WebLog Journal of Ophthalmology and Visual Sciences Published: 04 Jun, 2025 Published: 04 Jun, 2025

Case Report പ്പ

Management of Ocular Pathology in Patient with Schwartz-Jampel Syndrome: About a Clinical Case



OPEN ACCESS

*Correspondence:

Dr. Ana Morales Becerra, MD., Department of Ophthalmology, Pediatric Ophthalmology Unit, Hospital Arruzafa, 14012 Cordoba, Tel: 0034 957340118, Spain,

> E-mail: anamb1986@gmail.com Received Date: 28 May 2025 Accepted Date: 02 Jun 2025 Published Date: 04 Jun 2025

Citation.

Becerra AM, Torres García DJ, Morenilla BP, Gómez AÁ, Mesa VD, Palacios DC. Management of Ocular Pathology in Patient with Schwartz-Jampel Syndrome: About a Clinical Case. WebLog J Ophthalmol Vis Sci. wjovs.2025.f0403. https://doi. org/10.5281/zenodo.16040597

Copyright© 2025 Dr. Ana Morales Becerra. This is an open access article distributed under the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.

Dr. Ana Morales Becerra^{1*}, Diego José Torres García¹, Beatriz Perez Morenilla¹, Ana Álvarez Gómez¹, Vanessa Díaz Mesa², David Cerdán Palacios²

¹Department of Ophthalmology, Pediatric Ophthalmology Unit, Hospital Arruzafa, 14012 Cordoba, Spain

²Department of Ophthalmology, RDI Unit, Hospital Arruzafa, 14012 Cordoba, Spain

Abstract

Importance: Schwartz-Jampel syndrome (SJS) is a rare genetic disorder that affects bone and muscle development, causing muscle rigidity, joint contractures, and skeletal anomalies.

Ophthalmological involvement in SJS is often associated with facial features such as blepharophimosis, ptosis and blepharospasm.

Objective(s): To evaluate the management of strabismus in a patient with Schwartz-Jampel syndrome and assess the efficacy of botulinum toxin injection followed by surgical intervention.

Design: Case report and treatment analysis.

Setting: Ophthalmology in a tertiary care hospital.

Participants: A 30-year-old woman with a known diagnosis of Schwartz-Jampel syndrome presenting with double vision and worsening strabismus.

Intervention for Clinical Trials or Exposure(s) for Observational Studies: The patient initially received botulinum toxin injections in both medial rectus muscles (7.5 IU on the right and 10 IU on the left). Following partial improvement in strabismus, surgical intervention under general anesthesia was performed, involving medial rectus weakening and lateral rectus reinforcement.

Main Outcome(s) and Measure(s): Primary outcomes included the degree of strabismus as measured by cover test (alternating esotropia) and the resolution of diplopia. Secondary outcomes included the limitation of eye abduction and the success of surgical correction.

Results: Botulinum toxin injections reduced the magnitude of strabismus from 90 prism diopters (PD) to 60 PD. Surgical correction was performed with medial rectus weakening and lateral rectus reinforcement. Postoperatively, the patient showed no diplopia, and cover test revealed orthotropia. One month post-surgery, the patient developed a minor left esotropia (10-12 PD) with minimal right eye abduction limitation (1+).

Conclusion & Relevance: This case highlights the successful management of strabismus in a patient with Schwartz-Jampel syndrome using an initial treatment of botulinum toxin injections followed by surgical correction. The approach offers an effective strategy for managing complex strabismus in rare conditions, emphasizing the importance of individualized treatment and multidisciplinary care. Further studies are needed to better understand the ophthalmological manifestations of SJS and refine treatment protocols.

Keywords: Adult Acquired Esotropia, Diplopia, Lateral Rectus Resection, Prisms

Introduction

Schwartz-Jampel syndrome (SJS) or myotonic chondrodystrophy is a disease that affects the development of bones and muscles [1-4], a rare entity of autosomal recessive inheritance, although occasionally cases of dominant inheritance have been described [5]. Classic form appears in childhood, while neonatal variant (more severe and rare) is usually diagnosed at birth.

Ophthalmological involvement in these patients is associated with their muscular alteration, generating a 8 mask fascies associated with blepharophimosis.

In our clinical case, we present a 30-year-old woman who came to our clinic with double vision and

subacute strabismus of esotropia without signs of blepharophimosis and with a genetic, radiological and electromyography diagnosis of SJS.

Materials and Methods

30-year-old woman comes for strabismus evaluation. In her ophthalmological history, she describes esotropia since she was a child, which was treated with optical correction without complete resolution. She never had diplopia, but 3-4 months ago she started having it because strabismus has increased, associated with a loss of vision in the right visual field initially and later in the left.

As personal history, she reports a diagnosis of SJS, which is not related to strabismus. When she has had flare-ups due to his illness, they have not been associated with worsening of strabismus. Currently, she has not suffered a crisis for a long time and is very controlled.

In ophthalmological examination, visual acuity was LogMAR 0 in both eyes (OU) with this optical correction: +1.25-1.25 x 135° and +1.25-1.25 x 65°. Normal anterior segment, normal posterior segment, OU intraocular pressure: 12 mmHg, Worth Lights: suppress left eye, cover test (without and with correction): alternating esotropia 90 PD, but patient only tolerates 40 PD. If we increase the amount, she reports diplopia. Extrinsic ocular motility (EOM): limitation of abduction in OU.

Patient does not report any other type of associated symptoms.

Initially we requested imaging proof; an emergency CT scan and MRI with normal result.

Due to patient's pathology and risk of malignant hyperthermia with general anesthesia, as first treatment we carry out a botulinum toxin injection in both medial rectums: 7.5 IU in right and 10 IU in left.

Three months after treatment, patient showed improvement in strabismus, but not complete resolution. In her exploration, in cover test: alternating esotropia of 60 PD, well tolerated. Therefore, there was a correspondence between magnitude of strabismus and need for surgical treatment to resolve diplopia. In EOM: Abduction limitation persists, although, she passes of midline.

Despite this, her inability to lead a normal life led us to consider surgical treatment under general anesthesia after speaking with anesthesia team.

In examination under general anesthesia, we observed a positive duction test with 3+ to abduction in OU. We weakened both medial rectus muscles by 6 millimeters and a 6 mm left lateral rectus reinforcement. During surgery we found thickened medial rectus and a very thinned lateral rectus. Surgery was uneventful.

In initial postoperative period, patient did not present diplopia and cover test examination was orthotropia, with normal EOM except for a limitation of right eye abduction 1+.

One month after surgery, there is not diplopia, but in exploration of cover test: left esotropia 10-12 PD appears and limitation of abduction of right eye of 1+.

Results and Discussion

SJS presents with hypertrophy and muscle rigidity associated with joint contractures and various skeletal anomalies (chondrodysplasia:

short stature, pectus carinatum, kyphoscoliosis, bowing of long bones, epiphyseal, metaphyseal or hip dysplasia) and facial, with characteristic mask fascies, with blepharospasm, narrow palpebral fissures (blepharophimosis), small mouth with "furrowed" lips and chin [1-4]. Muscle stiffness is due to permanent and marked myotonia by continuous muscle activity on electromyogram (EMG), without there being an electric silence with muscle at rest [6]. Diagnosis is usually made in neonatal or childhood period, and a diagnosis in adulthood is exceptional.

We can observe ophthalmological involvement of these patients directly in their face appearance: eye folds due to a narrowing of eyelids depression (blepharophimosis), eyes almost closed due to upper eyelid drooping (ptosis) and strongly contracted eyelids because myotonia prevents relaxation of eyelid muscles (blepharospasm).

Its discovery was through Schwartz and Jampel: two American doctors, who worked at Brooklyn hospital, pediatrician and ophthalmologist [6]. In 1962, they jointly reported the observation of two children, brother and sister, born to an unrelated and healthy couple. These children presented with congenital blepharophimosis associated with myopathy, joint deformities, and a height below the third percentile. Myotonic component of syndrome was not identified at that time because it was not possible to perform an electromyogram (EMG) in any of patients. Lacking this crucial element, authors proposed to classify this original observation rather in arthrogryposis group. Years later, they could perform EMG that clearly demonstrated presence of anomalies, that resemble myotonic phenomena [7]. Muscle biopsy is inconclusive, detecting only discrete atrophy of muscle fibers of non-specific nature [8-10].

Until year 2000, SJS could not be confirmed genetically [11-12]. These patients had loss of function in gene HSPG2 (1p36), which encoded perlecan, a large protein located in the extracellular matrix of endothelial and epithelial cells [13].

Diagnosis is established based on demonstration of myotonia by electromyography and chondrodysplasia by radiographs. Also, genetic study is decisive.

Currently, SJS is divided into two types, 1 and 2. Type 2, or neonatal Schwartz Jampel 70 syndrome, is now considered a different, more serious disease called Stuve-Wiedemann syndrome, caused by mutations in the LIFR gene. Type 1, according to severity and age at which it begins, is subdivided into type 1A and 1B. 1A is considered the classic and most commonly recognized type. People with 1A generally have milder symptoms that begin later in childhood, while 1B has more severe symptoms that become apparent after birth.

Treatment of patients with SJS is mainly supportive and should be offered by a multidisciplinary team composed of neurologists, geneticists, physiotherapists, orthopedic surgeons, ophthalmologists and psychologists. Medical treatment is based on muscle relaxants and antiepileptic drugs. Effect of botulinum toxin injections for management of blepharospasm has been investigated, with limited and variable results [14].

There is no official registry of patients with SJS. To date it is estimated that around 150 the number of published cases of SJS worldwide [8].

Therefore, in the literature, only one clinical case has been described, which an alteration of extrinsic ocular motility was associated through a dissociated vertical deviation [15]; a type of

strabismus completely different from our case.

We are faced with patient with not previously described pathology and with anesthetic risks that make us question the treatment to be carried out. We initially decided on treatment using botulinum toxin which, although it was not a solution, it reduced the amount of surgical treatment and we could evaluate whether lateral rectus has functionality when we have medial rectus paralyzed.

Result after botulinum toxin was successful in our objective since we managed to reduce the magnitude of strabismus and restriction of abduction. Despite this, we had to face general anesthesia, which, with caution, did not pose any problem.

Surgical treatment and results, with indicated surgical figures, were successful. SJS is a rare disease with a difficult approach due to its pathology and lack of literature. It is very important to individualize each case. But we consider that the approach to strabismus with an initial treatment with botulinum toxin and subsequently treatment on necessary extraocular muscles is a success.

Funding

The authors received no financial support for the research, authorship, and/or publication of this article.

Declaration of Conflicting Interests

The Authors declare(s) that there is no conflict of interest.

References

- Spaans F, Theunissen P, Reekers AD, Smit L, Veldman H. SchwartzJampel syndrome: I. Clinical, electromyographic, and histologic studies. Muscle Nerve 1990; 13: 516-27. doi: 10.1002/mus.880130608.
- Greze J, Baldet P, Dumas R, Cadilkac J, Pages A, Jean R. Dystrophie osteochondro-musculaire de Schwartz-Jampel. Arch Fr Pediatr 1975; 32: 59-75.
- Pavone L, Mollica F, Grasso A, Cao A, Gullotta F. Schwartz-Jampel syndrome in two daughters of first cousins. J Neurol Neurosurg Psychiatry 1978; 41: 161-9. doi: 10.1136/jnnp.41.2.161.
- 4. Becher S, El Mabrouk J, Debbiche A, Hammou A, Ghram N, Makni S, et al. Le syndrome de Schwartz-Jampel. Arch Fr Pediatr 1992; 49: 799-802.
- Ferrannini E, Perniola T, Krajewska G, Serlenga L, Trizio M. Schwartz-Jampel syndrome with autosomal dominant inheritance. Eur Neurol 1982; 21: 137-46. doi: 10.1159/000115471.

- Schwartz O, Jampel RS. Congenital blepharophimosis associated with a unique generalized myopathy. Arch Ophthalmol. 1962 Jul. 68: 52-7. doi: 10.1001/archopht.1962.00960030056011.
- Ishpekova B, Rasheva M, Moskov. Schwartz-Jampel syndrome: clinical, electromyographic and genetic studies. Electromyogr Clin Neurophysiol 1996; 36: 151-5.
- Urtizberea JA, Severa G, Ropars J, Malfatti E. Le syndrome de Schwartz-Jampel [The Schwartz-Jampel syndrome]. Med Sci (Paris). 2023 Nov; 39 Hors série nº 1: 37-46. French. doi: 10.1051/medsci/2023133.
- Huttenlocher PR, Landwirth J, Hanson V, et al. Osteo-chondro-muscular dystrophy. A disorder manifested by multiple skeletal deformities, myotonia, and dystrophic changes in muscle. Pediatrics 1969; 44: 945-58.
- Aberfeld DC, Hinterbuchner LP, Schneider M. Myotonia, dwarfism, diffuse bone disease 116 and unusual ocular and facial abnormalities (a new syndrome). Brain 1965; 88: 313-22. doi: 10.1093/brain/88.2.313.
- 11. Arikawa-Hirasawa E, Le AH, Nishino I, et al. Structural and functional mutations of the perlecan gene cause Schwartz-Jampel syndrome, with myotonic myopathy and chondrodysplasia. Am J Hum Genet 2002; 70: 1368-75. doi: 10.1086/340390.
- Dagoneau N, Scheffer D, Huber C, et al. Null leukemia inhibitory factor receptor (LIFR) mutations in Stüve-Wiedemann/Schwartz-Jampel Type 2 syndrome. Am J Hum Genet 2004; 74: 298–305. doi: 10.1086/381715.
- 13. Arikawa-Hirasawa E, Le AH, Nishino I, Nonaka I, Ho NC, Francomano CA, Govindraj P, Hassell JR, Devaney JM, Spranger J, Stevenson RE, Iannaccone S, Dalakas MC, Yamada Y. Structural and functional mutations of the perlecan gene cause Schwartz-Jampel syndrome, with myotonic myopathy and chondrodysplasia. Am J Hum Genet. 2002 May; 70(5): 1368-75. https://doi.org/10.1086/340390.
- 14. Nessler M, Puchala J, Kwiatkowski S, Kobylarz K, Mojsa I, Chrapusta-Klimeczek A. Multidisciplinary approach to the treatment of a patient with chondrodystrophic myotonia (Schwartz-Jampel vel Aberfeld syndrome): case report and literature review. Ann Plast Surg. 2011 Sep; 67(3): 315-9. doi: 10.1097/SAP.0b013e3181fac1ec.
- Rebolleda Fernández G, Muñoz Negrete FJ, Lozano C. Manifestaciones oculares en el Síndrome de Schwartz-Jampel. Archivos de la Sociedad Española de Oftalmologia, ISSN 0365-6691, Vol. 59, Nº. 6 (DIC), 1990, págs. 669-672.