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Prophylactic Treatments for Retinal Tears/Detachment in Stickler Syndrome: Single Institution Experience

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Abstract

Background: To report the clinical features and prophylactic treatment methods for retinal pathology in Stickler syndrome (SS) patients (pts).

Methods: A retrospective non-comparative interventional case series.

Results: The median age of the pts was 12 (range 0.2-68) years. Systemic features of SS were present in 23 pts. High myopia was present in 19 pts. Treatment was by laser retinopexy and or cryotherapy (n=15) and vitrectomy plus scleral buckle (SB) (n=6). Five fellow eyes with extensive lattice degeneration with or without associated retinal tears treated by prophylactic SB. The median follow-up was 65.5 months (range 5-226). Seven fellow eyes developed RD over a median duration of 22.5 (range 5-123) months, who were treated by vitrectomy plus SB (n=6) and vitrectomy, silicon oil and laser retinopexy (n=1).

Conclusions: Prophylactic treatments such scleral buckle and laser and cryotherapy prevented the development of retinal tears or detachment in pts with Stickler syndrome.

Keywords: Cryotherapy; Laser retinopexy; Lattice degeneration; Retinal detachment; Retinal tear; Sclera buckle; Stickler syndrome; Vitrectomy; Vitreous syneresis

Brief summary

Stickler syndrome patients are prone to develop multiple retinal tears and detachment at an early age due to the vitreoretinal degeneration. Prompt and prophylactic treatment of retinal abnormalities and regular follow-ups preserves the retinal anatomy and maintains vision.

Introduction

Stickler syndrome (SS) is an autosomal dominant connective tissue disorder affecting mostly the orofacial, ocular, auditory and musculoskeletal systems [1]. It is usually of 3 types and is associated with various vitreous anomalies [1-6]. Type I or membranous vitreous is the most common type (SS 1) and is caused by mutations in the COL2A1 gene. Type II or beaded vitreous (SS 2) is caused by mutations in COL11A1 and COL11A2 genes [2]. Type 3 SS, a rare entity is non-ocular type [2]. The characteristic systemic abnormalities of SS include sensorineural hearing loss, Pierre-Robin sequence, joint hypermobility, premature arthritis, flat mid face, depressed nasal bridge, and cleft palate and the common ocular abnormalities include high myopia, cataract, vitreoretinal degeneration and retinal detachment (RD) [1-6]. The purpose of the study was to report various clinical features of patients with Stickler syndrome, who presented to our center and discuss outcomes of prophylactic treatment modalities.

Material and methods

A retrospective non-comparative chart review (interventional case series) of patients with Stickler syndrome from March 1991 to February 2012 was performed. The data analysis included demographic information, family history of SS, systemic disease status, prior ocular treatment, ocular symptoms, clinical features and treatment methods. Duration to develop retinal tears or retinal detachment (RD) after initial treatment and whether prophylactic scleral buckle was performed in eyes with extensive lattice degeneration and retinal tears were noted. Status of the retina at the last follow-up (FU) and outcomes were also included in the study. Failure at last FU was noted as those patients who developed RD after initial treatment.

Statistical analysis: Data analysis was done using the SPSS version 17.0 (SPSS Inc., Chicago IL).

Results

Thirty-one patients with Stickler syndrome presented with various ocular symptoms. Table 1 describes the demographic information, family history and systemic features.

Characteristic feature	Total (n=31 pts)
Age (mean, median; range)	20, 12; 0.2-68
Gender	
Male	22
Female	9

Family history	
Yes	17
No	14
Affected family member	
Parents	12
Siblings	8
Grandparents	4
Systemic features	
Cleft palate	12
Musculoskeletal abnormalities	7
Hearing loss	1
Others	5
High myopia	
Yes	19
No	12
Initial treatment (before presenting to our clinic)	Eyes (n=62)
Laser retinopexy	6
Cryotherapy	4
Scleral buckle	1
Combination treatment	8
Enucleation	2
Observation	41

Table 1: Demographics, Family history and Systemic features (n=31 pts, 62 eyes).

The median age was 12 (range 0.2-68) years. Of the 31, 9 were females and 22 were males. Family history of SS was positive in 17 patients (pts). The most common associated systemic features of SS were cleft palate in 13, musculoskeletal in 7 pts. High myopia was present in 19 pts. Past eye treatments prior to presenting us were laser retinopexy (6 eyes), cryotherapy (4 eyes), scleral buckle (1 eye), combination treatment (vitrectomy, laser retinopexy or cryotherapy; eight eyes), enucleation (2 eyes), and observation (41 eyes). The most common presenting symptom was poor vision in 13 patients, floaters in 11 patients and 10 were asymptomatic (Table 2) and sent for referral ocular evaluation due to presence of systemic features of stickler's syndrome.

Characteristic feature	Total (n=31 pts)
Ocular symptoms	
Asymptomatic	10
Decreased vision	13
Floaters	11
Visual acuity	
Better than 20/40	26
20/50-20/200	18
Worse than 20/200	8
Fixes and follows	8
Enucleated at presentation	2
Intraocular pressure (mean, median; range) mm Hg	16.3, 16; 4-34
Fundus examination	Eyes (n=62)
Vitreous abnormalities	60
Lattice degeneration	13
Giant retinal tear	2
Retina detachment	5
Treatment	
Laser	14
Cryotherapy	1
Combination therapy	6
Observation	40
Retina abnormalities in fellow eye	
Exensive lattice degeneration	5
Retina tears	16
Retina detachment	7
Duration to develop retina detachment in fellow eyes (mean, median; range), months	40.7, 22.5; 5-123
Treatment of fellow eyes	
Prophylactic scleral buckle in fellow eyes	5
Laser	15
Cryotherapy	1
Combination treatment (vitrectomy, laser, cryo,± scleral buckle)	7

Table 2: Ophthalmic features at initial presentation, (n=31 pts, 62 eyes).

Visual acuity was better than 20/40 in 26 eyes, between 20/50 to 20/200 in 18 eyes, worse than 20/200 in 8 eyes; fix/follows light in eight eyes and two eyes were enucleated prior to visiting us. The median intraocular pressure was 16 (range 4-34) mm Hg. Anterior segment examination was within normal limits in 38 eyes, band keratopathy in 1 eye, cataract in 10 eyes, pseudophakia in 7 eyes and aphakia in 4 eyes. Vitreous syneresis was present in 25 eyes, membranes in 4 eyes, veils and opacities in 8, posterior vitreous detachment in 3, hemorrhage in 1, clear in 12, empty in 7. Indirect ophthalmoscopic examination revealed normal optic disc in 43 eyes, hyperemic in 2,

pale in 6 and myopic tilt in 9 eyes. Macula was normal in 41 eyes, pigment changes were seen in 11, detachment in 6 eyes and a macular hole in 1 eye. Peripheral retina examination showed small tears in four eyes, lattice degeneration in 13, giant retinal tear in two, chorioretinal atrophic scars in 15, vitreoretinal tufts in eight, retinal detachment in 5, and no retinal abnormalities in 13 eyes. Treatment was by observation in 40 eyes, laser retinopexy in 14 eyes, cryotherapy in one eye and combination of vitrectomy, laser retinopexy, cryotherapy and or sclera buckle in six eyes. All patients were followed up periodically and were asked to visit clinic early if they developed any symptoms of retina tear or detachment. Retinal tears developed in 16 fellow eyes were treated by laser retinopexy (15 eyes) and cryotherapy (1 eye) (Table 2). Prophylactic scleral buckle was performed on five eyes that had extensive lattice degeneration with or without associated retinal tears in the fellow eye. Retinal detachment developed in seven fellow eyes. The median time to develop the retinal detachment was 22.5 (range 5-123) months. Six eyes were treated by vitrectomy with sclera buckle and one eye with vitrectomy, silicon oil and laser retinopexy. The median duration of follow-up was 65.5 months (range 5-226) (Table 3).

Characteristic feature	Total (n=26 pts)
Total follow-up duration (mean, median; range), months	77, 65.5; 5-226
Visual acuity	
Better than 20/40	27
20/50-20/200	13
Worse than 20/200	6
Fixes and follows	4
Enucleated at presentation	2
Intraocular pressure (mean, median; range) mm Hg	15, 14; 6-27
Fundus examination	Eyes (n=52)
Lattice degeneration	4
Retina tear	2
Retina detachment (focal)	1
Treatment	
Laser	2
Scleral buckle	1
Observation	47

Table 3: Outcome at last follow-up visit, (n=26 pts, 52 eyes).

Five patients were lost to follow-up. Visual acuity was better than 20/40 in 27 eyes, between 20/50 to 20/200 in 13 eyes, worse than 20/200 in 6 eyes, fix/follows light in four eyes and two eyes were enucleated prior to initial presentation. The median intraocular pressure was 14 (range 6-27) mm Hg. Anterior segment examination was within normal limits in 25 eyes, band keratopathy in 1 eye, cataract in 4 eyes, pseudophakia in 8 eyes and aphakia in 12 eyes. Vitreous syneresis was present in 14 eyes, membranes in 5 eyes, veils and opacities in 5, posterior vitreous detachment in 4, clear in 2, empty in 20. Macula was normal in 32 eyes, pigment changes were seen in 17, detachment in 1 eye. Peripheral retina examination showed small tears in one, lattice degeneration in four, chorioretinal scars in 36, vitreoretinal tufts in 1, focal detachment in 2, and normal in 6. Treatment was by observation 47 eyes. Small retinal tears and lattice degeneration were treated by laser retinopexy in two eyes and by scleral buckle in one eye. None of the patients developed RD at the time of last FU. There were two patients who had extensive RD at initial follow-up, which became phthisical over years.

Discussion

The common ocular features in Stickler syndrome are vitreous degeneration at an younger age; absence of vitreous or vitreous veils and membranes in the anterior vitreous; peripheral retinal abnormalities such as lattice degeneration, retinal tears & holes, retinoschisis, chorioretinal degeneration; myopic degeneration changes; bilateral retinal detachment (RD) and rarely nuclear sclerosis cataract at younger age [1-7]. Nineteen of 31 patients in our study had features of high myopia. Ten of 62 eyes had cataract and almost all eyes had various vitreous abnormalities. Majority of our patients showed peripheral retinal abnormalities such as lattice degeneration, small retinal tears, giant retinal tears, chorioretinal atrophic scars, vitreoretinal tufts, or retinal detachment. In Stickler syndrome, retinal detachment is common cause for the guarded visual acuity. Families with high suspicion of retinal detachment need to be investigated for genetic abnormalities of SS [4-7]. In a study by Edwards and associates, 22 family members with high rates of RD tested genetically showed positive C192A COL2A1 exon mutations [4]. However, none of our patients had genetic results available, but the associated systemic and ocular features were highly suggestive of SS.

Early detection and treatment of vitreoretinal abnormalities in SS patients not only preserves vision but also prevents further vision threatening complications such as RD and phthisis bulbi [8-11]. Prophylactic treatment of retinal tears and detachment described in the literature were cyrotherapy, 360 degree laser retinopexy (barrier), vitrectomy in combination with cryo or laser therapy and scleral buckle [9-15]. Ang and associates performed a retrospective study on 204 patients with type I SS and reported the results of prophylactic cryotherapy in preventing retina detachment development [10]. Seventy three percent of patients in the control group (n=111) who did not receive any prophylactic ocular treatment developed RD. Whereas among the patients who had bilateral 360 degree cryotherapy (group 2, n=62), only 8% developed RD [10]. In their 3rd group (n=31), approximately 10% of patients developed RD in the fellow eye after prophylactic cryotherapy [10]. In our study, laser retinopexy and or cryotherapy was performed in 15 eyes, combined vitrectomy and laser retinopexy or cryotherapy and or sclera buckle was performed in 7 eyes. All of them were stable without development of any complications until the last follow-up. The effect of laser treatment to prevent development of RD in SS was reported by Leiba and associates in a retrospective study of 22 patients [11]. Ten patients of non-treated group (n=34 eyes) developed retina detachment. Whereas nine of the 10 eyes treated with prophylactic argon laser had attached retina at a follow-up period ranging from 1-15 years and one eye developed RD from a non-lasered lesion after 5 years. They summarized that prophylactic argon laser treatment in family members of SS with vitreoretinal abnormalities prevented the development of RD compared with those not treated with laser retinopexy [11].

Apart from the prophylactic laser retinopexy or cryotherapy, surgery also provided good anatomical and functional outcomes in patients with Stickler syndrome with RD. In a twenty year cohort study by Abeysiri and associates, of the 30 eyes that had RD, 9 eyes had scleral buckle with cryotherapy and 19 eyes had primary vitrectomy and 2 eyes underwent barrier laser retinopexy [12]. Following the above surgeries, the complete re-attachment rate at was 66.7%, 84.2%, 78.5% in primary scleral buckle, primary vitrectomy and all surgeries groups respectively. The study results also showed that approximately 33% of patients had increase in the average Logmar visual acuity after primary scleral buckle and vitrectomy surgeries [12]. In our study, five fellow eyes with extensive lattice degeneration with or without associated retinal tears were treated by prophylactic scleral buckle. None of them developed retinal tears or detachment until the last follow-up visit. However, at a median follow-up of 22.5 months, seven eyes developed RD, that was subsequently treated by vitrectomy and scleral buckle. Whereas 16 of the fellow eyes developed retinal tears which were treated by either cryotherapy or laser retinopexy. The visual acuity was stable among our patients after various treatment methods. In another cohort study by Billington and associates, of the 33 patients with Wagner-Stickler syndrome (36 eyes with multiple tears and RD), 27 RD's were successfully reattached following the scleral buckling (n=13) and vitrectomy (n=14) [13].

Our paper has its few limitations. First, we did not divide the patients into various groups for different surgical procedures unlike other studies, and did not compare the outcomes with each procedure. We also did not have genetic test results of each patient. There was no detailed data of the asymptomatic eyes from the old patients in the patient sheets.

In summary Stickler syndrome patients who present with multiple retinal tears, giant retinal tears or retinal detachment may benefit from prophylactic barrier laser retinopexy or cryotherapy and scleral buckle or combination therapies. It is important regularly follow-up the patients after the complex and multiple retinal surgeries to check for both for anatomic status of the retina and the visual outcome.

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