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Keratoconus: Are the Earliest Warning Signs in our Genes?

A new addition to the clinician's toolbox uses next-generation sequencing technology and a custom gene panel for early disease detection

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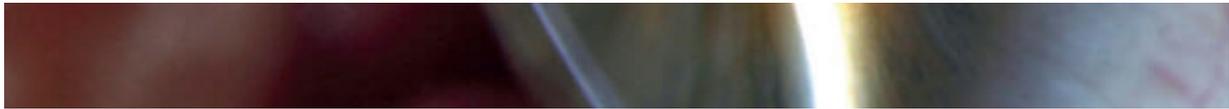


Clinicians on the lookout for keratoconus are about to benefit from a new addition to their armamentarium; Avellino Labs is developing a diagnostic genetic test for keratoconus risk factors.

So how does the genetic test work? First of all, a swab is used to collect DNA from the patient's cheek. The sample is then sent to Avellino's lab for next-generation sequencing (NGS) and analysis. The NGS custom panel targets over 1,000 variants across 75 genes for keratoconus (KC) and over 70 TGFBI mutations for corneal dystrophies (CD). Sequence results are aligned to the Genome Reference Human Build 37, and a relative risk (RR) score is calculated for the detected keratoconus variant. Risk scores were derived from a Bayesian logistic regression model constructed from NGS results, including whole exome sequencing and targeted sequencing platforms.

Current corneal cross-linking treatments are unable to reverse the damage caused by keratoconus – and changes to the cornea can result in vision deterioration, meaning that early detection of keratoconus is a serious need. For surgery candidates, early diagnosis of keratoconus is extremely important, as it can prevent post-surgery pathology progression. Holland explains: “By identifying those at-risk patients earlier, we can improve the monitoring for younger patients and potentially implement preventative treatments, such as collagen cross-linking. Genetic testing will also allow us to have additional information in the evaluation of refractive surgery patients. Knowing a patient's potential to progress to keratoconus could be a deciding factor in choosing one refractive procedure over another – or possibly not recommending corneal refractive surgery.”





Eye with keratoconus, with visible central scarring, and thinning with ectasia. Credit: David Yorston, Community Eye Health.

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