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The national ophthalmic genotyping and phenotyping network (eyegene®): Preserving vision through genetics

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Molecular genetics is revolutionizing the diagnosis and treatment of inherited eye diseases. In 2003, the National Eye Institute of the National Institutes of Health-in an effort to facilitate future basic and clinical research in inherited eye disease-created The National Ophthalmic Disease Genotyping and Phenotyping Network (eyeGENE®). eyeGENE® is a DNA repository and patient registry for inherited eye diseases coupled to phenotypic descriptors and molecular genetic information for which health care professionals throughout the United States and Canada can enroll their patients. A portion of the DNA extracted from a blood sample from the donor may be used for CLIA-certified molecular diagnostics that can be returned to the referring clinician to be used in patient care. Most recently the program added the option for vision researchers to be able to request access to a de-identified database of phenotype and genotype information for participants enrolled in eyeGENE® and request DNA aliquots for their research studies. eyeGENE® also offers participants the option to be included in a patient registry so that they may be contacted if an approved clinical study for which they might qualify is offered. Several studies have now successfully recruited participants to studies and developed high throughput diagnostics using patient DNA samples. eyeGENE® anticipates increased study proposals on a variety of issues in the near future as more researchers discover the resource.

Biography

Kerry Goetz attended West Virginia University, where she earned a Bachelor of Science in Animal and Veterinary Science in 2002, a Multidisciplinary Studies Degree in Professional Writing, Business, and Communication in 2008, and a Master of Science in Genetics and Developmental Biology in 2006. In 2006, she began working under Dr. Visvanathan Ramamurthy at the West Virginia Eye Institute, where she studied Leber congenital amaurosis and congenital stationary night blindness. Kerry Goetz joined the NIH in the Office of Director Elias Zerhouni in the summer of 2008 and transitioned to the NEI in October 2008 working as a coordinator for the National Ophthalmic Disease Genotyping Network (eyeGENE®). Her knowledge and expertise in the field of genetics has expanded the number of patients and doctors participating in eyeGENE® throughout the United States and Canada and opened the eyeGENE® Biorepository to scientists to facilitate vision research. The eyeGENE® Network is working to bring advances in identifying ophthalmic disease genes to the clinical practice. It is a component of the NEI Intramural Research Program and is conducted in partnership with extramural academic clinic and research laboratories. Goetz interacts with patients and their families, eye healthcare providers, and vision researchers on a daily basis. She maintains the two eyeGENE® clinical protocols and shepherds them through various review processes both domestic and abroad. She also serves as the liaison between individuals and offices involved in the oversight of human subject research at NIH and expanding the science of biobanking. Kerry also is involved in several efforts on the NIH campus to standardize data sets to facilitate data sharing across studies. Outside of work, is active in the National Organization of Albinism and Hypopigmentation and The Way Eye See It, a Northern Virginia-based group of parents with visually impaired children. She is a Girl Scout leader and enjoys being outdoors with her family and dog. She also has a passion for cooking.

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