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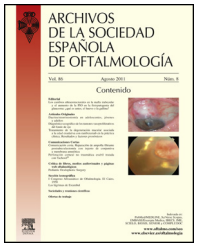

Original article

Stickler syndrome. Epidemiology of retinal detachment☆

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ABSTRACT

Objective: A review was performed on all patients with Stickler syndrome that had been treated in our Center since it was diagnosed, in order to evaluate the risk of suffering a retinal detachment (RD).

Methods: A total of 14 patients, diagnosed by clinical criteria, were included. The following variables were evaluated: age, gender, ocular background, follow-up, initial and final visual acuity (VA), optical prescription, prophylactic treatment, surgery and techniques performed. The risk age to suffer a RD, as well as cataracts, was determined by using the Kaplan–Meier survival curve analysis.

Results: From a total of 5 men and 9 women, the median initial VA was 0.35, which was the same as the final VA. The median optical prescription was −9.5 D myopia. The median of follow-up was 7 years. Ocular background was 4 RD cases and 2 Lasik surgeries. The operations performed were 8 RD, 12 cataract, 2 glaucoma, 2 macular hole, and one endotropia. The median age of RD was 20 years and cataract 34 years. As regards surgical tecnique, 4 scleral buckle cases, and 4 scleral buckle + pars plana vitrectomy cases were formed. The prophylactic treatments performed were: one scleral buckle case, 4 endolaser photocoagulation, and one cryotherapy. Two of which presented with RD.

Conclusion: In the series presented, retinal detachment in Stickler syndrome mainly occurs in the second decade of life, with cataracts mainly developing in the fourth decade.

Síndrome de Stickler. Epidemiología del desprendimiento de la retina

Palabras clave:
Síndrome de Stickler
Desprendimiento de retina

RESUMEN

Objetivo: Revisión de todos los pacientes con síndrome de Stickler que se han tratado en nuestro centro desde su descripción, para valorar el riesgo de padecer desprendimiento de la retina (DR).

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Vitrectomía
Catarata
Glaucoma
Agujero macular

Métodos: Un total de 14 pacientes, diagnosticados por criterios clínicos, en los que hemos valorado las siguientes variables: sexo, edad, antecedentes oculares, seguimiento, agudeza visual (AV) inicial y final, refracción, tratamiento profiláctico, cirugías y la técnica empleada. Mediante el análisis de curva de supervivencia de Kaplan-Meier hemos determinado la edad riesgo de padecer DR así como cataratas.

Resultados: En total fueron 5 hombres y 9 mujeres. La AV mediana inicial fue de 0,35, igual que la AV final. Refracción mediana de −9,5 dioptrías de miopía. La mediana de seguimiento de 7 años. Los antecedentes oculares fueron 4 DR y 2 Lasik. Las cirugías que realizamos han sido 8 DR, 12 cataratas, 2 glaucomas, 2 agujeros maculares y una endotropía. La mediana de años en el que sucedió el DR fue de 20 y las cataratas a los 34. La técnica quirúrgica utilizada en 4 casos ha sido el cerclaje y en los otros 4 restantes cerclaje con vitrectomía vía pars plana. Un total de 6 ojos han sido tratados profilácticamente: uno con cerclaje, 4 con fotocoagulación láser y otro con criocoagulación; de estos, 2 han presentado DR.

Conclusiones: En nuestra serie, el DR en esta dolencia se desarrolla mayoritariamente en la segunda década de la vida. Las cataratas, fundamentalmente en la cuarta década.

Introduction

The Stickler syndrome was first described in 1965 by Gunnar Stickler.[1] This syndrome is a hereditary arthro-ophthalmopathy with an estimated incidence of one case for every 10,000 births.[2] Inheritance is autosomic dominant in the majority of cases.[2] It is caused by collagen alteration with bilateral ophthalmological as well as orofacial (Pierre Robin complex: micrognathia, palatine fissure and glossoptosis)[3] and skeletal involvement (Fig. 1). Typical ophthalmological findings of this disease are congenital high myopia, cataracts and retinal problems such as changes in the vitreous, radial degeneration in the periphery and high risk of regmatogenous retina detachment (RD)[4] (Fig. 1). Diagnostic criteria for the most frequent type of the Stickler syndrome were established in 2005.[4] The main purpose was to assess the incidence of RD in our population as well as treatment, evolution and prophylaxis.

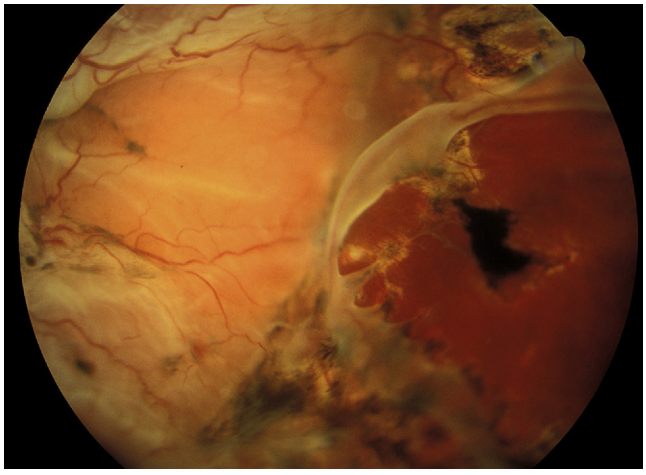

Fig. 1 – Retina detachment with vitreoretinal proliferation and peripheral tear at 2 h. Case #3.

Subjects, material and method

Since the Stickler syndrome was first described up to now we have documented 14 cases in the Barraquer Ophthalmology Center. The diagnostic was based on clinical criteria: ocular, orofacial and skeletal alterations. The cases involved 5 males and 9 females with ages comprised between 2 and 42 years (median 16). The follow-up ranged between 1 and 23 years (median 7).

We have assessed initial and final visual acuity (VA), refraction error, age, ocular antecedents, follow-up surgeries performed, age of RD expression, age at cataract surgery, surgical technique applied for RD, prophylactic treatment and subsequent RD recurrence.

By means of the Kaplan–Meier survival curve analysis we have determined the retina detachment and cataract risk age in said population.

Results

The results obtained from the 14 cases are shown in Tables 1 and 2. Table 1 shows the RD patients totaling 9 cases. The 5 remaining cases without RD are shown in Table 2.

Initial VA ranges were between no perception of light and 1.2 (median 0.35). Cases 2 and 4 are patients who did not perceive light in one eye due to long-term chronic RD. The final VA also ranged between no perception of light and 1.2 (median 0.35). Case 3 evolved to no perception of light due to poor surgical evolution in the left eye.

All patients were myopic, between 1.5 and 17 diopters (median 9.5), with the exception of case 1 who was not submitted to refraction due to poor light perception and case 10 who had undergone LASIK operation for myopia.

Overall, 4 eyes had undergone RD operations previously in another service and 8 eyes underwent RD operation in our center.