GLAUCOMA VISIONARIES

JANEY L. WIGGS, MD, PHD

Dr. Wiggs is the Paul Austin Chandler Professor of Ophthalmology and Vice Chair for Clinical Research in Ophthalmology at Harvard Medical School and Associate Director of the Ocular Genomics Institute at Massachusetts Eye and Ear in Boston.

1 What first drew you to ophthalmology and, specifically, to glaucoma?

As a medical student, I loved medicine, surgery, and pathology, and I was excited to find that ophthalmology included aspects of all three. I was particularly intrigued by the diagnostic and therapeutic challenges in glaucoma.

2 When you first became interested in the genetic component of glaucoma, what was the general knowledge of this association like at the time?

As a glaucoma fellow in 1991, I saw a patient who had multiple family members affected by glaucoma, and it was then that I learned that the disease could be inherited. At that time, the concept of glaucoma genetics was limited to these relatively rare affected families. It wasn't until the development of the statistical methods and genotyping technologies needed for large-scale genome-wide association studies that the more general contribution of genetics to glaucoma pathophysiology was understood.

3 Of the many significant "a-ha" moments that you have encountered throughout your pivotal work in genetics, which is most memorable?

Whenever we learn something new, it is exciting. Recently, we learned that *EFEMP1* mutations can cause juvenile glaucoma, and this was surprising because *EFEMP1* is a known cause of Doyne honeycomb retinal dystrophy, a seemingly unrelated disease. However, we have subsequently learned that *EFEMP1* contributes to a range of ocular conditions, likely through mechanisms that involve the extracellular matrix.

4 What are some of your research goals for the next few years?

Our ultimate goal is to use genetic information for clinical diagnosis, risk determination, and therapeutics.

NOMINATED BY ARSHAM SHEYBANI, MD

"Dr. Wiggs is a rare visionary who has helped us to understand the root cause for many glaucomas. Discoveries like hers will one day be utilized for personalized therapy."

"OVER THE NEXT SEVERAL YEARS, WE HOPE TO BETTER UNDERSTAND HOW TO INTEGRATE AND IMPLEMENT POLYGENIC RISK SCORES IN THE GLAUCOMA CLINIC."

Over the next several years, we hope to better understand how to integrate and implement polygenic risk scores in the glaucoma clinic. We are also conducting genetic analyses to identify novel glaucoma genes as well as understand the relationships between known genes and clinical features. The development of gene-based therapies and therapeutic strategies is also an important area of future investigations.

5 When did glaucoma last surprise you?

Glaucoma is continuously surprising, both in the clinic and in the laboratory. ■

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