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Optometry's Role in Diagnosing and Managing Patients with Stickler Syndrome

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INTRODUCTION

Stickler Syndrome (SS) is a hereditary multisystem collagen disorder with ocular, auditory, craniofacial, and musculoskeletal manifestations. The most common and earliest ocular finding is childhood myopia, occurring in 80-90% of SS patients, with 40% of those having a severe refractive error of -10.00 diopters or worse. Retinal detachments (RD) have been reported to occur in 50-70% of SS patients. Pre-senile cataracts can also be present (36-59%) with both nuclear and quadrant lamellar cataracts that spare the visual axis described. Less common ocular manifestations include glaucoma (11%), either secondary to RD surgery or, more rarely, infantile-onset glaucoma. Due to the high rates of ocular signs in SS, eye care professionals play a vital role in the health care team. Particularly, optometrists can fit these pediatric patients with contact lenses to improve visual quality.

CASE PRESENTATION

A 15-year-old male with Stickler Syndrome was referred for a specialty contact lens fitting. He had been wearing glasses for most of his life with symptoms of blurry vision OU.

Ocular History

- degenerative myopia OU
- lattice degeneration s/p 360-degree prophylactic barricade laser OU (Figure 1)
- quadrant cortical cataracts, not within the visual axes OS>OD
- glaucoma, managed with latanoprost qhs OU by ophthalmology

Manifest Refraction

OD: -22.00 -1.25 x180 VA 20/80
OS: -24.25 -0.75 x180 VA 20/60

Corneal Topography (Figure 2)

OD: Keratometry 42.0D / 44.0D @107, HVID 11.3mm
OS: Keratometry 42.3D / 43.6D @093, HVID 11.3mm

MANAGEMENT

The patient was determined to be a good candidate for corneal gas permeable (GP) contact lenses and was fit diagnostically. Table 1 shows the final contact lens order, resulting in improved visual acuity to 20/25- OD/OS.

However, after 8 months of wear, the patient complained of discomfort with GPs and only wore the lenses 1-2 days per week for about 8 hours per day. He was happy with the improved visual quality but was unable to tolerate wearing the lenses daily. The patient was scheduled for a refit into scleral contact lenses, which has not been completed at the time of publication.

FIGURE 1

Optos fundus photography shows prophylactic barricade laser OU to treat degenerative myopia, and an artifact OS due to quadrant cataract.

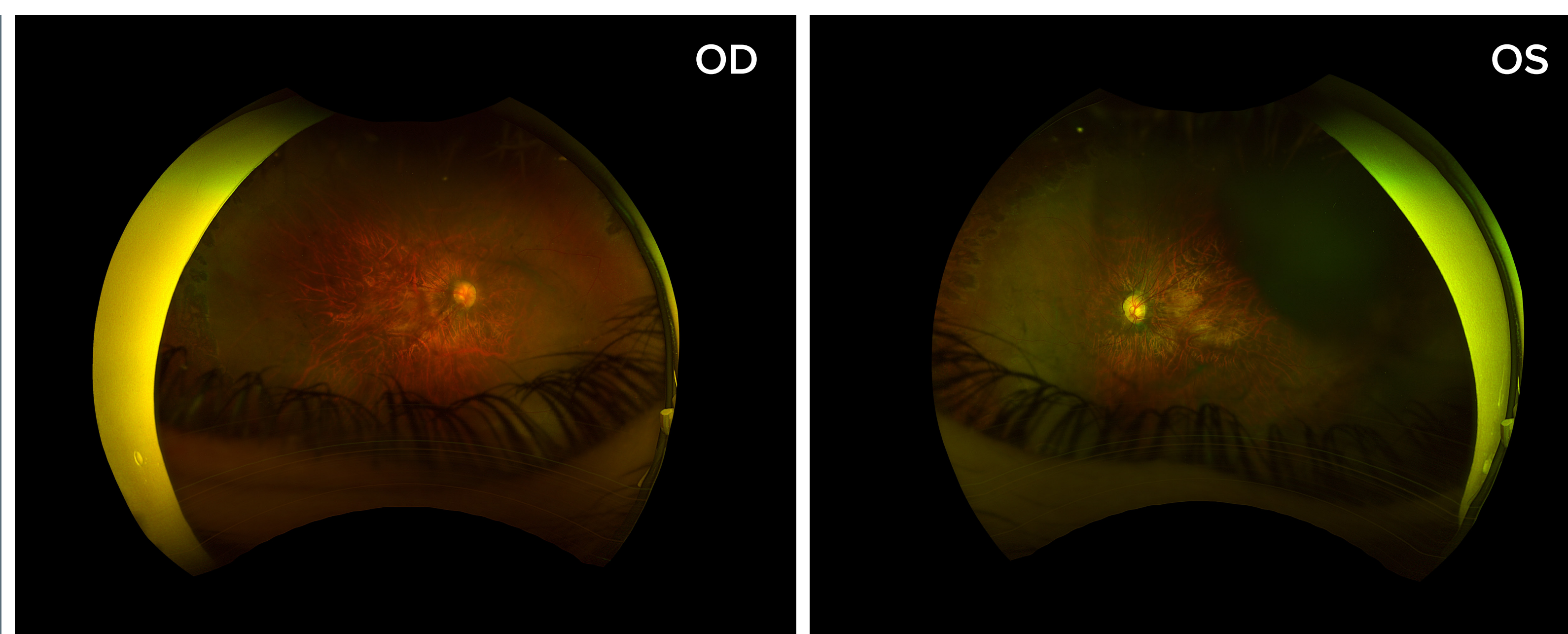


FIGURE 2

Axial topography maps reveal regular with-the-rule (WTR) astigmatism OU.

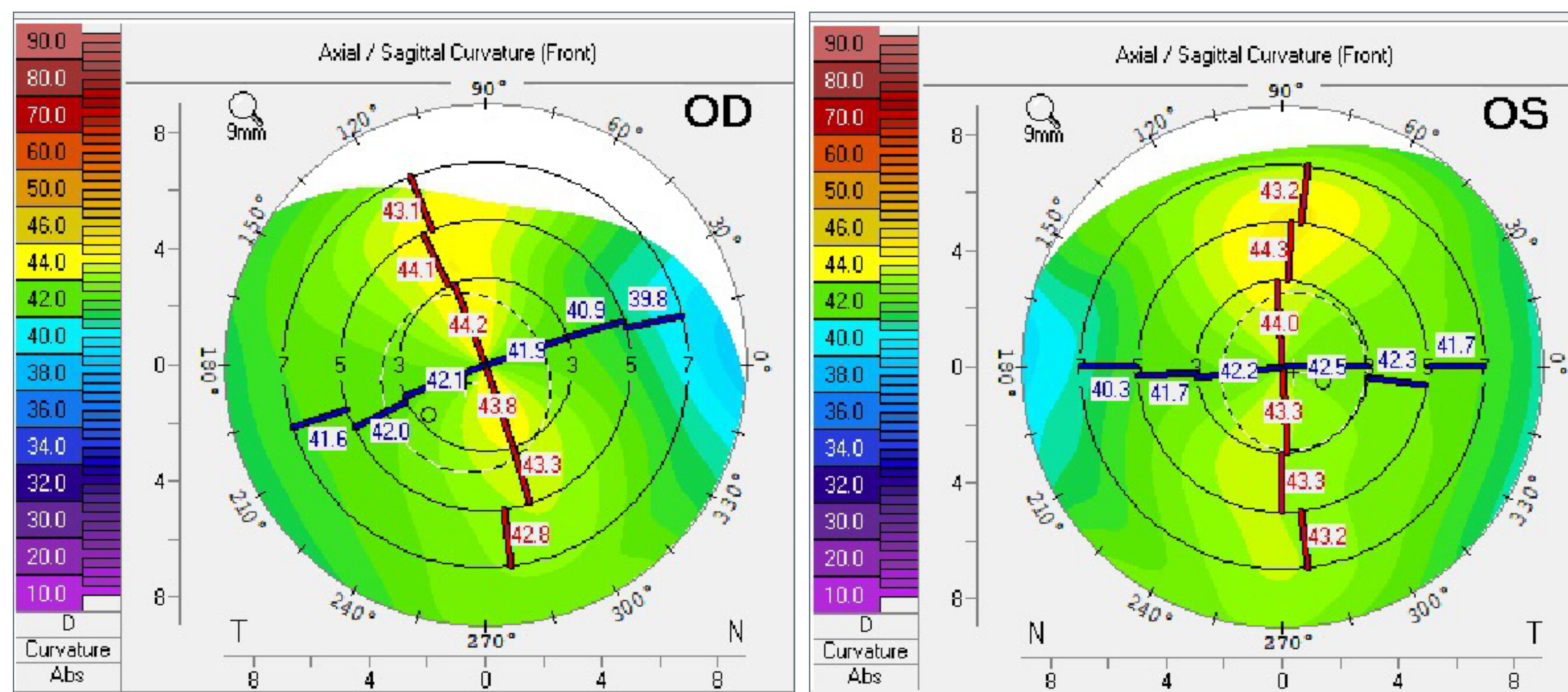


TABLE 1

Contact Lens Parameters

	Design	Material	Power	BC	Dia	Edge	VA
OD	Spherical GP	Boston XO	-22.50	8.08	9.2	TPC (vertical steep)	20/25-
OS	Spherical GP	Boston XO	-21.25	7.80	9.2	TPC (vertical steep)	20/25-

TABLE 2

Ocular Manifestations of Stickler Syndrome

Congenital myopic refractive error (> -3.00 D)
Retinal detachment (1st and 2nd decades of life)
Cataracts (nuclear and quadrant lamellar)
Glaucoma

CONCLUSION

Stickler Syndrome is commonly encountered at a young age and eye care professionals may be the first to diagnose this condition due to the many ocular manifestations (Table 2). Optometrists are essential members of the health care team and can monitor for RD, glaucoma, and cataracts in conjunction with ophthalmology, referring for surgical intervention when warranted. Prophylactic laser and cryotherapy can be considered to prevent RD.

This case promotes awareness of Stickler Syndrome as a condition that often results in degenerative myopia. The unique role of optometry is to manage SS patients with contact lenses to rehabilitate vision.

Furthermore, some children with Stickler Syndrome may exhibit ocular findings without other systemic involvement (auditory, craniofacial, and musculoskeletal findings). Therefore, molecular genetic testing for SS should be considered when examining a young patient with high myopic refractive error and other comorbid findings such as RD, glaucoma, or congenital cataracts.

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