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Results of surgical management of the retinal detachment in pediatric patients with Marfan and Stickler syndrome

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ABSTRACT

BACKGROUND: The purpose of this study was to report the anatomical and functional outcomes in the surgical treatment of retinal detachment (RD) in pediatric patients suffering from Marfan (MFS) and Stickler syndrome (SS).
MATERIAL AND METHODS: Retrospective consecutive case series of eight eyes of four patients with SS and MFS. Retinal reattachment and visual acuity, as well as complications of surgical procedures have been evaluated during the follow-up period.
RESULTS: Total RD was reported in five of the eight eyes of these patients. In one patient, it was bilateral. As a surgical treatment, pars plana vitrectomy (PPV) with silicone oil was performed in four eyes. Proliferative vitreoretinopathy was the reason for the secondary PPV in three eyes. Scleral buckling was conducted in two eyes of bilateral RD. Laser treatment was performed as prophylaxis in all fellow eyes. The visual acuity in eyes with RD ranged from light perception to 0.1 in two cases. The retina was finally attached in all cases. Imaging with OCT-A in a patient with SS revealed a reduced vascular density in the eye after PPV compared to the eye treated with laser.
CONCLUSIONS: Multiple surgical procedures are needed to attach the retina in patients with SS and MFS. However, despite the improvement of anatomical conditions — reattachment of the retina, the visual acuity after RD surgery is not satisfactory in patients suffering from SS and MFS.

KEY WORDS: retinal detachment; pediatric patients; Marfan syndrome; Stickler syndrome

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INTRODUCTION

Marfan and Stickler syndromes are rare multisystem disorders with manifestations typically involving the skeletal and ocular systems.

Stickler syndrome (SS) is an inherited progressive disorder of collagen connective tissues described first in 1965 (Stickler, 1965). It may be expressed by various symptoms, e.g., orofacial defects (cleft palate, micrognathia, and midface flattening), conductive and/or sensorineural hearing loss, and ocular abnormalities (nonprogressive high myopia, vitreal abnormalities, and a high risk of retinal detachment

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(RD) [1]. Moreover, SS is the most commonly identified inherited cause of RD in childhood. The rate of RD, potentially leading to loss of vision, in patients with SS has been suggested to be as high as about 60% in one eye [2]; 8% percent of affected children have RD between the age of 0 and 9 years and 26% between the age of 10 and 19 years [3]. The pathognomonic feature of SS is a congenital abnormality in vitreous embryological development, which manifests as an abnormal architecture of the vitreous visible on slit lamp biomicroscopy [4].

Marfan syndrome (MFS) is an autosomal dominant genetic connective tissue disorder that results from *de novo* or inherited mutations in the fibrillin-1 gene (FBN1, MMI 134797) [5]. Mutations in the genes for fibrillin-1 (FBN1) result in multiple distinct pleiotropic disorders. These genetic disorders demonstrate that specific domains of fibrillin-1 perform roles important to musculoskeletal growth. A disproportionate linear bone growth that causes serious malformations of the limbs, spine, and anterior chest wall is the most striking and immediately evident manifestation in MFS patients [6–8].

The diagnosis of MFS depends on the clinical evaluation, family history, and molecular data in accordance with the Ghent criteria [9] revised in 2010. Many individuals diagnosed with the disorder do not carry the FBN1 mutation, which resulted in the establishment of revised criteria that were more specific and stringent. The zonular defect leads to ectopia lentis, which is a hallmark of Marfan ocular abnormalities, and occurs in 60% to 80% of cases. Other less common ocular features of MFS are increased axial length and axial myopia. The RD prevalence in MFS ranges from 5 to 25% and increases to 8–38% in those with ectopia lentis or patients that have undergone cataract surgery. Most patients develop RD at a young age in both syndromes [10], and usually, they require multiple surgeries [11].

The aim of this study is to present a case series of patients with Stickler and Marfan syndrome and to analyze the results of different surgical procedures performed due to RD.

CASE PRESENTATIONS

Case 1

An 18-year-old male patient was admitted to the ophthalmology department with complaints of loss of vision in the right eye for the last few days. His short stature (150 cm), short midface, retrognathia, and high myopia revealed the clinical diagnosis of SS (Fig. 1).

FIGURE 1. General appearance of patient 1 with clinical features of Stikler syndrome — short stature, short midface, and retrognathia

Best-corrected visual acuity (BCVA) was 0.05 in the right eye and 0.1 in the left eye. The patient had been diagnosed with SS before and had already undergone a lensectomy in both eyes due to a congenital cataract. Additionally, he had also undergone a complete scleral buckling procedure in both eyes due to RD in the past. During the examination, total RD was detected in the right eye. During hospitalization in the Ophthalmology Department, he received complete PPV (Alcon, Constellation, Fort Worth, US) with 5000 centistokes silicone oil tamponade and scleral fixation of the intraocular lens (IOL) implantation, as the postoperative aphakia was present. At 6 months after the surgery, the right eye visual acuity was assessed as only light perception, although the retina was anatomically reattached.

Case 2

A 13-year-old patient was admitted to our hospital due to the sudden loss of vision in the left eye. His flat face, short stature (145 cm), dispropor-

FIGURE 2. General appearance (**A**) of patient 2 with clinical features of SS — short stature, flat midface, and short limbs. Anterior segment (**B**) of the left eye of patient 2 at the end of treatment: pseudophakia and Ahmed valve

tionately short limbs, and ocular findings suggested a diagnosis of SS (Fig. 2).

BCVA during examination in the emergency room was 0.3 (20/63) cc –7.0 D in the right eye and 0.05 (20/400) cc –7.0 D in the left eye. The fundus examination showed total RD in the left eye and high myopic degeneration in both eyes. The patient received PPV with silicon tamponade with intraocular lenses (IOL) implantation in the left eye. The visual acuity on the first day after this procedure was 0.1 (20/200) with a correction +3.0 D in the left eye.

After 2 weeks, high intraocular pressure (IOP) (34 mm Hg) was noticed in the left eye. Using local and systemic treatment (beta blocker eye drops, both eye drops, and oral carbonic anhydrase inhibitors), IOP decreased to 19 mm Hg. In the right eye, extensive degenerations were diagnosed in the periphery of the retina, and laser treatment was applied.

After six months of the follow-up, a fundus examination showed recurrent RD in the left eye. The patient received PPV with 5000 centistokes silicone oil exchange. During subsequent control visits, IOP in the left eye was still raised, and the pharmacological therapy was not satisfactory. Thus, an XEN implant (a stent that imitates the principle of trabeculectomy by generating subconjunctival filtration) was implemented to achieve effective pressure reduction.

After a few weeks, IOP was again raised in the left eye, and revision of the implant was needed. Some pressure reduction was sufficient for about a month, but the left eye's high pressure returned to 50 mm Hg. The patient received the Ahmed implant, which reduced IOP to the satisfying level of 18 mm Hg. Five years after PPV, BCVA was 0.1 (20/200) in the left eye due to RD. In the right eye, only laser treatment has been applied.

At the end of the follow-up, the retina was attached in both eyes (Fig. 3), and both eyes' wide-field fundus fluorescence (Fig. 4) revealed a predominantly hypofluorescent pattern with a hypofluorescent photocoagulation scars and areas of atrophy. In the right eye, hypofluorecent lesions along vessels and in the periphery were located on the opposite side of the laser treatment area. In the optical coherence tomography (Fig. 5) there was inverted foveal contour with slight epiretinal membrane in the right eye after laser treatment and thinning of the retina in the eye with silicone oil after vitrectomy.

Case 3

A 12-year-old girl was admitted to our hospital with a visual acuity of 1.0 cc (20/20)+13 Dsph in both eyes. Her height (182 cm), deformed chest due to scoliosis, and long fingers indicated connective tissue disorders characteristic of MFS (Fig. 6). Lensectomy was done in the past in both eyes because of lens subluxation, which is a "big" diagnostic feature of MFS according to Ghent nosology 9. Moreover, there was a positive family history of MFS.

We offered the patient secondary IOL implantation to the right eye. After surgery, the patient had BCVA of 0.7 in the right eye. Later, secondary IOL was also implanted in the left eye. BCVA was 1.0

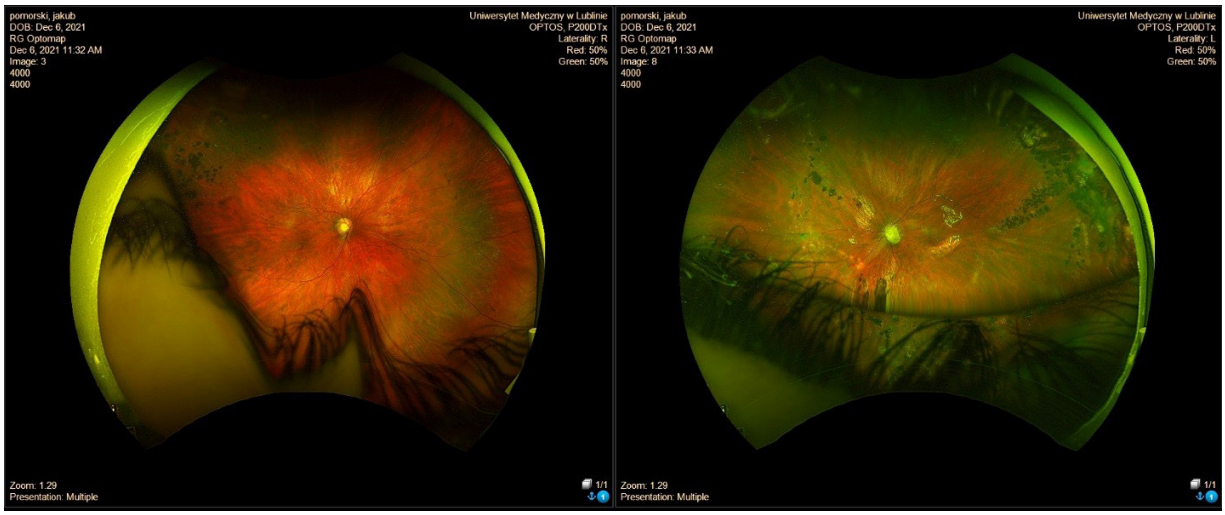

FIGURE 3. Ultra wide-field fundus photos (Optos) of the fundus of both eyes of patient no 2 with SS. In the right eye, laser spot in the inferio-temporal quadrant, and in the left eye, silicone oil, enlarged excavation of the optic disc due to secondary glaucoma

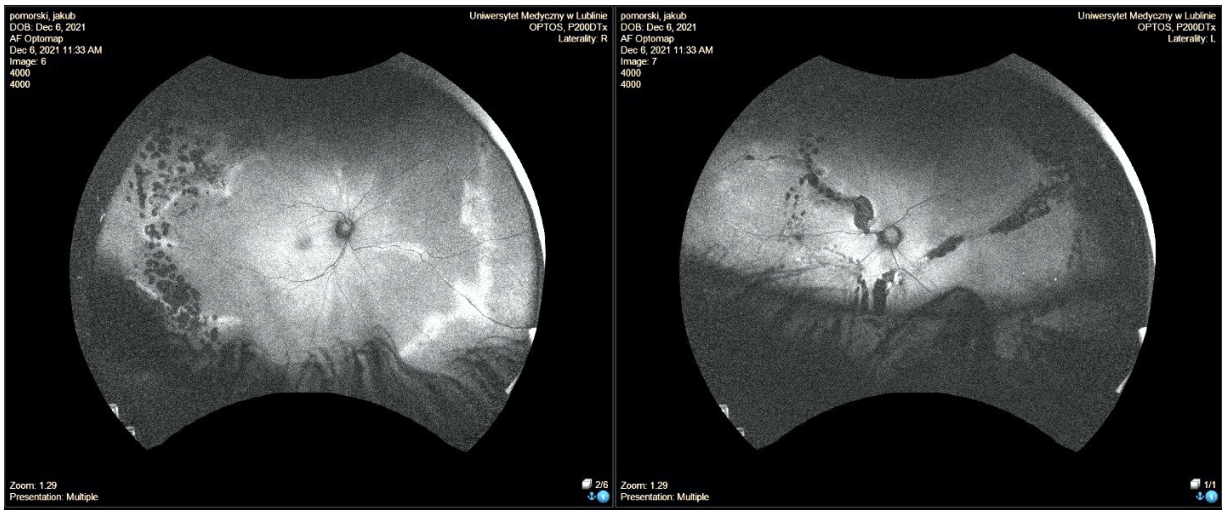

FIGURE 4. Ultra wide-field fundus autofluorescence images of both eyes of patient no 2 with Stickler syndrome representing a hyperfluorescent pattern with hypofluorescent photocoagulation scars and areas of atrophy. In the right eye, hypofluorecent lesions along vessels and in the periphery on the opposite side to the laser treatment area

in the right eye, and 0.9 was achieved in the left eye. Seven months later, the patient returned with complaints of loss of vision in the left eye. YAG capsulotomy did not give a satisfactory result because of the persistence of "cloudiness" on the IOL surfaces. Anterior vitrectomy with IOL cleaning was applied. After a few days, the fundus examination showed total RD in the left eye. The patient received complete PPV with 5000 centistokes silicone oil. After a few months, this procedure had to be repeated because of the existing redetachment with PVR. At the last follow-up visit, the visual acuity of the left eye was 0.05 (20/400). Prophylactic laser treatment was performed in the fellow eye.

Case 4

A 15-year-old boy with diagnosed MFS (based on ascending aorta dilatation and ectopia lentis) and Axenfeld-Rieger's anomaly was referred to our department because of corectopia in the left eye (Fig. 7). The patient received surgery for the iris in the left eye. After the surgery, BCVA was 0.7 cc +3.0 ax 60° in the right eye and 0.1cc –1.5/+4.0 ax 105° in the left eye.

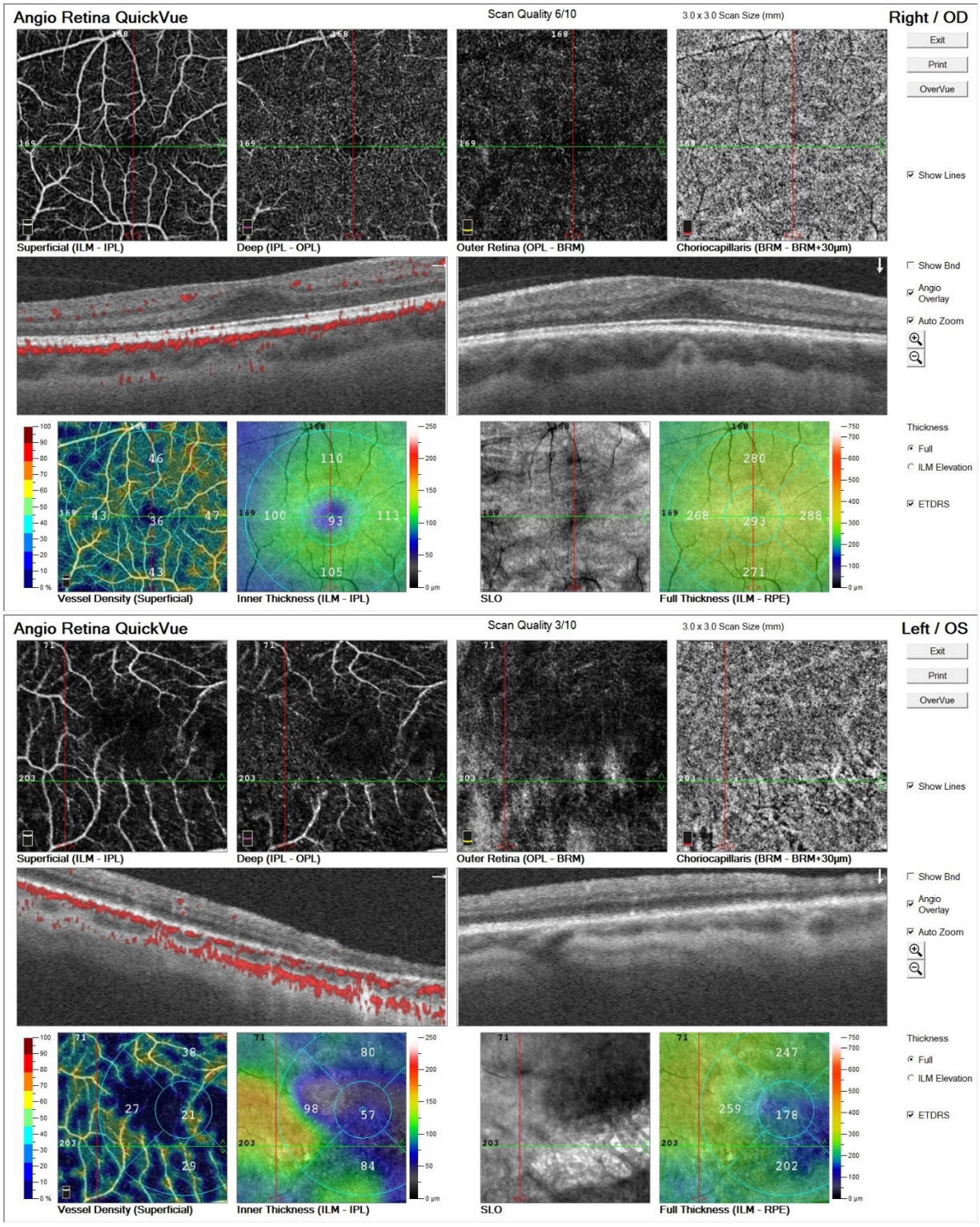

FIGURE 5. Optical coherence tomography (OCT) and optical coherence tomography angiography (OCT-A) (Optovue) images of both eyes (right eye in the upper part, left eye — in the lower part) of patient no 2 with Stickler syndrome. Right eye after laser treatment (inverted foveal contour, slight epiretinal membrane), left eye after vitrectomy with silicone oil (thinned retina). OCT-A reveals decreased vessel density and decreased full thickness in the left eye

After that, the patient returned with complaints of worse visual acuity in the left eye (0.05). The fundus examination and B scan ultrasonography confirmed the diagnosis of total RD in the left eye.

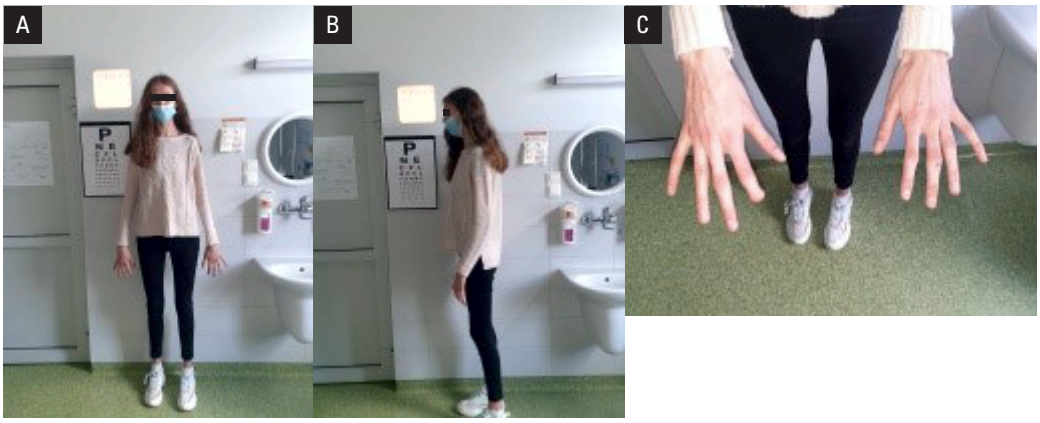

FIGURE 6. The general appearance of the patient 3: 182 cm high, long limbs, deformed chest (**A, B**), and arachnoidal fingers (**C**)

FIGURE 7AB. General appearance of patient 4: over 180 cm high, long limbs. Anterior segment of the left eye: pseudophakia, anterior capsule fibrosis, anterior adhesions, silicone oil under the conjunctiva, status post iris plastic surgery

The patient underwent complete PPV with 5000 centistokes silicone oil tamponade with ILM peeling combined with phacoemulsification and implanting IOL in the left eye. As a result, BCVA was 0.2 with correction +3 D. After 3 weeks at the follow-up visit, the visual acuity in the left eye dropped to 0.05, and we discovered recurrent RD in the left eye. Urgent complete PPV with 360 degrees of retinotomy, ILM peeling, and silicone oil tamponade was performed in the left eye, but the visual acuity was not improved. Laser treatment was performed in the right eye in the periphery.

DISCUSSION

Patients with connective tissue disorders present significant challenges in the surgical treatment related to abnormal vitreoretinal adhesions [12]. The presence of multiple breaks or giant retinal tear [13], thin sclera, miotic pupils [14], and liquefied abnormal vitreous complicates the management of these patients. The vitreous cavity of these patients is only partially filled with gel vitreous from birth [15], and the vitreous gel adheres to the retina in a grossly anomalous and unpredictable fashion. Moreover, RD is complex and consistently difficult to manage in patients with Stickler and MFS, with a lower success rate of repair compared with the typical degenerative RRD in other pediatric patients with RD [16].

Studies performed on the pediatric population indicate that anatomical retinal reattachment can be achieved in 70–80% of cases. Thus it is lower than in adult patients [17]. This success rate highly depends on the number of retinal clock hours involved and whether previous retinal surgery has been performed [18]. Successful surgery treatment in these cases is strongly connected with phenotype/genotype correlations between the mutation types. Such correlations exist in MFS, for example, with ectopia

Table 1. Surgical procedures performed in two patients with Stickler syndrome (SS) and two patients with Marfan syndrome (MFS)

Case/ disease	Gender	Age of the first surgery	Age of onset	Surgical procedures of the right eye	Surgical procedures of the left eye	Final visual acuity of the right eye (Snellen charts)	Final visual acuity of the left eye (Snellen charts)	Follow-up period
Patient 1/SS	Male	18		Lensectomy Scleral buckling PPV + silicone oil tamponade	Lensectomy Scleral buckling	Light perception	0.1	2 years
Patient 2/SS	Male	13		Laser treatment	Phacoemusification + IOL implantation + PPV + silicone oil tamponade PPV + silicone oil exchange Xen implant Ahmed valve	0.3 cc – 7.0 D	0.1 cc + 3.0 D	5 years
Patient 3/MFS	Female	12		Lensectomy Laser treatment	Lensectomy Artisan lens PPV + silicone oil tamponade PPV + silicone oil exchange	1.0 cc + 13.0 D	0.05cc + 3.0 D	2 years
Patient 4/MFS	Male	15		Laser treatment	Iris plastic surgery Phacoemulsification+ IOL implantation + PPV + silicone oil tamponade PPV + silicone oil exchange	0.7 cc + 3.0 D ax 60	0.05 cc + 3.0 D	2 years

PPV — pars plana vitrectomy; IOL — intraocular lenses

lentis much more frequently observed than in other disorders. The prognosis for retinal reattachment in these cases should be studied or established [19, 20]. In our case series, the patients had the clinical diagnosis of MFS and SS based on the phenotype features without genotyping confirmation [21].

It has already been observed that patients with MFS with a normal or subluxated lens without any interference with fundoscopy or the retinal breaks are at or anterior to the equator should undergo scleral buckling.

Vitreoretinal surgery is recommended for patients with a history of failed scleral buckling, posterior lens dislocation, and subluxated or cataractous lens with interference with fundoscopy and giant retinal tears [22]. Surgery failure in MFS is mainly due to proliferative retinopathy and poor visualization of the retinal periphery [23].

In an extensive series of MFS patients, it has been reported that 70% of 160 patients with RD were below the age of 20 years. Bilateral RD is common and may reach 70% [3]. Predisposing factors for retinal breaks in MFS include ectopia lentis, long axial length, early vitreous liquefaction, posterior vitreous detachment without any dehiscence at the vitreoretinal interface, and abnormal peripheral vitreoretinal adhesions [18]. RD may occur spontaneously in eyes with axial myopia or following cataract extraction, especially in longer eyes. Maumenee found that 21% of the eyes of patients with MFS had myopia of 7 D or more, astigmatism, and a flat cornea [1].

Loewenstein et al. identified and retrospectively reviewed the charts of one cohort of 12 patients (15 eyes) with MFS and RD who were operated on at the Wilmer Institute and the second cohort of 16 such patients (24 eyes) who were operated on several years earlier elsewhere. They concluded that the results of the RD surgery in the past were worse when the eye was aphakic. In most cases operated more recently, the prognosis for successful repair was good regardless of whether the eye was phakic [24]. In another study, Abboud et al. found bilateral RD in 9 of 13 (69%) patients. The lens was ectopic in all eyes. The retinal breaks were small horseshoe tears or holes located anterior to the equator in 11

of 16 (69%) eyes. The retina of 12 of 16 (75%) eyes remained successfully reattached after a follow-up ranging from 4 to 132 months. All 12 eyes had a VA of 20/300 or better (range: 20/30 to 20/300); 8 had a VA of 20/125 or better, and the cause of failure in the other 4 eyes was proliferative vitreoretinopathy [25].

Compared to adults, pediatric RDs have higher rates of macula-off detachment, proliferative vitreoretinopathy, and worse presenting visual acuity [17]. In one series, about 40% were related to trauma, whereas retinopathy of prematurity, MFS, and SS accounted for up to 50% of cases. This was confirmed by Dotrelova and colleagues in thirteen patients (18 eyes) with MFS who underwent surgery for RD. The characteristic findings were as follows: RD in three or more quadrants (12 eyes) and advanced proliferative vitreoretinopathy (PVR) (7 eyes). Nine uncomplicated RDs were managed with scleral buckling, and nine complicated RDs were managed with PPV, scleral buckling, and retinal tamponade, mostly with silicone oil. Complete retinal reattachment was achieved in 89% of uncomplicated RDs and in 56% of complicated RDs. Additional partial anatomic success was achieved in two eyes with complicated RDs where the macula was attached. Visual acuity improved significantly in five eyes with uncomplicated RDs (median final vision 20/80) and in six eyes with complicated RDs (median final vision, 20/200) 23. In our case series of four patients with Marfan and SS, all the patients had total RD, as reported by Dotrelova and coworkers18. Regarding visual acuity, in a study of 206 patients with pediatric DR, SS accounted for 4.3% and 70% of patients had visual acuity worse than 0.1, and the recurrence was resent in 80% of operated eyes [11].

Functional results of the retinal surgery in Stickler and Marfan syndrome obtained in our study are very poor. The worst results were found in a patent with bilateral RD and SS (patient no 1). He had recurrent RD instead of scleral buckling. According to a recent large systematic literature review, visual impairment is rare in SS (blind: 6%; vision loss in one eye: 10%) [26]. It seems that the retina is only detached but also intrinsically damaged in patients with connective tissue disorders. Possibly, there is existing photoreceptor's or other retinal layer's damage, or the retinal cells are very prone to surgical interventions. In the multimodal imaging obtained in one patient with SS (patient no 2), we observed thinned retina and decreased retinal vascularization in the eye after PPV with silicone oil. Predominantly hypofluorescent fundus autofluorescence images with hypofluorescent lesions along vessels and in the periphery in the untreated with PPV are similar to those described by Fujimoto in the large series of SS patients [27].

In our study, we reported the final anatomical attachment of the retina in all eyes of the patients with SS and MFS. Lee et al. [28] reported the same results in a recently published study. Successful reattachment was achieved in 28 of 29 eyes (97%) with RD due to SS with an average of 2.3 surgeries (including silicone oil removal surgeries). In our case series, if a vitrectomy was performed, there was a need for silicone oil tamponade in all cases. In a study by Read et al. [11], SB was performed in 40% of cases, SB with PPV and gas in 30%, and SB with PPV and silicone oil in 30% of pediatric cases.

Vitreoretinal surgeons have great difficulties in achieving retinal reattachment in patients with connective tissue disorders, and it is even possible that the functional success is very low. In view of these challenges, some researchers have emphasized the importance of prophylactic laser treatment to the retina in patients with SS to reduce the occurrence of and/or prevent RD [2]. In our case series, laser treatment was applied to all fellow eyes. It is more important as we consider that RD in patients with MFS tends to occur bilaterally (30–42% of cases) [29]. The prophylactic treatment purportedly creates firm chorioretinal adhesions and thus potentially prevents the development of retinal tears and subsequent RD. It is common practice that patients with SS undergoing surgical repair also undergo indirect barrage laser treatment of the companion eye even without the apparent disease. Three retrospective studies have addressed the benefits and efficacy of prophylactic laser treatment to prevent RD in SS type I. All three studies, in which either 360 peripheral cryotherapy or argon laser photocoagulation were used, reported statistically significant beneficial long-term outcomes and good safety profiles [30–32]. Thus, there is a need for screening in children diagnosed with MFS and SS and a need for prophylactic treatment in the unaffected eye.

To conclude, different surgical strategies can be applied in patients with SS and MFS; however, the functional results are unsatisfactory after multiple PPV. The patients' phenotypes are very complex, and although success was not secured upfront, we tried to improve the patients' quality of life.

REFERENCES

1. Maumenee IH, Traboulsi EI. The ocular findings in Kniest dysplasia. Am J Ophthalmol. 1985; 100(1): 155–160, doi: 10.1016/s0002-9394(14)74998-0, indexed in Pubmed: 4014370.
2. Carroll C, Papaioannou D, Rees A, et al. The clinical effectiveness and safety of prophylactic retinal interventions to reduce the risk of retinal detachment and subsequent vision loss in adults and children with Stickler syndrome: a systematic review. Health Technol Assess. 2011; 15(16): iii–xiv, 1, doi: 10.3310/hta15160, indexed in Pubmed: 21466760.
3. Yi'an GN, Wai-Ching L. Pediatric retinal deteachment. Taiwan J Ophthalmol. 2018; 8(4): 222–36.
4. Snead MP, McNinch AM, Poulson AV, et al. Stickler syndrome, ocular-only variants and a key diagnostic role for the ophthalmologist. Eye (Lond). 2011; 25(11): 1389–1400, doi: 10.1038/eye.2011.201, indexed in Pubmed: 21921955.
5. Dietz HC, Cutting GR, Pyeritz RE, et al. Marfan syndrome caused by a recurrent de novo missense mutation in the fibrillin gene. Nature. 1991; 352(6333): 337–339, doi: 10.1038/352337a0, indexed in Pubmed: 1852208.
6. Smaldone S, Clayton NP, del Solar M, et al. Fibrillin-1 Regulates Skeletal Stem Cell Differentiation by Modulating TGF Activity Within the Marrow Niche. J Bone Miner Res. 2016; 31(1): 86–97, doi: 10.1002/jbmr.2598, indexed in Pubmed: 26189658.
7. Lee B, Godfrey M, Vitale E, et al. Linkage of Marfan syndrome and a phenotypically related disorder to two different fibrillin genes. Nature. 1991; 352(6333): 330–334, doi: 10.1038/352330a0, indexed in Pubmed: 1852206.
8. Judge D, Dietz H. Marfan's syndrome. Lancet. 2005; 366(9501): 1965–1976, doi: 10.1016/s0140-6736(05)67789-6, indexed in Pubmed: 16325700 .
9. Loeys BL, Dietz HC, Braverman AC, et al. Revised diagnostic criteria for the Marfan syndrome. Am J Med Genet. 1996; 62(4): 417–426, doi: 10.1002/(SICI)1096-8628(19960424)62:4<417::AID-AJMG15>3.0.CO;2-R, indexed in Pubmed: 8723076.
10. Esfandiari H, Ansari S, Mohammad-Rabei H, et al. Management Strategies of Ocular Abnormalities in Patients with Marfan Syndrome: Current Perspective. J Ophthal Vis Res. 2019; 14(1): 73, doi: 10.4103/jovr.jovr_29_18.
11. Read SP, Aziz HA, Kuriyan A, et al. Retinal Detachment Surgery in a Pediatric Population: Visual and Anatomic Outcomes. Retina. 2018; 38(7): 1393–1402, doi: 10.1097/IAE.0000000000001725, indexed in Pubmed: 28858062.
12. Edwards A, Robertson J. Hereditary Vitreoretinal Degenerations. Retina. 2012: 836–851.
13. Yonekawa Y, Haddock LJ, Eliott D. A 360° giant retinal tear in Stickler syndrome. JAMA Ophthalmol. 2015; 133(1): e141812, doi: 10.1001/jamaophthalmol.2014.1812, indexed in Pubmed: 25569319.
14. Nahum Y, Spierer A. Ocular features of Marfan syndrome: Diagnosis and management. Israel Med Assoc J. 2008; 10: 179–81.
15. Watanabe Y, Ueda M, Adachi-Usami E. Retinal detachment in identical twins with Stickler syndrome type 1. Br J Ophthalmol. 1996; 80(11): 976–981, doi: 10.1136/bjo.80.11.976, indexed in Pubmed: 8976725.
16. Gonzales CR, Singh S, Yu F, et al. Pediatric rhegmatogenous retinal detachment: clinical features and surgical outcomes. Retina. 2008; 28(6): 847–852, doi: 10.1097/IAE.0b013e3181679f79, indexed in Pubmed: 18536601.
17. Rejdak R, Nowakowska D, Wrona K, et al. Outcomes of Vitrectomy in Pediatric Retinal Detachment with Proliferative Vitreoretinopathy. J Ophthalmol. 2017; 2017: 8109390, doi: 10.1155/2017/8109390, indexed in Pubmed: 28840039.
18. Smith JM, Ward LT, Townsend JH, et al. Rhegmatogenous Retinal Detachment in Children: Clinical Factors Predictive of Successful Surgical Repair. Ophthalmology. 2019; 126(9): 1263–1270, doi: 10.1016/j.ophtha.2018.11.001, indexed in Pubmed: 30419297.
19. Latasiewicz M, Fontecilla C, Millá E, et al. Marfan syndrome: ocular findings and novel mutations-in pursuit of genotype-phenotype associations. Can J Ophthalmol. 2016; 51(2): 113–118, doi: 10.1016/j.jcjo.2015.12.019, indexed in Pubmed: 27085269.
20. Li Y, Xu J, Chen M, et al. A FBN1 mutation association with different phenotypes of Marfan syndrome in a Chinese family. Clin Chim Acta. 2016; 460: 102–106, doi: 10.1016/j.cca.2016.06.031, indexed in Pubmed: 27353645.
21. 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.
22. Lee SY, Ang CL. Results of retinal detachment surgery in Marfan syndrome in asians. Retina. 2003; 23(1): 24–29, doi: 10.1097/00006982-200302000-00004, indexed in Pubmed: 12652227.
23. Dotrelova D, Dotrelova D, Karel I, et al. Retinal detachment in Marfan's syndrome. Characteristics and surgical results. Retina. 1997; 17(5): 390–396, doi: 10.1097/00006982-199709000-00006, indexed in Pubmed: 9355186.
24. Loewenstein A, Barequet I, Juan EDe, et al. Retinal detachment in Marfan syndrome. Retina. 2000; 20(4): 358–363, doi: 10.1097/00006982-200004000-00006, indexed in Pubmed: 10950412.
25. Abboud E. Retinal detachment surgery in Marfan's syndrome. Retina. 1998; 18(5): 405–409, doi: 10.1097/00006982-199805000-00003, indexed in Pubmed: 9801033.
26. Boysen KB, La Cour M, Kessel L. Ocular complications and prophylactic strategies in Stickler syndrome: a systematic literature review. Ophthalmic Genet. 2020; 41(3): 223–234, doi: 10.1080/13816810.2020.1747092, indexed in Pubmed: 32316871.
27. Fujimoto K, Nagata T, Matsushita I, et al. Ultra-wide field fundus autofluorescence imaging of eyes with Stickler syndrome. Retina. 2021; 41(3): 638–645, doi: 10.1097/IAE.0000000000002879, indexed in Pubmed: 32639332.
28. Lee AC, Greaves GH, Rosenblatt BJ, et al. Long-Term Follow-Up of Retinal Detachment Repair in Patients With Stickler Syndrome. Ophthalmic Surg Lasers Imaging Retina. 2020; 51(11): 612–616, doi: 10.3928/23258160-20201104-02, indexed in Pubmed: 33231693.
29. Dotrelova D. Bilateral retinal detachment in Marfan's syndrome. Eur J Ophthalmol. 1998; 8(2): 102–105, indexed in Pubmed: 9673479.
30. Ang A, Poulson AV, Goodburn SF, et al. Retinal detachment and prophylaxis in type 1 Stickler syndrome. Ophthalmology. 2008; 115(1): 164–168, doi: 10.1016/j.ophtha.2007.03.059, indexed in Pubmed: 17675240.
31. 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.
32. 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.