

GENETIC TESTING INFORMS RISK FOR KERATOCONUS & CONFIRMS DYSTROPHIES



AvaGen™, The Genetic Eye Test (Avellino) is used to create an individual's genetic risk score for keratoconus and detect eight types of transforming growth factor beta-induced corneal dystrophies. This next-generation test sequences 75 genes to determine the number of risk and protective variants a person has. These variants determine the patient's polygenic risk score (PRS), which falls into low-, moderate-, or high-risk categories.

In a 2022 educational series, Brandon Ayres, MD; Melissa Barnett, OD, FAAO, FSLs, FBCLA; Barry Eiden, OD; Mitch Ibach, OD; David Kading, OD, FAAO, FCLSA; Elise Kramer, OD; Steven Sorkin, OD, FSLs; and Stephanie Woo, OD, described how they incorporated the AvaGen™ test into their practices. Here they shared case studies that highlight the usefulness of AvaGen™ for confirming or correcting treatment plans. Also, be sure to watch the webinar that Drs. Ayres, Eiden, and Barnett recorded for World Keratoconus Day, where they shared more clinical cases in which AvaGen™ helped guide their treatment plans (<https://www.geneticsandeyecare.com/webinar/world-keratoconus-day/>).

INCORPORATING GENETIC TESTING IN A PRACTICE

Genetic testing holds promising potential for vision care, although there is a learning curve for incorporating it into the ophthalmic practice, says Brandon Ayres, MD. He has found the technology a valuable addition to his diagnostic armamentarium. He and his staff underwent training for AvaGen™ via an online instructional video that covers all aspects of the test collection process. The online training is followed by an in-person review with a member of the Avellino team so staff members have an opportunity to ask questions.

THE AVAGEN™ TESTING PROCESS

After Dr. Ayres examines a patient's eyes, including imaging, there are usually two main reasons he recommends the AvaGen™ test for patients, either those referred to him for corneal pathology evaluation or refractive surgical

candidates: (1) if he suspects or is aware that a patient has a family member with keratoconus, or (2) if a refractive surgery candidate has suspicious findings via diagnostic testing.

- A patient test requisition form will be completed and, once a patient agrees to the Avagen™ test, a technician will take four buccal swabs and, with pre-paid label, mail the sample back to Avellino.
- Once the test is processed in the lab, the results are uploaded to a HIPAA-compliant portal, and Dr. Ayres receives an email notice that the results are available.
- Dr. Ayres or a technician reviews the test results and calls the patient. He may ask the patient to schedule a follow-up visit to go over his treatment plan. As appropriate, Dr. Ayres includes the complimentary genetic counseling offered, where a genetic counselor can further review the test results with the patient and answer any of his or her questions related to the findings.

AVAGEN™ CASE STUDIES

By giving eye care providers insight into a patient's potentially complex myopia pathology, genetic testing can help them make better-informed treatment plans, or help them correct the course of treatment if warranted.

CASE 1

Stephanie Woo, OD, used the AvaGen™ test in a 10-year-old patient with progressive myopia (the presentation began with -11.00 D with some cylinder OUS and increased by 2 lines in 2 years). The topographical map did not show a traditional keratoconic pattern, however it did appear irregular, even after a 2-week wash-out period. Fortunately, the AvaGen™ test assigned this patient a risk score of 3, which gave Dr. Woo the confidence to continue recommending ortho-k lenses for him. "Had his risk factor been substantially higher, I would have used a Pentacam on him every 6 months to monitor progression, but I would not have risked the ortho-k lenses," Dr. Woo said. She further explained, "Any time I can rely on a test to confirm a gut feeling, it makes me that much more confident in my treatment strategy. AvaGen™ is just that test."

CASE 2

A 12-year-old female presented to David Kading, OD, FAAO, FCLSA, with a prescription of -1.50 D and -1.75 D from a different provider the previous year.

Dr. Kading tested her at -3.50 D and -3.00 D, and subsequently recommended a myopia management program that included orthokeratology and 0.05% Atropine to help slow the myopia's progression.

The patient's topographies were not strongly indicative of keratoconus (they did not show steepening in the inferior), but they were also not completely symmetrical, and they had some unusual indices. With permission from the patient's mother, Dr. Kading conducted the AvaGen™ test. The patient's score was 83 out of a polygenic risk score range of 0 to 100. Thus, she was at high risk for developing keratoconus. Based on this new information, Dr. Kading switched this patient's treatment plan:

- Continue myopia management
- Keep using Atropine 0.05%
- Order spectacle lenses to wear
- Switch to a soft multifocal lens (-3.50 DS -3.25 DS).

"Before the availability of AvaGen™, The Genetic Eye Test, these topographies would not have worried me," Dr. Kading said. "I would have probably considered this patient at low risk for keratoconus, and I could have quite possibly missed it if she developed the condition in the future. With the information gained from AvaGen™, I was able to safely adjust my patient's treatment plan and begin regularly monitoring her for keratoconus."

GENETIC TESTING IS AN ADJUNCTIVE TOOL THAT DESERVES FURTHER EDUCATION

Genetic testing offers a novel way to advance vision care and prevent vision loss, and Melissa Barnett, OD, FAAO, FSLs, FBCLA, suggests that any practitioners who wish to include this technology in their diagnostic tool kit would benefit from learning the basics of how genetic testing works. It is useful for practitioners to understand these differences so they may vet which type of genetic test might

best suit a particular patient. It is useful for practitioners to understand how these genetic tests can aid in the diagnostic assessment of a patient.

Furthermore, Dr. Barnett cautions that genetic testing and its utility in medicine is in its infancy, and it should always be used alongside existing diagnostic eyecare tools. Genetic tests are not a replacement for other necessary screening, such as corneal imaging.