Complex Retinal Detachments - They Run in the Family

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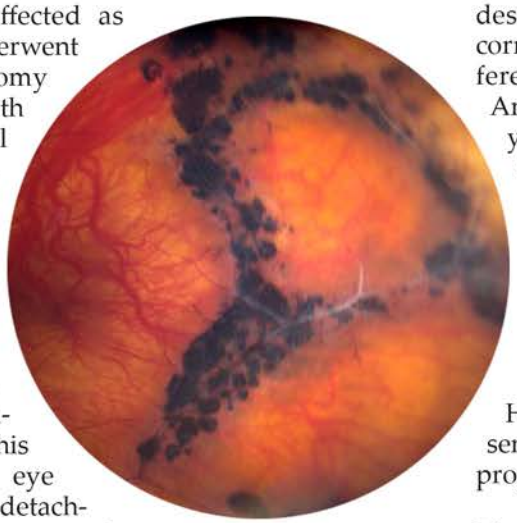
Introduction:

A 54-year-old man presented with temporal field loss in the left eye. The patient's acuity was 20/25, OU. Both eyes were pseudophakic. The posterior segments were remarkable for unusual vitreous veils, extensive circumferentially oriented lattice degeneration, some of which was perivascular. The left retina was shallowly detached with an inferior giant tear.

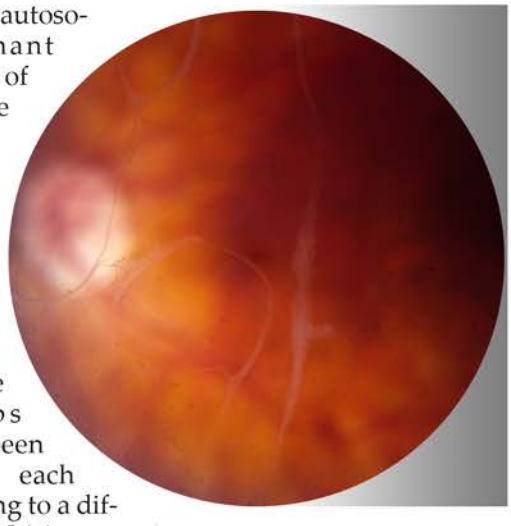
Diagnosis:

The patient had been diagnosed several years earlier with Stickler's Syndrome. Several family members were affected as well. The left eye underwent successful vitrectomy surgery combined with an encircling scleral buckle, but eventually all useful vision was lost to severe glaucomatous optic neuropathy. The lattice degeneration in the remaining eye was treated with prophylactic photocoagulation. In spite of this intervention, this eye developed a retinal detachment complicated by a similar giant tear. Two years after a vitrectomy procedure, this eye remains 20/30 with a securely attached retina.



Stickler's Syndrome is usually diagnosed in early childhood. It was first described by Gunnar Stickler, a pediatrician in 1965. This disorder is one of the most common hereditary connective tissue disorders in the United States.

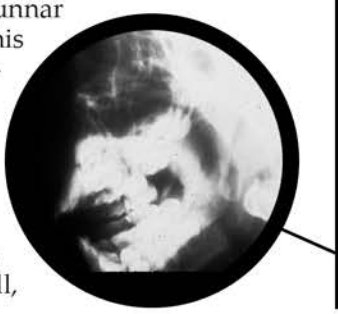
Mutations in the structural gene for Type II collagen (COL2A1) are responsible for most, if not all, cases. An autosomal-dominant pattern of inheritance with variable expressivity and nearly complete penetrance is the rule. At least five sub-groups have been described, each corresponding to a different COL2A1 mutation.



An autosomal-recessive form exists and additional, as yet, undiscovered loci are suspected. A classic presenting manifestation is the Pierre Robin malformation sequence which consists of a small lower jaw (micrognathia), a tongue that tends to fall backwards (glossoptosis), and incomplete closure of the roof of the mouth. Fifty per cent of patients with the Pierre Robin malformation have Stickler's Syndrome.

Hearing loss is common and may be conductive or sensorineural. The degree of loss varies and may be progressive.

Musculoskeletal complications are common and may include generalized joint laxity, scoliosis, chest and hip deformities, and early onset osteoarthritis. For this



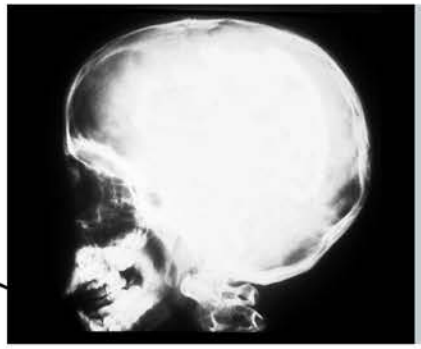
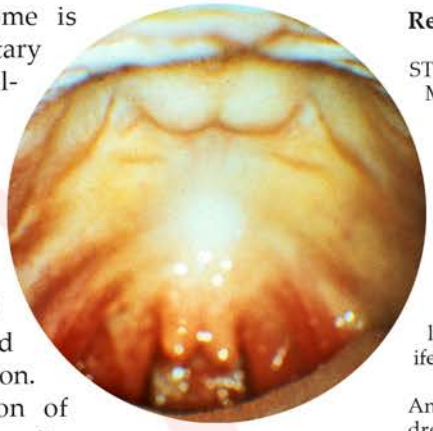


Figure 1: (Top) Stickler's veil

Figure 2: (Middle) Radial lattice

Figure 3: (Bottom) Pierre Robin micrognathia

reason, Stickler's Syndrome is often known as "Hereditary Progressive Artho-ophthalmopathy".



Ocular manifestations of Stickler's Syndrome include axial myopia, which is usually present early and varies from mild to severe. Wedged-shaped cortical cataracts are common. Fibrovascular condensation of the vitreous gel results in vitreous membranes in the retrolental space. The vitreous elsewhere is often optically empty. Radial perivascular pigmentation and radial lattice degeneration are frequent retinal findings.

Rhegmatogenous retinal detachments are the most serious ophthalmic complication and may occur in up to 65% of patients. The retinal breaks are often tractional, frequently multiple, and arise in markedly varying distances from the ora. The breaks are commonly posterior. Giant retinal tears on proliferative vitreoretinopathy increase the challenge of repair. These detachments are most commonly addressed with a primary vitrectomy, often combined with scleral buckling. In light of the complexity of these retinal detachments, the recommendations for prophylactic treatment are liberal. Three hundred sixty degree photocoagulation of the peripheral retina (ora secunda circlage) is often effective in reducing risk. The above patient's son is also followed by The Retina Institute and is doing well in one eye (20/30) following a vitrectomy and scleral buckling procedure, and eventually cataract extraction. The fellow eye is phthisical.

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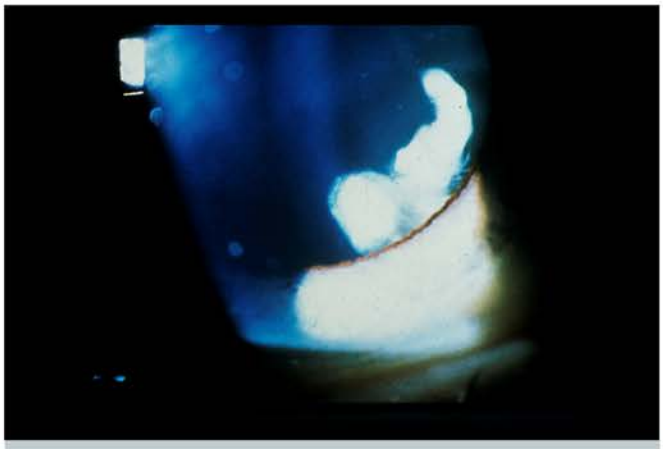

Figure 4: (top) High-arched palate
Figure 5: (bottom) Wedge-shaped cataract

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