

eyeGENE™- National Ophthalmic Disease Genotyping and Phenotyping Network

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Genetic mutations are associated with many ocular diseases including glaucoma, cataracts, strabismus, corneal dystrophies, and many forms of retinal degenerative disease. As a result, gene-based therapies are actively being pursued to ameliorate genetic eye diseases that once were considered untreatable. However, molecular diagnostic testing for these diseases is not widely available to patients. The National Ophthalmic Disease Genotyping and Phenotyping Network (eyeGENE™) was created three years ago by the National Eye Institute (NEI), part of the National Institutes of Health (NIH), in partnership with research laboratories, ophthalmic clinics and CLIA-certified molecular diagnostic laboratories in the United States and Canada. The major goals of this network are to 1) broaden patient accessibility to diagnostic genetic testing, 2) facilitate research into the genetic causes of ophthalmic disease by establishing a genotype/phenotype database and a DNA repository, and 3) provide a de-identified genotype/phenotype-linked samples resource for ophthalmic disease researchers. This Network currently includes the eyeGENE Coordinating Center, more than 120 ophthalmic clinical organizations in the US and Canada, and 12 clinical diagnostic laboratories. The Coordinating Center, based at NEI, manages a centralized repository for blood/DNA and a secure web-based genotype/phenotype database. To date, Network molecular laboratories offer testing for mutations in more than 65 genes. Mutations in these genes correlate with 32 different clinical diagnoses. At the time of this submission, more than 1200 patient samples have been collected in the repository. Public access to de-identified clinical and genetic information is under development and will be available to researchers in the vision community upon NIH IRB approval. The eyeGENE Network will continue to serve the vision community by promoting accessibility of genetic testing to patients with ocular diseases and by promoting clinical and basic research for a better understanding and treatment of ocular diseases.