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## DISTINGUISHED FACULTY LECTURESHIP AWARD IN BIOMEDICAL RESEARCH

### **Anand Swaroop, Ph.D.**

*Harold F. Falls Collegiate Professor of Ophthalmology and Visual Sciences*

*Professor, Department of Human Genetics*

*Director, Center for Retinal and Macular Degeneration*

*Director, Sensory Gene Microarray Node*

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Internationally recognized for his research on retinal gene discovery, gene regulation and diseases, Dr. Anand Swaroop leads major research programs on the genetics of retinal diseases such as retinitis pigmentosa, diabetic retinopathy and age-related macular degeneration.

Dr. Swaroop's publication record started early in his career, when as a postdoctoral student he discovered a novel gene (*p150glued*) in *drosophila*, demonstrating that mutations in this gene cause an eye phenotype. Dr. Swaroop's discovery represented one of the first descriptions of an autosomal dominant disease gene and resulted in two first-author publications in *PNAS (Proceedings of the National Academy of the*

*Sciences of the United States of America)*. Now *p150glued's* function is recognized as an essential component of microtubule motor cytoplasmic dynein-dynactin, involved in numerous cellular activities and for the pathogenesis of motor neuron diseases and certain ciliopathies.

Dr. Swaroop moved to human genetics and went on to make outstanding contributions to the understanding of the genetics of retinal diseases, by identifying novel genes that play a major role in retinal development and pathogenesis of RP, such as the *neural retina-specific leucine zipper (Nrl)* gene. He made major contributions to the molecular genetics of the retinitis pigmentosa GTPase regulator (RPGR), Usher syndrome, *CRX*, *RP3*, *CTRP5*, *SEC13R* and the photoreceptor-specific nuclear receptor gene (*NR2E3*). Recently, Dr. Swaroop's group revealed new insights into the role of genetic susceptibility variants associated with AMD. Articles in *Nature Genetics* and *PNAS* describe the in-depth analysis of variants in the region of complement factor H (CFH) and *ARMS2* and reveal new insights that are valuable for AMD diagnosis and therapy.

These scientific insights and achievements have advanced the genetics of RP and AMD – and have contributed greatly to the understanding of basic molecular mechanisms in cell and developmental biology.

Earlier this year the Foundation Fighting Blindness awarded Dr. Swaroop the Board of Director's Award for outstanding research achievement for his role in discovering a gene that will advance the diagnosis and understanding of LCA (Leber's Congenital Amaurosis), an inherited degenerative eye disease that occurs at birth or in infancy. The discovery of the gene *CEP290* is a significant addition to the knowledge of this early-onset degenerative disease. When coupled with previous discoveries, scientists can now account for 70 percent of the genetic contributors to LCA. The gene *CEP290* also may have implications for related diseases such as RP and Joubert Syndrome.

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In *Nature*, Dr. Swaroop and his colleagues reported on the successful transplantation of precursors to rod photoreceptors (the cells that provide vision in dim light) in the mouse retina. They were able to isolate the committed rod precursors and show that, upon transplantation in degenerating retina, these cells survived, differentiated and properly integrated with other cells. The mice responded to light, as indicated by pupil response. The research has profound implications for people who suffer from diseases caused by the loss of photoreceptors.

The National Institutes of Health, the Foundation Fighting Blindness, Macula Vision Research Foundation, Juvenile Diabetes Research Foundation and Research to Prevent Blindness, among others, support Dr. Swaroop's research. He was one of the first vision scientists in the country to receive funding from the National Eye Institute to establish a microarray facility, using technology that dramatically speeds the process by which scientists can scan for disease genes. In 2003, Dr. Swaroop received a grant from the Elmer and Sylvia Sramek Charitable Foundation to establish a genetics database and data analysis programs at the Kellogg Eye Center. He has been the director of the Microarray and Molecular Biology Core since 1997 and the director of the Center for Retinal and Macular Degeneration at the Kellogg Eye Center since 2001.

In recognition of his leadership in vision research, Dr. Swaroop has been the recipient of numerous honors, including the Harrington Senior Scientific Award from Research to Prevent Blindness, and has been a finalist for the prestigious Alcon award three years in a row. He is highly regarded by his colleagues and the numerous students (including almost 100 medical and undergraduate students), research fellows and junior faculty he mentors.

Dr. Swaroop obtained his Ph.D. in Biochemistry at the Indian Institute of Science in Bangalore, India, in 1982. He received his postdoctoral training in molecular biophysics and biochemistry at Yale University, and in human genetics at Yale University School of Medicine.

He joined the faculty at the University of Michigan in 1990 as an assistant professor in the departments of Ophthalmology and Human Genetics. He became professor in 2000 and Harold F. Falls Collegiate Professor in 2003.

*Dr. Swaroop spends his free time with his wife Manju, son Alok and daughter Kanchan. They like to travel and have been to many places in the U.S. and abroad. Dr. Swaroop is an avid follower of political events and enjoys discussions with friends. He is passionate about good Indian music and Michigan football.*

*Dr. Swaroop credits Manju and his wonderful children for a stable and loving environment, which has enabled him to do his research. He is grateful to his parents (his dad has AMD) for teaching basic human values and his numerous friends and colleagues for encouragement and support.*