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 ocular 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 technique, 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.

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Síndrome de Stickler. Epidemiología del desprendimiento de la 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).

Palabras clave:
Síndrome de Stickler
Desprendimiento de retina

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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 fue 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-ophthalmalopathy with an estimated incidence of one case for every 10,000 births.2 Inheritance is autosomic dominant in the majority of cases.3 It is caused by collagen alteration with bilateral ophthalmological as well as orofacial (Pierre Robin complex: micrognathia, palatine fissure and glossophtosis)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.

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.
Other ocular conditions which required surgery comprised 12 cataracts, 2 glaucoma, 2 macular holes and one endotropia.

Overall, 7 patients (50%) had cataracts, of which 5 were bilateral and 2 unilateral. Ages ranged between 7 and 40 years. Kaplan–Meier survival curve analyses indicated that the median age at which they underwent cataract surgery was 34.

Nine of all 14 patients (64.2%) developed RD (Fig. 1). Of these, 3 were bilateral (33.3%) and 6 unilateral (66.6%). Kaplan–Meier survival curve analyses indicated that the median age of retina detachment was 20.

Bilaterality risk was not assessed as it expressed in only 3 patients (cases 1, 2 and 3 shown in Table 1). The number of years elapsed prior to the involvement of the second eye was highly variable: 3, 6 and 13 years respectively. Case 1 had been intervened in another service for both eyes with poor anatomic and functional results, leaving an inoperable fundoscopic condition. Case 2 also visited our center after undergoing 3 previous operations with poor anatomic and functional results in the left eye. This case exhibited RD in the right eye without previous prophylactic treatment, being intervened with cerclage which achieved retinal reapplication and a vision of 0.7. Case 3 was intervened for both eyes in our center. The first eye was operated at age 8, applying cerclage associated to vitrectomy due to blood remains in the vitreous cavity. This patient exhibited relapse which was intervened with vitrectomy, ending in a poorer visual result. The second RD appeared at age 21 in the right eye despite prophylactic cryocoagulation. The technique applied was cerclage with vitrectomy, with anatomic and functional success.

Overall, the surgical technique was scleral cerclage in 4 eyes (cases 2, 5, 7 and 9) and combined surgery comprising cerclage and vitrectomy in the remaining 4 cases (case 3 in both eyes, 6 and 8). Gas was used as tamponade in the left eye relapse.

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<th>Table 1 – Patients with RD.</th>
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<td>Follow-up (years)</td>
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<td>Refraction RE</td>
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<td>Age RD</td>
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<td>Age cataract</td>
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Ant.: antecedents; BE: both eyes; VA: visual acuity; Cat: cataract; RD: retina detachment; ET: endotropia; G: glaucoma; F: female; HM: hand movements; PLP: poor light to perception; ND: not documented; RE: right eye; LE: left eye; M: Male.

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<th>Table 2 – Patients without RD.</th>
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MH: macular hole; Ant.: antecedents; BE: both eyes; VA: visual acuity; Cat: cataract; T: tear; E: emmetropia; ETI: left endotropia; F: female; HM: hand movements; PLP: poor light perception; NA: not assessable; RE: right eye; LE: left eye; M: male.
of case 3, air in the contralateral eye and silicone in case 6. Vitrectomy was aborted in case 8 due to the impossibility of extracting vitreoretinal proliferations. As for the 4 remaining cases, the surgical technique is unknown because they were intervened in another service.

The majority of the patients operated by us improved or stabilized their VA, excepting cases 3 commented above and case 6 that exhibited diminished final VA.

Only 2 cases exhibited RD relapse, cases 2 and 3 commented above.

Prophylactic treatments were carried out in 6 eyes that exhibited vitreoretinal degeneration (Figs. 2 and 3) and were considered as population at risk. Four eyes were treated with laser photocoagulation (FCG), one with cryocoagulation and the other with cerclage. Despite prophylaxis, 2 of the 6 treated eyes, 1 with FCG laser and the other with cryocoagulation, developed RD (33.3%).

**Discussion**

The Stickler syndrome is an extraordinarily infrequent condition. In our center we have found only 14 cases since it was first described in 1965. Recently, Doray et al. found a single patient affected by the Stickler syndrome out of 321 orofacial alterations when these occur in a proportion of 2.1 hour 1000 inhabitants, representing an incidence of approximately one of for every 150,000.

The Stickler syndrome is the most frequent hereditary cause of RD. In our series of 14 eyes, 64.2% of cases exhibited RD. Some authors, such as Fincham et al. demonstrated the efficacy of prevention by means of cryocoagulation, or Leiba et al. with FCG. We have performed prophylactic treatment for vitreoretinal degeneration by means of FCG or cryocoagulation depending on patient cooperation, which was related to their age as some patients were very young. For patients who did not cooperate, cryocoagulation was performed in the operating theater under locoregional or general anesthesia. Even so, a high percentage of failures (33.3%) has occurred.

In what concerns patients with bilateral processes, case 3 is worthy of note as the intervention in the first eye (1992) failed and the second (2005) was successful. It is also worth mentioning that we utilized the same methodology but with different instruments. It must be taken into account that experience was gained with time and new technologies were developed in the interim which have improved prognosis.

The Stickler syndrome is not only associated to RD but can include other ocular conditions such as congenital glaucoma. In our series, said syndrome was associated to cataracts, glaucoma, macular hole and endotropia. Palisades degeneration are frequent, as well as the association with myopia. The present cases were all myopic in varying degrees. Half of the patients in this series (50%) had cataracts, which confirms the findings of other published series such as that of Seery et al. with 49.8%.

Diagnosing the Stickler syndrome at an early age is essential for preventing or treating its consequences which could be irreversible on some occasions.

As conclusion, it can be said that in the present series retina detachment developed mainly in the second decade of life and that surgery exhibited a low failure rate. Cataracts developed mainly in the fourth decade of life.

**Conflict of interests**

No conflict of interests has been declared by the authors.

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