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Genetics in Stickler Syndrome

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Message from the Guest Editor

The Stickler syndromes form part of the spectrum of type II, IX, and XI collagenopathies. They are the most common cause of retinal detachment in children and the most common cause of familial retinal detachment.

In contrast to many other blinding retinal disorders, blindness from retinal detachment is both relatively common and preventable with accurate prediction of those at risk. Recent advances in the molecular genetic analysis of Stickler syndrome have allowed clinicians to stratify with far greater accuracy the risk of blindness in affected individuals and offer prophylactic surgery to reduce the risk of avoidable blindness, particularly in children.

The genes for type II, IX, and XI collagen are also expressed in elastic and hyaline cartilage, and patients with Stickler syndrome may also suffer from deafness, cleft palate, and premature arthropathy.

This review focusses on recent molecular genetic advances in all the multisystem aspects of Stickler syndrome, together with an overview of how these relate to myopia and retinal detachment in the population at large.

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Message from the Editor-in-Chief

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