QUESTION: What is the Universal Test by Avellino Labs?

ANSWER: It is a molecular pathology test that detects genetic mutations in the TGFBI gene responsible for five distinct corneal dystrophies.

- Granular corneal dystrophy - type 1 (GCD1)
- Granular corneal dystrophy - type 2 (GCD2, aka Avellino dystrophy)
- Lattice corneal dystrophy – type 1
- Thiel Behnke corneal dystrophy
- Reis-Bücklers corneal dystrophy

This is the most specific test available to ascertain the risk of these corneal dystrophies.

QUESTION: How does it work?

ANSWER: The DNA is obtained in the physician’s office using a buccal mucosal swab. The patient rinses his/her mouth with water, the physician or medical assistant wipes the swab inside the mouth 10 times, places the swab in the plastic tube, packages it in a containment bag and sends it to Avellino Labs. The lab performs the test to identify the mutations responsible for these corneal dystrophies. The results are then available through a secure web portal within 24-48 hours after it is received by Avellino Labs.

QUESTION: What are the risks of genetic testing?

ANSWER: While the risk of collecting DNA by buccal swab is minimal, “A genetic test can affect a patient’s plans to have children, can create a sense of anxiety or guilt, and can even perturb a patient’s relationships with other family members. For these reasons, skilled counseling should be provided…to maximize the benefits and minimize the risks associated with each test.”

QUESTION: Is genetic testing a good idea?

ANSWER: The American Academy of Ophthalmology (AAO) Task Force on Genetics states, “Genetic testing can make a very positive impact on individuals and families affected with inherited eye disease. “…mutations in a single gene, and the detection of the responsible mutations can predict the development of the disease with relatively high accuracy (e.g., TGFBI-related corneal dystrophy).” The AAO Task Force recommends that clinicians “Offer genetic testing to patients with clinical findings suggestive of Mendelian disorder whose causative gene(s) have been identified” such as the TGFBI gene. However, the AAO Task Force also suggests all genetic testing should be done under the direction of a physician or genetic counselor through a CLIA certified lab.

QUESTION: What is the clinical relevance of this genetic test?

ANSWER: The results can guide clinical decision-making. In regards to GCD2, the International Committee for Classification of Corneal Dystrophies (IC3D) states, “Injury to the central cornea results in exacerbation of this corneal dystrophy with accelerated opacification. Hence, laser in situ keratomileusis, photorefractive keratectomy, and laser assisted in situ epithelial keratomileusis are strongly contraindicated in this dystrophy.” This test assists physicians to diagnose corneal dystrophies that may not be clear and present on slit lamp examination and guide decision making. Refractive surgery candidates, for example, should have genetic testing when the clinical findings, personal medical history, and family medical history indicate an increased risk of GCD2 or any of the other TGFBI dystrophies.

March 13, 2019

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Provided Courtesy of Avellino Labs USA, Inc

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6 QUESTION: Provide an example of a case where genetic testing is warranted.

ANSWER: A 25 y/o female presents for refractive surgery evaluation. Slit lamp examination reveals a few, small, nonspecific corneal opacities of uncertain origin in both eyes. She has a history of contact lens related problems including corneal ulceration with residual scarring. The patient is now contact lens intolerant, and desires refractive surgery. Family history is suggestive for corneal dystrophy. The physician orders genetic testing to ensure that the corneal opacities are not secondary to atypical GCD2. A decision to perform LASIK surgery will await results of the genetic testing.

7 QUESTION: Is this test accurate?

ANSWER: Yes. Internal data from a CLIA validation study compared genetic sequencing results of 734 clinically confirmed corneal dystrophy subjects and 136 normal controls to results of the Universal Test by Avellino Labs. The Universal Test results were identical to the genetic sequencing results, indicating 100% sensitivity and 100% specificity (no false positives and no false negatives).

8 QUESTION: Is this testing covered by insurance?

ANSWER: Sometimes. CMS states, “…genetic tests used to diagnose or determine treatment in the presence of signs and symptoms of disease can be covered by Medicare.” Conversely, CMS states, “Medicare does not pay for preventive screening tests except for those specifically authorized by statute (e.g., prostate-specific antigen test). Since CMS considers predictive tests to be screening tests, genetic tests for this purpose are not covered by Medicare.” Other third party payers have similar attitudes.

9 QUESTION: What is the Medicare payment rate?

ANSWER: The 2019 CMS Clinical Laboratory Fee Schedule for 81333 (TGFBI gene analysis) is $137.

10 QUESTION: Who bills for the genetic test?

ANSWER: Avellino Labs is the provider of the genetic test and bills insurance for this procedure when it is medically necessary. When the Test Requisition Form (TRF) indicates that the testing is screening, then Avellino Labs does not bill insurance; they bill the ophthalmologist or optometrist. In turn, the physician bills the patient for a noncovered service. Use an Advance Beneficiary Notice of Noncoverage (ABN) or similar form or process prior to rendering a noncovered service.

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