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Commentary

# The Spectrum of Hereditary Disorders Associated with Retinal Detachment

**Gloria Stewart***

*Department of Anesthesiology, University of Oklahoma Health Sciences Center, Oklahoma City, USA*

## INTRODUCTION

Retinal detachment is a serious and sight-threatening condition that occurs when the retina, the thin layer of tissue at the back of the eye responsible for capturing light and transmitting visual information to the brain, separates from its underlying support tissue. This separation can lead to irreversible vision loss if not promptly treated. While retinal detachment can occur spontaneously or as a result of trauma or other ocular conditions, there is a significant subset of cases linked to hereditary disorders. These genetic conditions often predispose individuals to structural weaknesses or degenerative changes in the retina, increasing the risk of detachment. Understanding the spectrum of these disorders is crucial for early detection, management, and potential genetic counseling.

## DESCRIPTION

### Stickler syndrome

Stickler syndrome is one of the most common genetic conditions associated with retinal detachment. It is an inherited disorder that affects connective tissue, impacting the eyes, joints, and sometimes the auditory and craniofacial systems. Stickler syndrome results from mutations in several genes, including *COL2A1*, *COL11A1*, and *COL11A2*, which encode collagen proteins crucial for the structural integrity of various tissues. Ocular complications in Stickler syndrome are frequent and include high myopia (nearsightedness), vitreoretinal degeneration, and a high risk of retinal detachment, sometimes occurring at a young age. Prophylactic laser therapy to reinforce the retina may be recommended to reduce the risk of detachment in individuals diagnosed with Stickler syndrome.

### Marfan syndrome

Marfan syndrome is another connective tissue disorder that has ocular manifestations predisposing individuals to retinal detachment. It is caused by mutations in the *FBN1* gene, which encodes fibrillin-1, a protein essential for the formation of elastic fibers in connective tissue. Marfan syndrome is characterized by features affecting the cardiovascular system, skeletal system, and eyes. Ocular abnormalities include ectopia lentis (dislocation of the lens), high axial myopia, and an increased likelihood of retinal detachment. The risk of retinal detachment in Marfan syndrome is compounded by the elongation of the eye, which places stress on the retina. Management strategies focus on regular ophthalmic monitoring and surgical intervention when necessary.

### Ehlers-Danlos Syndrome (EDS)

Ehlers-Danlos syndrome encompasses a group of connective tissue disorders characterized by hyperelastic skin, joint hypermobility, and vascular fragility. Several subtypes of EDS, particularly the vascular and classical types, are associated with ocular complications, including retinal detachment. Mutations in genes such as *COL3A1* and *COL5A1* disrupt the production of collagen, compromising the structural integrity of the retina and increasing the risk of detachment. In patients with EDS, minor trauma or normal eye movements may be sufficient to precipitate retinal detachment, emphasizing the need for caution and proactive ophthalmic care.

### X-linked Juvenile Retinoschisis

X-Linked Juvenile Retinoschisis (XLRS) is an inherited retinal dystrophy that primarily affects young males and is caused by mutations in the *RS1* gene. This gene encodes retinoschisin, a protein essential for the structural and functional organization of the retina. In XLRS, splitting of the retinal layers occurs, usually in the macula, leading to reduced central vision. However, as the disease progresses, the structural instability of the retina can result in retinal detachment. Treatment options are limited, but ongoing research into gene therapy and other molecular approaches may offer future therapeutic possibilities.

### Wagner syndrome

Wagner syndrome is a rare autosomal dominant vitreoretinopathy that results in progressive degeneration of the vitreous and retina. It is caused by mutations in the *VCAN* gene,

**Correspondence to:** Gloria Stewart, Department of Anesthesiology, University of Oklahoma Health Sciences Center, Oklahoma City, USA; E-mail: gloria.s@gmail.com

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which encodes versican, an extracellular matrix protein crucial for the structural support of the eye. Patients with Wagner syndrome often present with myopia, cataracts, and a liquefied, optically empty vitreous. As the vitreous degenerates, tractional forces on the retina can lead to detachment. Management of Wagner syndrome typically involves vigilant monitoring and early surgical intervention if retinal detachment occurs.

### Best disease (Best vitelliform macular dystrophy)

Best disease is an autosomal dominant macular dystrophy caused by mutations in the *BEST1* gene, which encodes bestrophin-1, a protein involved in ion transport in the Retinal Pigment Epithelium (RPE). Although the disease primarily affects the macula, some patients experience retinal detachment due to subretinal fluid accumulation or atrophic changes. Best disease often presents in childhood with a characteristic "egg-yolk" lesion in the macula. Regular monitoring of retinal changes is critical for preventing complications such as detachment.

### Retinitis Pigmentosa (RP)

Retinitis Pigmentosa (RP) is a group of inherited retinal degenerative disorders characterized by progressive peripheral vision loss and night blindness. It results from mutations in various genes involved in photoreceptor function. While RP typically leads to gradual vision loss, some patients are at increased risk of developing retinal detachment, particularly those with associated vitreous abnormalities or high myopia. Surgical repair of retinal detachment in RP patients can be challenging due to the compromised retinal architecture, but early detection and intervention remain essential.

## CONCLUSION

Hereditary disorders associated with retinal detachment present a diverse and complex spectrum of conditions. The genetic mutations underlying these disorders often result in structural abnormalities of the retina, vitreous, or connective tissue, predisposing individuals to detachment. Early recognition of these genetic conditions through genetic testing and regular ophthalmologic evaluations is vital for minimizing the risk of vision loss. Advances in genetic research and emerging therapies hold promise for more effective prevention and treatment strategies in the future.