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Avellino Labs Begins Commercial Push for Expanded Ophthalmology Genetic Test Panel

By Molika Ashford

NEW YORK – Avellino Labs, a company working for close to a decade to bring genetic testing into the treatment and management of eye diseases, has launched an updated and expanded test product, now called AvaGen, focused on diseases and disorders of the cornea.

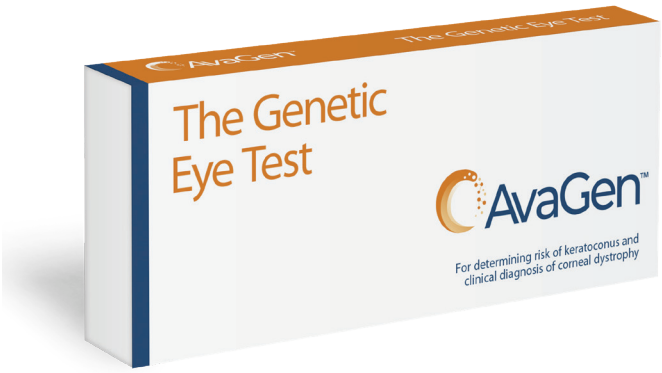
The assay, which is performed on a cheek swab, uses next-generation sequencing to detect variants across 75 genes. The results are intended to help clinicians diagnose and subtype corneal dystrophies, as well as to identify patients with elevated risk of a condition called keratoconus, which, when diagnosed early, can be managed with techniques to slow progressive corneal damage.

In both cases, the assay results can also influence an eye care professional's decision about patient eligibility for refractive vision correction surgery.

For corneal dystrophies, AvaGen determines the presence of a group of 70 known disease-causing TGFBI gene variants, providing a conclusive diagnosis of corneal dystrophy subtypes including epithelial basement membrane, granular and lattice disease distinctions, Reis-Bucklers, Schnyder, and Thiel-Behnke.

For keratoconus, the company analyzes detected variants across the 75-gene panel to create a polygenic risk score.

Avellino began its efforts several years ago, initially planning to commercialize a simpler test, focused just on TGFBI corneal dystrophy variants. By early 2020, though, the firm had developed the polygenic approach behind the new more comprehensive AvaGen assay and was ready to launch it commercially in the US. The COVID-19 pandemic put that on hold, however, and the firm pivoted last year to exclusively providing virus testing. This brought in a significant influx of cash, which is now fueling Avellino as it sets out to market AvaGen to the ophthalmology and optometry communities.



Avellino's AvaGen™, The Genetic Eye Test

In an interview, Avellino Labs CEO Jim Mazzo said that current corneal diagnostic imaging tools are relatively imprecise, especially for keratoconus, a disease in which the cornea lengthens and changes shape. It can be unclear to clinicians whether imaging results and other clinical signs point to keratoconus or to astigmatism or myopia. Some practitioners may also lack access to more advanced corneal mapping technology.

The result, Mazzo said, is that doctors adopt a wait-and-see policy, which can mean that patients only receive a definitive diagnosis once significant damage to their cornea has accumulated. Meanwhile, there is an available treatment for the disease, called cross-linking, which can prevent further degeneration. But once damage has occurred to the cornea, it can't be reversed, highlighting the value of early definitive diagnosis.

Gary Wortz, a Lexington, Kentucky, ophthalmologist specializing in cataract and refractive surgery who is integrating AvaGen into his practice, said in an email that despite sophisticated ocular measurement instruments, it remains challenging for clinicians to detect keratoconus in its early stages.

AvaGen doesn't diagnose keratoconus, but it identifies patients at a high genetic risk. Knowing a patient has this high risk, ophthalmologists can examine them more closely, and interpret findings less equivocally.

The hope is that this can lead to improved outcomes, for example, through earlier treatment with cross-linking therapy, before damage to the cornea has accumulated. In addition, earlier knowledge of keratoconus status could prevent negative LASIK outcomes by identifying more individuals for whom the surgery is contraindicated.

"Once we identify keratoconus, we can make clinical recommendations to reduce [a patient's] risk of progression, such as disqualifying them for LASIK surgery and recommending corneal cross-linking at the first clinical sign of the disease. Additionally, I can test a patient's family members. ... I can examine an entire family and see who may need additional care based on their test results," Wortz said in his email.

"Specifically, if patients have younger children, they're going to want to know this earlier because then you can proactively manage these diseases," Mazzo added.

Unlike keratoconus, which has complex polygenic origins, corneal dystrophies have more defined genetic contributors. TGFBI variants can mark patients as having specific disease subtypes with different clinical courses. According to Avellino CSO Nazneen Aziz, knowing a patient's subtype can help doctors better personalize monitoring or intervention.

Mazzo said that because LASIK and associated vision correction assessments are largely a patient-pay market, Avellino is treating AvaGen as an adjunct to that — something that would be billed as part of a larger package of clinical care. That said, diagnostic usage for corneal dystrophy subtyping could be reimbursed in some cases as professional recommendations and evidence for clinical utility evolve.

A report from the American Academy of Ophthalmology Task Force on Genetics in 2014, for example, recommends that clinicians "offer genetic testing to patients with clinical findings suggestive of Mendelian disorder whose causative gene(s) have been identified," such as TGFBI gene.

"When it comes to technology, you will have early adopters and late adopters. However, what I can say is that existing diagnostic technologies — as well-designed as they are — simply do not have the ability to help us detect keratoconus early," Wortz said.

"Given this reality, this entry into genetics for eye care should be, in general, accepted by a majority of ophthalmologists as it's a safe and effective way to improve patient care," he added, noting that Avellino's reported sensitivity and variant precision give him "high confidence" in the test results — enough to incorporate it into his practice.

Moving forward, the proof will be in the pudding to some extent, Wortz added. "As a practitioner, as I begin to integrate AvaGen into my work, I will be able to track my success rates, such as ... the number of patients I screen out for LASIK based on [their] test results," he said.

Overall, he expressed optimism. "Genetic [risk] findings have proven very valuable for oncologists and cardiologists in managing their patients, and I expect this similar genetic information will also prove valuable to eye care professionals," Wortz said. "In our field we are often faced with test results that are inconclusive, so the addition of genetic information will make a difference in decision-making around patient management and treatment as the results are much more definitive in nature."

Wortz also highlighted the fact that AvaGen is simple to administer. Avellino claims a turnaround time of 14 days or less and has created what it calls an "intuitive" report framework, which ordering clinicians access through a HIPAA-secured portal. To help providers and their patients understand the results, the firm also offers genetic counseling services.

As its efforts to drive adoption of AvaGen move forward this year with what Mazzo called a "full sales force," Avellino is also expanding its pipeline — first to other ocular disorders and then to additional areas in dermatology and dentistry.

The company plans to release a test for glaucoma — another polygenic disorder that suffers from issues of delayed diagnosis and where Avellino sees genetic information as helping improve and increase early intervention — by early next year. And it is working on developing genetic tools for atopic dermatitis and periodontitis.

Aziz explained that the connection between these new applications and the firm's current work in ophthalmology is that these oral and skin conditions share inflammatory features with keratoconus. As a result, the company expects their polygenic aspects to overlap.

Finally, Mazzo said the firm is also focused on commercializing its data by licensing it for pharmaceutical development.

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