm, respectively, and both eyes had positive Bell's phenomenon. The cornea and lens were clear, and the fundoscopic exam was unremarkable.

Due to intolerance to general anesthetic drugs, botulinum toxin injection for the treatment of severe blepharospasm was tried by another ophthalmologist who injected 20 units of botulinum toxin (Dysport, Speywood Pharmaceuticals Ltd., Maidenhead, UK) into the orbicularis muscles on each side. After 1 month, no relief of blepharospasm was observed in both eyes. Because of the severity of the blepharospasm and resulting significant visual disability, we decided to try bilateral orbicularis oculi myectomy. Under general anesthesia, an upper lid crease incision was made and in the lower lid, a subciliary incision was made. Pretarsal and preseptal orbicularis muscle exposure and myectomy were performed; during the procedure, vermiform contractions of the orbicularis muscle were apparent and after the procedure, these movements were decreased but had not disappeared completely. These movements were a useful guide for the main part of the myectomy. We also did a procerus muscle myectomy, but the corrugator was left intact and the levator muscle was tucked.

On the first postoperation day, his blepharospasm had decreased and he had mild lagophthalmos (OD, 2 mm; OS, 3 mm). Frequent lubrication was prescribed and 1 month later on the second follow-up visit, the patient was able to open his eyes without using his fingers and his blepharospasm had significantly diminished [Figures 3 and 4]. His cycloplegic refraction showed hyperopic astigmatism in both eyes (OD, +3.75 – 3.75 × 180; OS, +3.75 – 3.50 × 170). The vertical palpebral fissure in the right and left eyes was 7 and 8 mm, respectively.

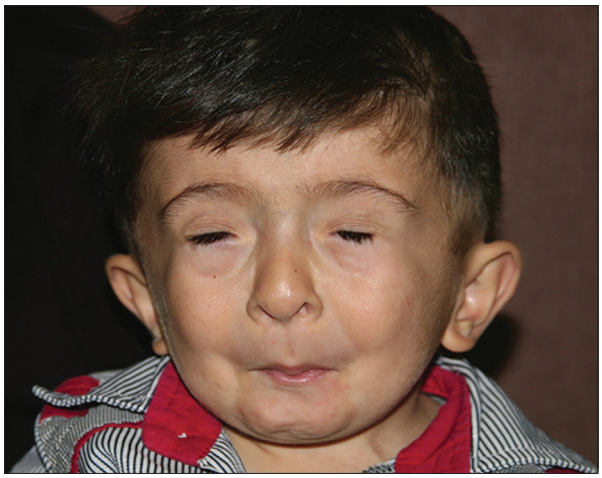

Figure 1. Mask-like face and severe bilateral blepharospasm.

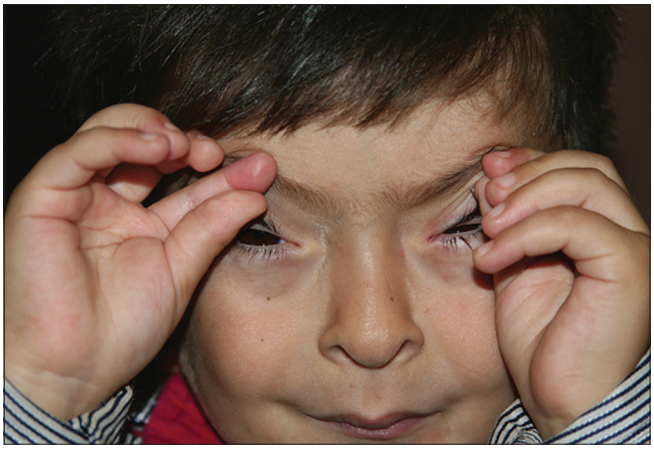

Figure 2. Preoperatively, due to severe blepharospasm, the patient was not able to open his eyes without using his fingers.

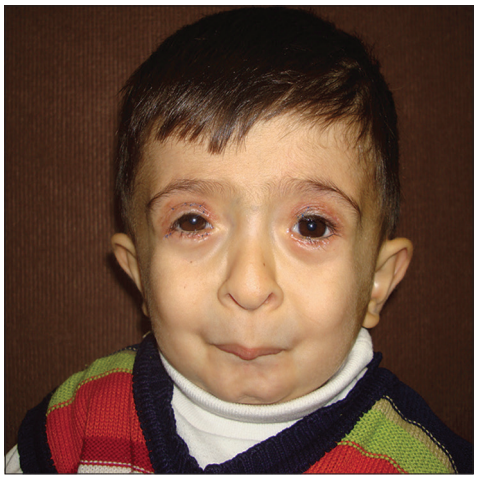

Figure 3. In the second follow-up visit, a month after surgery, the patient was able to open his eyes without using his fingers and his blepharospasm had diminished significantly.

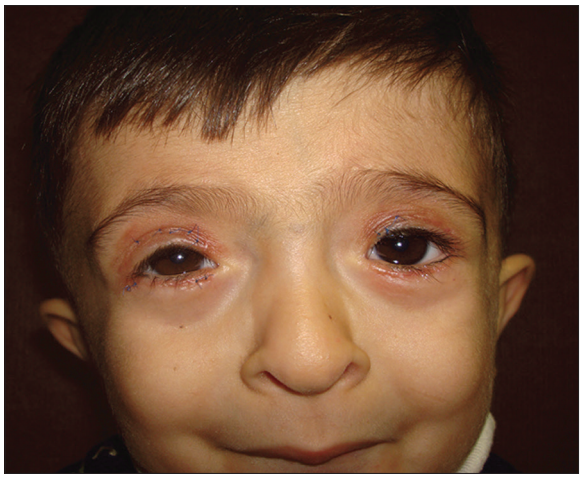

Figure 4. Near view of reduced blepharospasm 1 month postoperatively.

He had mild lagophthalmos in both eyes (OD: 1 mm and OS: 2-3 mm) but he did not have corneal exposure keratopathy. He had hyperopic astigmatism and full cycloplegic refraction was prescribed for him. In monthly follow-ups up to month 6, he had mild blepharospasm that minimally interfered in his daily life and also mild lagophthalmos without corneal exposure similar to the findings 1 month after surgery.

DISCUSSION

According to the history of our patient given by his parents, clinical symptoms, and his developmental delay (e.g., delay in talking and walking), and consultation with a neurologist, he was considered to have Type IB SJS.

In the diagnosis of SJS, clinical manifestations should be documented by electromyography for myotonia and X-rays for the chondrodysplasia.[1] Electromyography of our patient reported by the neurologist demonstrated continuous electrical activity in the muscle fibers. Ocular manifestations of SJS include microcornea, microphthalmia, myopia, juvenile cataract, hypertrichosis, maldevelopment of the eyelid, and repeated contractions of the orbicularis oculi muscle that results in blepharospasm, blepharophimosis, and ptosis in these patients.[3,5] Our patient had most of these manifestations but did not have microcornea, myopia, or juvenile cataract. He had blepharophimosis and was hyperopic, not myopic. His blepharospasm progressively worsened after birth.

In a literature review on the treatment of blepharospasm in patients with SJS, a few reports are available of surgical and nonsurgical treatment methods such as botulinum toxin injection; however, the efficacy and comparison of these 2 treatment modalities have not been documented with any controlled trial. Surgical treatment in these patients has some problems. They are difficult to intubate due to jaw muscle rigidity and micrognathia, and they also prone to malignant hyperthermia while under general anesthesia.[6]

Eikermann et al demonstrated that in this syndrome higher amounts of muscle relaxants are required in comparison to unaffected patients. Our patient did not respond to 20 units bilateral injection of botulinum toxin in the orbicularis muscle as initial nonsurgical modality for treatment of blepharospasm. The injection was performed by an ophthalmologist after considering the high risk of general anesthesia; some previous reports have shown that botulinum toxin does not have any effect on blepharospasm in SJS.[3,7]

Surgical treatment for blepharospasm and blepharophimosis in these patients includes orbicularis myectomy, levator muscle advancement, lateral canthopexy, and resection of the lower lid retractors.[8] Lucci et al suggested that the treatment of choice for lid problems in SJS is orbicularis myectomy, lateral canthopexy, and repair of the ptosis.[3] Cruz et al demonstrated that advancement of the levator aponeurosis with lower eyelid retractors resection had some functional efficacy and increased palpebral fissure height.[7] Morrison et al suggested that the treatment of choice is orbicularis myectomy; however, some procedures such as lateral canthopexy, levator advancement, and lower eyelid retractors resection can be combined with that method for better functional and cosmetic effects.[9]

Because our patient was visually disabled and required a faster treatment, a different surgical method was chosen, and preseptal and pretarsal orbicularis myectomy and levator muscle tucking were performed. During the procedure under general anesthesia, spontaneous muscle spasms continued. These vermiform contractions were a useful guide for the amount of orbicularis myectomy required because they were decreased as the myectomy proceeded. The patient responded to surgical management very well and was relieved from constant blepharospasm.

In conclusion, we believe that although different methods have been described for the management of blepharospasm in SJS, the surgical method is faster and has a better functional effect. It seems that a single dose of botulinum toxin, which is mostly effective in essential blepharospasm, has insignificant and negligible results in severe blepharospasm due to SJS. Furthermore, during the surgical procedure, vermiform contraction of the orbicularis muscle may be utilized as a beneficial indicator for sufficient myectomy in these patients.

Declaration of Patient Consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their

images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial Support and Sponsorship

Nil.

Conflicts of Interest

There are no conflicts of interest.

REFERENCES

1. Vargel I, Canter HI, Topaloglu H, Erk Y. Results of botulinum toxin: An application to blepharospasm Schwartz-Jampel syndrome. *J Craniofac Surg* 2006;17:656-660.
2. Nicole S, Ben Hamida C, Beighton P, Bakouri S, Belal S, Romero N, et al. Localization of the Schwartz-Jampel syndrome (SJS) locus to chromosome 1p34-p36.1 by homozygosity mapping. *Hum Mol Genet* 1995;4:1633-1636.
3. Lucci LM, Yen MT, Anderson RL, Hwang IP, Black RE. Orbicularis myectomy with levator advancement in Schwartz-Jampel syndrome. *Am J Ophthalmol* 2001;132:799-801.
4. Giedion A, Boltshauser E, Briner J, Eich G, Exner G, Fendel H, et al. Heterogeneity in Schwartz-Jampel chondrodystrophic myotonia. *Eur J Pediatr* 1997;156:214-223.
5. Bastola P. Schwartz-Jampel syndrome. *Kathmandu Univ Med J (KUMJ)* 2010;8:348-351.
6. Eikermann M, Bredendiek M, Schaper J, Hövel M, Peters J. Resistance to rocuronium in a child with Schwartz-Jampel syndrome type 1 B. *Neuropediatrics* 2002;33:43-46.
7. Cruz AA, Souza CA, Plastino Júnior LS. Levator aponeurosis surgery in Schwartz-Jampel syndrome. *Ophthal Plast Reconstr Surg* 1998;14:271-276.
8. Viljoen D, Beighton P. Schwartz-Jampel syndrome (chondrodystrophic myotonia). *J Med Genet* 1992;29:58-62.
9. Morrison DA, Mellington FB, Hamada S, Moore AT. Schwartz-Jampel syndrome: Surgical management of the myotonia-induced blepharospasm and acquired ptosis after failure with botulinum toxin a injections. *Ophthal Plast Reconstr Surg* 2006;22:57-59.