CASE REPORT
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Surgical treatment of vitreoretinal complications in a patient with Stickler syndrome type 1: a case report

Nicolás Rivera-Valdivia[1], Carlos Salgado-Cerrate[1], Pablo Cabal-López[1], Hiroshi Maeda-Yasunaga[1], Laura Pacheco-Palomino[2], Carlos Abdala-Caballero[3]

[1]Retina and Vitreous Fellow, Grupo Oftalmológico Abdala-Figuerola AF, Barranquilla, Colombia
[2]Ophthalmology Resident, Universidad del Sinú, Cartagena de Indias, Colombia
[3]Retina and Vitreous Surgeon, Grupo Oftalmológico Abdala-Figuerola AF, Barranquilla, Colombia

ABSTRACT

We present the clinical case of an 8-years-old male patient with a genetic diagnosis of Stickler syndrome type 1 and the management of associated vitreoretinal complications.

The patient had an antecedent of no light perception in his left eye secondary to retinal detachment treated in another health centre. He consulted with a history of blunt trauma in his head with an unremarkable anterior segment and fundus exam. Scleral indentation showed no lesions to be treated in the right eye. We recommended follow-up every six months. The patient did not come to controls. He was consulted three years later because of visual loss in his right eye. Fundus showed a total retinal detachment secondary to a giant retinal tear of 320º with macular involvement and choroidal detachments. Therefore, scleral buckling, lensectomy, pars plana vitrectomy, inferior retinectomy, endovenous laser treatment (endolaser), and silicone oil tamponade were performed. After six weeks of follow-up, the patient presented an inferior re-detachment with grade C3 vitreoretinal proliferation. A new procedure of silicone oil removal, epiretinal membrane removal, enlargement of inferior retinectomy, endolaser, and silicone oil tamponade was indicated. After 6-months of follow-up, no new retinal detachment was presented, and the best-corrected visual acuity was 20/100.

KEY WORDS: Stickler syndrome; retinal detachment; pars plana vitrectomy; vitreous; genetics

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INTRODUCTION

Stickler syndrome is a well-known hereditary disease with an autosomal dominant or recessive genetic pattern depending on the pathogenic variant [1, 2]. It was first reported in 1965 by Stickler et al. [3]. He described ocular findings, auricular, orofacial (isolated or as part of the sequence of Robin), joints changes by defects in collagen type II, IX, and XI [2, 4–6]. Its annual incidence is estimated at 1:7.500 to 1:10.000 [5–8], and it is considered the most frequent cause of non-traumatic retinal detachment in the pediatric population [8].

In this case, we present a patient with a genetic diagnosis of Stickler syndrome type 1 and asymmetric bilateral rhegmatogenous retinal detachment.

CORRESPONDING AUTHOR:
Carlos Abdala-Caballero MD, Grupo Oftalmológico Abdala-Figuerola AF, Carrera 30 # Corredor Universitario 1-850, Torre Médica, Consultorio 812, Code 081007, Barranquilla, Colombia, tel: +57 (311) 296 8674; e-mail: fellow.retina.abdala@gmail.com

CASE PRESENTATION

An 8-years-old male patient with a genetic diagnosis of Stickler syndrome type 1 (COL2A1, autosomal dominant inheritance pattern with an association to androgenesis type 2 and osteoarthritis with chondrodysplasia media), history of lower maxillary osteosynthesis, tracheostomy, and palatorrhaphy was admitted to our facility with total retinal detachment in his left eye (OS). The patient was previously treated in another centre before arriving at our healthcare institution, where he consulted because of blunt trauma with a balloon in the frontal region. Best-corrected visual acuity (BCVA) was in the right eye (OD): 20/50 and OS: no light perception. Anterior segment examination revealed OD unremarkable and OS with band keratopathy, 30% hyphema, rubeosis iridis, and surgical aphakia. The OD fundus examination was unremarkable.

On the other hand, OS presented with grade 4 vitreous hemorrhage, not allowing display of the posterior pole. An eye ultrasound and examination under anesthesia were conducted after one month with indirect ophthalmoscopy and indentation, assessing that OD retina was applied without peripheral retinal degenerative lesions. OS we found partial vitreous hemorrhage in reabsorption and total retinal detachment with rigid retina and atrophic appearance. OS was diagnosed as retinal re-detachment with an amaurotic eye. We recommended observation and control every six months. The patient came to consult three years later because of an OD sudden vision loss. BCVA was OD: light perception and OS: no light perception. The examination under biomicroscopy in OD was unremarkable. OS had no changes with rubeosis iridis and surgical aphakia. The fundus of the OD eye showed a giant retinal tear of 320º with curled edges and total retinal detachment with macular involvement associated with choroidal detachments; OS was not assessable. Under this context, the surgery indicated was scleral buckling, lensectomy, pars plana vitrectomy, inferior retinectomy, endolaser, and 5000 cSt silicone oil tamponade. The procedure was carried out without complications. Post-operative follow-up was started according to the protocol. Six weeks after the procedure, the patient presented an inferior re-detachment with grade C3 vitreoretinal proliferation and stellar folds in the OD (Fig. 1). A new procedure was indicated to re-apply the retina. We performed silicone oil removal, epiretinal membrane removal, enlargement of inferior retinectomy, endolaser, and silicone oil tamponade. All went without complications (Fig. 2, 3). In the sixth post-operative month, OD progressed satisfactorily with retina and macula entirety attached with transparent silicone oil without signs of emulsification. BCVA OD was 20/100.

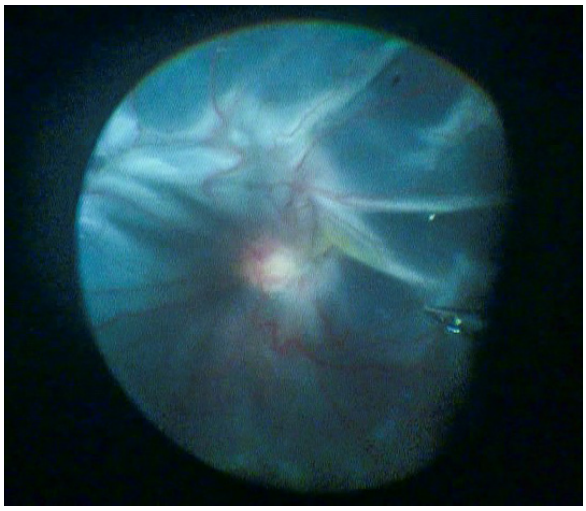

FIGURE 1. Retinal re-detachment with the presence of preretinal membranes

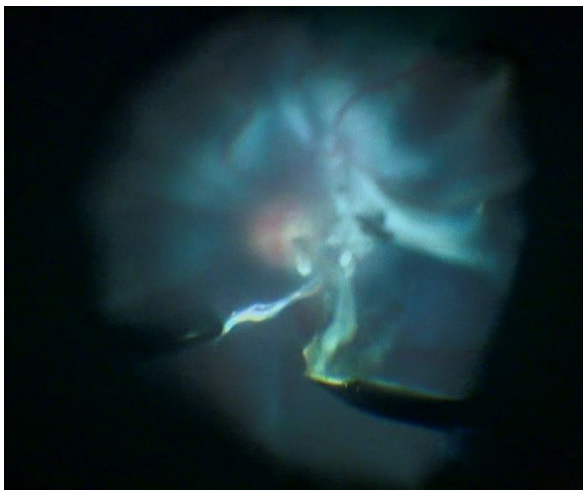

FIGURE 2. Dissected preretinal membrane

DISCUSSION

Stickler syndrome is an inherited connective tissue disorder, described for the first time in a family with ocular, auditory, orofacial, and musculoskeletal disorders [3], which occurs as a result of a collagen type II, IX, and XI defect presents in hyaline cartilage and secondary vitreous [5, 6]. The oph-

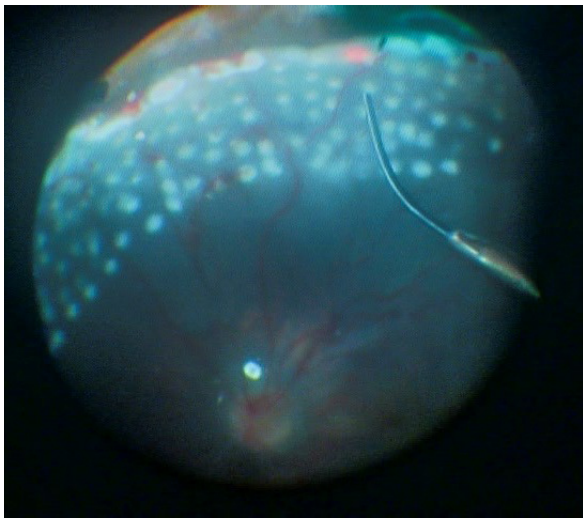

FIGURE 3. Inferior retinectomy surrounded to edges with 5-lines laser photocoagulation

thalmological manifestations can be of the anterior and posterior segment, the most frequent being: pre-senile cortical cataracts, open-angle glaucoma, high myopia, and lattice-type peripheral degenerations in the retina that can lead to the formation of giant retinal tears and retinal detachment [6,7,9], as it is the case of our patient.

The various disease subtypes are caused by a mutation in one of the six genes: COL2A1, COL11A1, COL11A2 — associated with dominant inheritance, and COL9A1, COL9A2, COL9A3 — associated with recessive inheritance [1,2,8]. Ophthalmologically, Stickler syndrome type 1, which presents a mutation in the COL2A1 gene [10], and is part of more than 80% of all cases [5], is associated with an "optically empty" vitreous with fibrillar and membranous degeneration, leading to at a high rate of rhegmatogenous retinal detachment, more frequently in pediatric age [11]. In this population, non-traumatic retinal detachment is a rare finding. However, in patients diagnosed with Stickler syndrome, it is estimated to be present in up to 70% of cases [12,13], followed by other complications such as cataract (49%) and ocular hypertension (10%) [12].

The anatomic success rate in the surgical repair of retinal detachments in the pediatric population depends on the etiology, showing rhegmatogenous detachment better results than tractional detachments [11]. However, surgical treatment represents significant challenges in this population given the vitreous's abnormal features, which are firmly adhered to borders of perivascular lattice degenerations in these patients [6]. Various procedures have been proposed for treatment and prophylaxis. Surgical techniques described in the literature have different modes of addressing these cases. Abeysiri et al. [8] obtained retina reattachment results after scleral buckling, pars plana vitrectomy, or a combination of both, 67%, 84%, and 79%, respectively. Reddy et al. [6] showed 100% anatomic success in any of the three modes.

Nevertheless, most of the patients required multiple procedures to reapply their retina (81.3%). In our case, the second surgery to reapply the retina was necessary. The procedure performed involved scleral buckling, pars plana vitrectomy, and retinectomy with silicone oil tamponade.

On the other hand, having a prophylaxis scheme is as important as surgical treatment. For this, a different approach has been proposed, such as the Cambridge Protocol for giant tears with cryotherapy, where Fincham et al. [5] demonstrated efficacy and safety in reducing the risk of retinal detachment in the unaffected eye in patients with Stickler syndrome type 1. Leiba et al. [14] reported that 10% of the treated group versus 44% of the control group developed retinal detachment in a study with focal laser photocoagulation or 360° with argon.

CONCLUSION

The patients with Stickler syndrome are a significant challenge in the pre-operative and post-operative management where technique varies the results obtained in different series. It is vital to choose the best approach based on the surgeon's experience.

We believe that in cases with Stickler syndrome and associated retinal detachment, a prophylaxis protocol and quick and efficient treatment of the contralateral eye is essential to reduce the ocular morbidity in this group of patients.

Conflict of interest

The authors declare that they have no conflict of interest.

Funding

None declared.

Informed consent and human and animal rights statements

Informed consent has been obtained from all individuals included in this study.

Authorization for the use of human subjects and ethical approval

The research related to human use complies with all the relevant national regulations, institutional policies, is in accordance with the tenets of the Helsinki Declaration and has been approved by the Ethics Committee of Grupo Oftalmológico Abdala-Figuerola AF, Barranquilla, Colombia.

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REFERENCES

1. Snead MP, Yates JR. Clinical and Molecular genetics of Stickler syndrome. J Med Genet. 1999; 36(5): 353–359, indexed in Pubmed: 10353778.
2. Robin NH, Moran RT, Ala-Kokko L, et al. Stickler Syndrome. US National Library of Medicine: Gene Reviews. 2020: 1–19.
3. Stickler GB, Belau PG, Farrell FJ, et al. Hereditary Progressive Arthro-Ophthalmopathy. Mayo Clin Proc. 1965; 40: 433–455, indexed in Pubmed: 14299791.
4. Öztürk C, Sarıgül Sezenöz A, Yılmaz G, et al. Bilateral Asymmetric Rhegmatogenous Retinal Detachment in a Patient with Stickler Syndrome. Turk J Ophthalmol. 2018; 48(2): 95–98, doi: 10.4274/tjo.60430, indexed in Pubmed: 29755825.
5. Fincham G, Pasea L, Carroll C, et al. Prevention of Retinal Detachment in Stickler Syndrome. Ophthalmology. 2014; 121(8): 1588–1597, doi: 10.1016/j.ophtha.2014.02.022, indexed in Pubmed: 24793526.
6. Reddy DN, Yonekawa Y, Thomas BJ, et al. Long-term surgical outcomes of retinal detachment in patients with Stickler syndrome. Clin Ophthalmol. 2016; 10: 1531–1534, doi: 10.2147/OPTH.S111526, indexed in Pubmed: 27574392.
7. Wubben TJ, Branham KH, Besirli CG, et al. Retinal detachment and infantile-onset glaucoma in Stickler syndrome associated with known and novel COL2A1 mutations. Ophthalmic Genet. 2018; 39(5): 615–618, doi: 10.1080/13816810.2018.1509355, indexed in Pubmed: 30130436.
8. Abeysiri P, Bunce C, da Cruz L. Outcomes of surgery for retinal detachment in patients with Stickler syndrome: a comparison of two sequential 20-year cohorts. Graefes Arch Clin Exp Ophthalmol. 2007; 245(11): 1633–1638, doi: 10.1007/s00417-007-0609-2, indexed in Pubmed: 17579881.
9. Stickler GB, Hughes W, Houchin P. Clinical features of hereditary progressive arthro-ophthalmopathy (Stickler syndrome): a survey. Genet Med. 2001; 3(3): 192–196, doi: 10.1097/00125817-200105000-00008, indexed in Pubmed: 11388760.
10. Rose PS, Levy HP, Liberfarb RM, et al. Stickler syndrome: clinical characteristics and diagnostic criteria. Am J Med Genet A. 2005; 138A(3): 199–207, doi: 10.1002/ajmg.a.30955, indexed in Pubmed: 16152640.
11. Ebert JJ, Utz VM, Sisk RA. Bilateral rhegmatogenous retinal detachments from giant retinal tears in an infant with abusive head trauma and Stickler syndrome. Am J Ophthalmol Case Rep. 2020; 17: 100581, doi: 10.1016/j.ajoc.2019.100581, indexed in Pubmed: 31909295.
12. Rishi P, Maheshwari A, Rishi E. Stickler syndrome. Indian J Ophthalmol. 2015; 63(7): 614–615, doi: 10.4103/0301-4738.167114, indexed in Pubmed: 26458481.
13. Coussa RG, Sears J, Traboulsi EI. Stickler syndrome: exploring prophylaxis for retinal detachment. Curr Opin Ophthalmol. 2019; 30(5): 306–313, doi: 10.1097/ICU.0000000000000599, indexed in Pubmed: 31313752.
14. Leiba H, Oliver M, Pollack A. Prophylactic laser photocoagulation in Stickler syndrome. Eye (Lond). 1996; 10 (Pt 6): 701–708, doi: 10.1038/eye.1996.164, indexed in Pubmed: 9091366.