which affects more than 64 million people worldwide—nerve damage results from high IOP, which occurs when the fluid that circulates in and out of the front part of the eye drains inadequately. Additionally, factors aside from elevated eye pressure, such as dysregulated optic nerve blood flow and low intracranial pressure, are also being pursued as possible causes of glaucoma. NEI-funded research has resulted in several Food and Drug Administration (FDA)-approved pressure-reducing drug therapies, which have been found by the NEI-sponsored Ocular Hypertension Treatment Study (OHTS) to delay disease onset. In addition to drug regimens, glaucoma is also treated through traditional and minimally invasive surgical techniques—many including the use of FDA-approved drainage devices to enhance aqueous humor outflow from the eye to reduce IOP.

Dr. Pasquale participates in NEI’s Glaucoma Human Genetics Collaboration (NEIGHBOR) Consortium and the Glaucoma Gene and Environment Initiative (GLAUGEN) Study. These collaborative efforts, under the umbrella of a larger consortium called NEIGHBORHOOD (The NEIGHBOR Heritable Overall Operational Database), involves clinicians and geneticists at multiple institutions throughout the United States whose goal is to identify genetic variants associated with POAG in the largest and most thoroughly characterized population of people with known glaucoma status. Through mid-2018, NEIGHBORHOOD in collaboration with other large consortia throughout the world, has identified 133 genetic variants that predict within 75 percent accuracy a person’s risk for developing primary open angle glaucoma. As Dr. Pasquale noted, “Of course, more work is needed to improve this accuracy rate. But it is remarkable that recently completed research found an almost direct correlation between the magnitude of the genetic variants’ effect on eye pressure and their effect on glaucoma risk, confirming that IOP is an important factor that determines whether someone develops glaucoma.”

Among the 133 gene variants identified associated with glaucoma risk, 68 had not been previously linked to IOP, and their loci point to cellular processes, such as lipid metabolism and nitric oxide signaling, that contribute to elevated pressure. Dr. Pasquale then spoke in depth about the cellular processes, biomedical mechanisms, and metabolic pathways that not only result in elevated IOP but increase the vulnerability of the optic nerve to resulting damage. Through understanding the genetic basis of glaucoma and these concomitant pathways involved, clinicians would move even closer to making an earlier diagnosis of POAG. Furthermore, since no current treatment paradigm completely stops POAG, this genomic research may also lead to neuroprotection strategies that could potentially halt disease progression.

Dr. Pasquale concluded by commenting that, “Since I last spoke on Capitol Hill in 2014, I am pleased to report that the NEI-funded NEIGHBORHOOD Consortium has greatly expanded our understanding of the genetic basis of POAG, and that research into the associated metabolic pathways is moving us further to an earlier diagnosis of the disease and the development of effective therapies to stop its progression.”

About World Glaucoma Week 2019...

The first World Glaucoma Day was held on March 6, 2008, and the United States House of Representatives passed H.R. 981, which recognized the event and supported the NEI’s efforts to research the causes of and treatments for glaucoma. Since 2010, the day has expanded into a week of educational events held worldwide, with all major glaucoma professional societies and research organizations co-sponsoring AEVR’s 2019 event, including:

- Research to Prevent Blindness (RPB)
- American Glaucoma Society (AGS)
- Association for Research in Vision and Ophthalmology (ARVO)
- Glaucoma Research Foundation (GRF)
- Optometric Glaucoma Society (OGS)

In addition to speaking at the Briefing, Dr. Pasquale also visited his New York Congressional delegation in NAEVR-hosted visits.

“Research into the associated metabolic pathways is moving us further to an earlier diagnosis of the disease and the development of effective therapies to stop its progression.” – Dr. Pasquale