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# A Unique Ophthalmologic Presentation of Stickler Syndrome

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## Abstract

**Purpose:** Stickler syndrome (STL) is an extremely rare multisystem collagenopathy characterized by auditory, ocular, musculoskeletal, and orofacial abnormalities. In this case report we present a unique ophthalmologic presentation in a patient with Stickler syndrome.

**Observations:** We report a case of a 66-year-old female with STL with several associated right retinal detachments status post retinectomy with membrane peel in the right eye who prophylactically underwent laser retinopexy of the left eye. In addition to retinal detachment, she has been treated for bilateral vertical diplopia, allergic dermatitis, dry eye syndrome, and bilateral punctate keratitis. It is unclear whether these additional ocular findings could be manifestations of her previously diagnosed STL.

**Conclusions and Importance:** STL is an exceedingly rare but devastating multisystem collagenopathy with often significant visual implications. Early detection and diagnosis are essential for improved visual outcomes. This case should raise the awareness of variable clinical presentations, treatment, and follow up of patients with STL.

**Keywords:** Stickler syndrome; Laser retinopexy; Retinal detachment

## Introduction

Stickler syndrome (STL) is a rare autosomal dominant genetic condition, affecting 1:7500-9000 neonates, that reduces the ability of collagen to crosslink [1]. STL can present with several clinically distinct presentations including: ocular pathologies, hearing abnormalities, craniofacial anomalies, and joint problems [2]. The diagnosis of STL is typically made through the observation of the myriad of clinical symptoms in addition to genetic analysis of various pathogenic collagen subtypes including collagen 2, 9, and 11 [2]. Of particular ophthalmologic significance, STL most commonly presents with ocular abnormalities [3]. This places ophthalmologists on the front line in diagnosis of this rare genetic disease. Some of the possible ocular presentations include myopia, cataracts, and retinal detachment [4].

STL is the most common cause of pediatric genetically determined retinal detachment, and carries a significant lifetime risk of subsequent detachment [5]. The treatment for ophthalmological sequelae of STL usually involves correction of myopia with refractive glasses, and surgical intervention in the treatment of retinal detachment [4]. This case is important as it highlights that STL, though typically diagnosed in the earlier years of life, can present and be diagnosed well into adulthood. This case also presents with several atypical STL characteristics, including dry eye syndrome, punctate keratitis, and diplopia.

## Case Presentation

This is an investigation into a 66-year-old woman with a history of STL confirmed by genetic testing, with an additional history of hypertension, asthma, type 2 diabetes mellitus, hypercholesterolemia, rheumatoid arthritis, and epilepsy. Since 2010, she has been followed by optometry and retina specialists for numerous supplementary complex ocular disorders, including cataract, myopia, diplopia, allergic dermatitis, and dry eye syndrome of the lacrimal glands in addition to her associated retinal disease.

During 2010, at the age of 56, she experienced three separate instances of retinal detachments of the right eye. Initially, treatment involved scleral buckle and pars planar vitrectomy (PPV) with endolaser and gas procedure. With her second retinal detachment she underwent a second PPV with silicone oil, and finally retinectomy with membrane peel upon her third retinal detachment.

Since her cataract extraction and retinal detachment repairs, her visual acuity was maintained at 20/80-100 in the right eye, and 20/20-25 in the left eye. Her baseline physical exam exhibited exophoria at distance and near; bilateral vertical diplopia, corrected with 7-8 vertical prism diopters; bilateral myopia; irregular right pupil shape; and fundoscopy of the right eye showing vitreous with silicon oil, scleral buckle, diffuse peripheral chorioretinal scarring and loss of foveal contour (Figure 1). Slit lamp examination demonstrated posterior capsule IOL (PCIOL) bilaterally with pigment on the right intraocular lens anterior surface due to posterior synechiae as well as corneal endothelial pigmentation secondary to iris rubbing and surgical trauma from her retinal procedures. Other exam findings were consistently unremarkable: extraocular movements full; pupils equal, round, and reactive to light; intraocular pressure within normal limits; peripheral vision full by confrontation visual fields; negative afferent pupillary defect; optic nerve cup to disc ratio of 0.40 left eye and 0.25 right eye with clear, distinct margins bilaterally.

In March 2015, five years following her retinal detachment repairs, routine fundoscopy of the left eye showed lattice degeneration of the superior, temporal, and inferior peripheral retina (Figure 1). She was referred to a retinal specialist for further evaluation. Considering the history of STL, retinal detachment in the right eye, previous myopia in the left eye, and lattice degeneration in the left eye, the patient was considered to be of higher risk of retinal tear or retinal detachment in the left eye. Subsequently, in 2017 she had prophylactic left laser retinopexy to prevent retinal detachment in her already compromised left eye.

Interestingly, throughout the years receiving routine optometry and retinal exams, she acquired several other, unexpected ocular conditions. In January 2016 she presented to her optometrist with complaints of sun irritation in both eyes with associated crusting and peeling of the surrounding skin. She was diagnosed with dry eye and allergic dermatitis with flaking, resolved with triamcinolone synthetic corticosteroid cream.

Several years later, in March 2022, the patient presented to her local optometrist with complaints of bilateral burning, aching, and irritation of eyes for approximately two weeks. Her slit lamp exam indicated diffuse superficial punctate keratitis, but no signs of infection or dendrites with fluorescein dye. She was diagnosed with a flare of dry eye syndrome of the bilateral lacrimal glands and her symptoms resolved following a taper with fluorometholone (ophthalmic corticosteroid) 0.1% suspension.

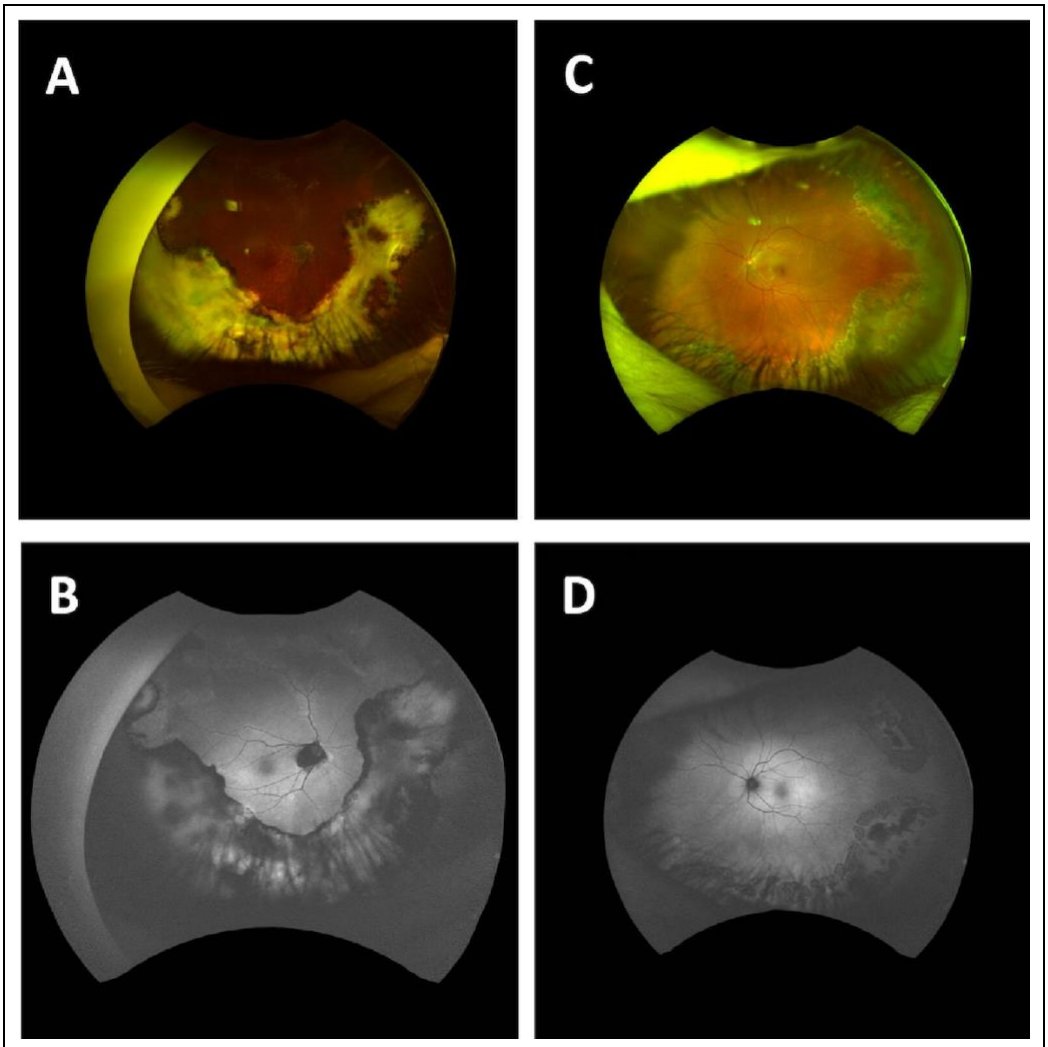


**Figure 1:** 2019 Optos photography of the right and left fundus. Optos images A (color fundus photography of the right eye) and B (autofluorescence imaging of the right eye) demonstrate diffuse peripheral chorioretinal scarring and loss of foveal contour. Right retinal changes occurred secondary to three retinal detachments in 2010 and associated retinal surgeries (scleral buckle and pars planar vitrectomy (PPV) with endolaser and gas, second PP with silicone oil, and retinectomy with membrane peel). Since having retinal surgeries in 2010, no right retinal changes have been appreciated with continued monitoring. Optos images C (color fundus photography of the left eye) and D (autofluorescence imaging of the left eye) show lattice degeneration of the superior, temporal, and inferior peripheral retina first observed in 2015 and treated with laser retinopexy in 2017.

## Discussion

This case of STL illustrates several unique features of which patients may present. Retinal detachment in patients with STL are reported to occur between the ages of 10-30 years, however this patient first experienced retinal detachment at age 56 [3]. Additionally, she had three total retinal detachments in her right eye which generated increased concern for left retinal detachment, leading to prophylactic left retinopexy. Continued follow up is needed to monitor the efficacy of these prophylactic measures.

Other unique ocular findings in this patient include bilateral vertical diplopia, allergic dermatitis, dry eye syndrome, and an episode of bilateral punctate keratitis, which is characterized by small intraepithelial lesions often in the center of the cornea [6]. These additional ocular presentations have previously not been described as sequalae of STL and it is unclear whether their etiology could be attributed to STL, either directly or indirectly. These additional manifestations are compelling, however, and warrant further investigations into unique, lesser seen ocular manifestations and symptoms of individuals diagnosed with STL.

Although the patient's personal history of type 2 diabetes mellitus and hypertension hold potential to contribute to further retinal disease, at the time of this report, both hypertension and diabetes had been well controlled with oral medication, diet, exercise, and annual evaluations. Of additional note, in April, 2018 she was diagnosed with Rheumatoid Arthritis and treated with Plaquenil 200 mg until 2021. During this time, she had no indication of toxic maculopathy on annual examinations. The authors do not feel that these elements of her medical history contributed to the ocular findings previously discussed.

## Conclusion

Although STL will often present in childhood to young adulthood, ophthalmologists and optometrists should be aware of the ocular abnormalities of the clinical diagnostic criteria for patients of all ages, which include lattice degeneration, retinal hole, retinal detachment, or retinal tear (ROBBIN 2000). In this case we also observed additional unique ophthalmic pathology of bilateral vertical diplopia, allergic dermatitis, dry eye syndrome, irregular pupil size, and bilateral punctate keratitis. It stands to be seen whether the unique ocular symptoms of our patient can be used as an aid to further help in the diagnosis and treatment of STL.

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