

Letters

A Different View on Genetic Testing

An Academy press release issued last fall¹ advised members to avoid genetic testing for complex eye disorders, stating that “currently marketed genetic tests for these eye disorders offer little benefit or additional insight regarding whether a patient is significantly predisposed to a particular disease. Furthermore, the organization strongly believes that a comprehensive eye exam is significantly more effective than any currently available genetic test for identifying treatable disease.”

In 1991, an editorial in the Academy journal *Ophthalmology* questioned the need for the new technique of phacoemulsification.² The Academy is cautious when assessing new technologies, so it is not surprising that it recommends that physicians refrain from routine genetic testing for AMD. I agree that routine testing is inappropriate and that tests should not be marketed directly to patients, but I disagree with the opinion that genetic testing does not offer real and measurable benefits.

A genetic test for AMD is not intended to determine the presence or severity of disease. Genetic testing enhances the predictive power of the clinical exam³ and

can identify which patients, among those of similar phenotype, may be at greater risk of disease progression. Genetic tests should be used in concert with a clinical exam. In fact, the newest commercial genetic AMD tests require the addition of clinical information to generate a test result.

Too many patients with AMD lose vision due to treatment delays. Those arguing that genetic testing for AMD offers no benefit ignore the practical reality that identifying and highlighting disease risk can lead to an overall reduction in vision loss, even with current therapies. Increased surveillance of higher-risk patients should lead to earlier detection of disease progression. For patients with neovascular AMD, earlier detection results in better vision.⁴

The understanding of the genetics of AMD is far from complete. Commercial tests will continue to change. Just as the early use of now-outdated OCT devices led to tremendous improvements in technology and interpretation, genetic testing by informed and thoughtful clinicians will speed the understanding and evolution of this revolutionary technology. Genetically guided preventive and therapeutic interventions will follow.

I have a financial interest in genetic testing, mainly because I think that genetic

testing for AMD will lead to better outcomes for our patients. The ultimate benefits of many medical technologies are not realized in a laboratory or pure research setting. Practicing physicians should be actively engaged in the responsible use, interpretation, and refinement of genetic tests for AMD.

Carl C. Awh, MD
Nashville, Tenn.

1 The press release titled “American Academy of Ophthalmology Discourages Genetic Testing for Age-Related Macular Degeneration” (Nov. 11, 2012) is available at www.aaopt.org/newsroom; select “News Releases,” then “2012 Releases.”

2 Lichter PR. *Ophthalmology*. 1991;98(4):415-416.

3 Seddon JM et al. *Ophthalmology*. 2011;118(11):2203-2211.

4 Boyer DS et al. *Ophthalmology*. 2007;114(2):246-252.

Dr. Awh is an equity investor in ArcticDx and a member of its scientific advisory board.

EDITOR'S NOTE: The editors checked the 1991 editorial referenced by Dr. Awh and found that it did not question the need for the new technique of phacoemulsification but, rather, was a general discussion of science, economics, and self-promotion. We have

CORRECTION Prices for the 2013 *Ophthalmic Coding Coach* book and online subscription in last month's issue were incorrect. The price of \$235 for members and \$350 for non-members is for each product, not both.



Last month's issue is online at www.eyenet.org/archives.

posted the editorial, along with Dr. Awh's letter, in the February 2013 online edition at www.eyenet.org. In addition, Ophthalmology editorials do not constitute official Academy policy. For the Academy policy on genetic testing, please read the Task Force recommendations, available at www.aaopt.org/one (select “Clinical Statements” under the Practice Guidelines tab).

Also, note that EyeNet discussed this topic in the Opinion “As We Look to Save Health Care Dollars, What to Do About the Valetudinarian?” (August 2012) and in the Clinical Update “Genetic Testing for Inherited Eye Disease: Why, How, and Who” (June 2012). Both of these articles can be found at www.eyenet.org/archives.