

**BIOGRAPHICAL SKETCH**

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NAME <b>Swaroop, Anand</b>	POSITION TITLE <b>Harold F. Falls Collegiate Professor</b>		
eRA COMMONS USER NAME <b>swaroop</b>			
EDUCATION/TRAINING <i>(Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.)</i>			
INSTITUTION AND LOCATION	DEGREE <i>(if applicable)</i>	YEAR(s)	FIELD OF STUDY
G.B. Pant University, Pant Nagar, India	M.Sc.	1977	Biochemistry
Indian Institute of Science, Bangalore, India	Ph.D.	1982	Biochemistry

**Professional Experience**

1982-86 Post-doctoral Associate, Dept. of Mol. Biophys. & Biochem., Yale University, New Haven, CT.  
 1986-87 Associate Research Scientist, Dept. Hum. Genet., Yale Univ. Sch. Med., New Haven, CT.  
 1987-88 NRSA Post-doctoral Fellow, Dept. Hum. Genet., Yale Univ. Sch. Med., New Haven, CT.  
 1988-90 Associate Research Scientist, Dept. Hum. Genet., Yale Univ. Sch. Med., New Haven, CT.  
 1990-96 Assistant Professor, Department of Ophthalmology, University of Michigan, Ann Arbor, MI.  
 1990-98 Assistant Professor, Department of Human Genetics, University of Michigan, Ann Arbor, MI.  
 1991- Faculty Member, Graduate Program in Cellular & Molecular Biology.  
 1996-2000 Associate Professor, Department of Ophthalmology & Visual Sciences.  
 1996- Faculty Member, Neuroscience Graduate Program.  
 1998-2002 Associate Professor, Department of Human Genetics  
 2000- Professor, Department of Ophthalmology and Visual Sciences  
 2000-2000 Scientist (on sabbatical for 6 months), Laboratory of Genetics, Salk Institute, La Jolla, CA  
 2001- Coordinator/Director, Center for Retinal and Macular Degeneration, University of Michigan  
 2002- Professor, Department of Human Genetics  
 2003- Harold F. Falls Collegiate Professor of Ophthalmology & Visual Sciences  
 2006- Member, Center for Computational Medicine and Biology, University of Michigan.

**Memberships in Professional Societies, Honors and Awards**

American Association for the Advancement of Science, American Society of Human Genetics, Association for Research in Vision and Ophthalmology, Society for Neuroscience.

First Young Investigator Award from the Retinitis Pigmentosa Foundation, 1990. Lew R. Wasserman Merit Award from the Research to Prevent Blindness (RPB), 1997. Employee Recognition Award for outstanding service to the Department of Ophthalmology and Visual Sciences, 1999. RPB Research Sabbatical Award, June 2000. Inventor Recognition Award, University of Michigan, 2001. RPB Senior Scientific Investigator Award, January 2004.

The Foundation Fighting Blindness Board of Directors Award in recognition of outstanding research achievements, January 26, 2007.

**Scientific Activities**

- Chair, Appointments and Promotions Committee in Ophthalmology & Visual Sciences, 2005– .
- Member, Medical School Advisory Committee for Appointments, Promotions and Tenure, 2006– .
- Member (ad hoc), NIH Mammalian Genetics Study Section, October 1992. Member, Special Review Committee (study section), NIH/NEI, May 1993. Member, Special Review Committee (VisB 01 study section), NIH/NEI, December 1995. Member (ad hoc), Visual Sciences C study section, February 1998, October 2001, February, June and October 2002. NEI special study section ZEY1 VSN (01), July 2006.
- Member, Executive Committee, Human Genome Center at University of Michigan, 1990-94.
- Reviewer for several journals, including AJHG, HMG, IOVS, JBC, Mol. Vis., PNAS, MCB & Nat. Genet.
- Grants Reviewer for The Foundation Fighting Blindness; The Wellcome Trust, U.K.; Comitato Promotore Telethon, Italy; The South Africa Retinitis Pigmentosa Foundation; The Medical Research Council of

Canada; Canadian Foundation Fighting Blindness; Juvenile Diabetes Research Foundation, National Science Foundation.

- Ad-hoc member, National Eye Institute Board of Scientific Counselors (BSC). June 2001.
- Regular Member, BDPE (previously Visual Sciences C) study section, February 2003–June 2005.
- Member, Stem Cell Biology Group, The Foundation Fighting Blindness. 2006 – .
- Member, Genetics Advisory Committee, Age – Related Eye Disease Study 2 (ARED2), National Eye Institute. 2006 – .
- Editorial Boards, *Investigative Ophthalmology & Visual Science*, June 2002– . *Molecular Vision*, 1995– .

#### Selected peer-reviewed publications since 2004 (total = 157)

1. Abecasis GR, Yashar BM, Zhao Y, Ghasvand N, Zarepari S, Branham KEH, Reddick AC, Trager EH, Yoshida S, Bahling J, Filippova E, Elner S, Johnson MW, Vine AK, Sieving PA, Jacobson SG, Richards JE, **Swaroop A**: Age-related macular degeneration: A high resolution genome scan for susceptibility loci in a population enriched for late-stage disease. *Am J Hum Genet* 74:482-494, 2004.
2. Akimoto M, Filippova E, Gage PJ, Zhu X, Craft CM, **Swaroop A**: Transgenic mice expressing Cre-recombinase specifically in M- or S-cone photoreceptors. *Invest Ophthalmol Vis Sci* 45:42-47, 2004.
3. Yoshida S, Mears AJ, Friedman JS, Carter T, He S, Oh E, Jing Y, Farjo R, Fleury G, Barlow C, Hero AO, **Swaroop A**: Expression profiling of the developing and mature Nrl<sup>-/-</sup> mouse retina: Identification of retinal disease candidates and transcriptional regulatory targets of Nrl. *Hum Mol Genet* 13:1487-1503, 2004.
4. Cheng H, Khanna H, Oh ECT, Hicks D, Mitton KP, **Swaroop A**: Photoreceptor specific nuclear receptor NR2E3 functions as a transcriptional activator in rod photoreceptors. *Hum Mol Genet* 13:1563-1575, 2004.
5. Zarepari S, Hero A, Zack DJ, Williams RW, **Swaroop A**: Seeing the unseen: Microarray-based gene expression profiling in vision. *Invest Ophthalmol Vis Sci* 45:2457-2462, 2004.
6. Strettoi E, Mears AJ and **Swaroop A**: Recruitment of the rod pathway by cones in the absence of rods. *J Neurosci.* 24:7576-7582, 2004.
7. Yu J, He S, Friedman JS, Ghosh D, Mears AJ, Hicks D, **Swaroop A**: Altered expression of genes of the Bmp/Smad and Wnt/Calcium signaling pathways in the cone-only Nrl-knockout mouse retina, revealed by gene profiling using custom cDNA microarrays. *J Biol Chem.* 279:42211-42220, 2004.
8. Friedman JS, Khanna H, Swain PK, DeNicola R, Cheng H, Mitton KP, Weber CH, Hicks D, **Swaroop A**: The minimal transactivation domain of the basic motif-leucine zipper transcription factor NRL interacts with TATA-binding protein. *J Biol Chem.* 279:47233-47241, 2004.
9. Nishiguchi KM, Friedman JS, Sandberg MA, **Swaroop A**, Berson EL, Dryja TP: Recessive NRL mutations in patients with clumped pigmentary retinal degeneration and relative preservation of blue cone function. *Proc Natl Acad Sci USA.* 101:17819-17824, 2004.
10. Otto EA, Loeys B, Khanna H, Hellemans J, Sudbrak R, Fan S, Muerb U, O'Toole JF, Helou J, Attanasio M, Utsch B, Sayer JA, Lillo C, Jimeno D, Coucke P, Paepe AD, Reinhardt R, Klages S, Tsuda M, Kawakami I, Kusakabe T, Omran H, Imm A, Tipples M, Raymond PA, Hill J, Beales P, He S, Kispert A, Margolis B, Williams DS, **Swaroop A**, Hildebrandt F: A novel ciliary IQ domain protein, NPHP5, is mutated in Senior-Loken syndrome (nephronophthisis with retinitis pigmentosa), and interacts with RPGR and calmodulin. *Nature Genet.* 37:282-288, 2005.
11. Zarepari S, Buraczynska M, Branham KEH, Shah S, Eng D, Pawar H, Yashar BM, Moroi SE, Lichter PR, Petty HR, Richards JE, Abecasis G, Elner VM, **Swaroop A**: Toll-like receptor 4 variant D299G is associated with susceptibility to age-related macular degeneration. *Hum Mol. Genet.* 14:1449-1455, 2005.
12. Carter TA, Greenhall JA, Yoshida S, Fuchs S, Helton R, **Swaroop A**, Lockhart DJ, Barlow C: Mechanisms of aging in senescence-accelerated mice. *Genome Biol.* 6:R48, 2005.

13. Zarepari S, Branham KEH, Shah S, Klein RJ, Ott J, Hoh J, Abecasis GR, **Swaroop A**: Strong association of the Y402H variant in Complement Factor H at 1q32 with susceptibility to age-related macular degeneration. *Am J Hum Genet.* 77:149-153, 2005.
14. Khanna H, Hurd TW, Lillo C, Shu X, Parapuram SK, He S, Akimoto M, Wright AF, Margolis B, Williams DS, **Swaroop A**: RPGR-ORF15, which is mutated in retinitis pigmentosa, associates with SMC1, SMC3, and microtubule transport proteins. *J Biol Chem.* 280:33580-33587, 2005.
15. Zhu D, Hero AO, Cheng H, Khanna R, **Swaroop A**: Network constrained clustering for gene microarray data. *Bioinformatics* 21:4014-4020, 2005.
16. Zacks DN, Han Y, Zeng Y, **Swaroop A**: Activation of signaling pathways and stress response genes in an experimental model of retinal detachment. *Invest Ophthalmol Vis Sci.* 47:1691-1695, 2006.
17. Akimoto M, Cheng H, Zhu D, Brzezinski JA, Khanna R, Filippova E, Oh EC, Jing Y, Linares JL, Brooks M, Zarepari S, Mears AJ, Hero A, Glaser T, **Swaroop A**: Targeting of GFP to newborn rods by Nrl promoter and temporal expression profiling of flow-sorted photoreceptors. *Proc Natl Acad Sci. USA* 103:3890-3895, 2006. [Cover].
18. Chang, B., Khanna, H., Hawes, N., Jimeno, D., He, S., Lillo, C., Parapuram, SK, Cheng, H., Scott, A., Hurd, RE, Sayer, JA, Otto, EA, Attanasio, M., O'Toole, JF, Jin, G., Shou, C., Hildebrandt, F., Williams, DS, Heckenlively, JR, **Swaroop A**: In-frame deletion in a novel centrosomal/ciliary protein CEP290/NPHP6 perturbs its interaction with RPGR and results in early-onset retinal degeneration in the *rd16* mouse. *Hum Mol Genet.* 15:1847-1857, 2006.
19. Sayer, JA, Otto EA, O'Toole JF, Nurnberg G, Kennedy MA, Becker C, Hennies HC, Helou J, Attanasio M, Fausett BV, Utsch B, Khanna H, Liu Y, Drummond I, Kawakami I, Kusakabe T, Tsuda M, Ma L, Lee H, Larson RG, Allen SJ, Wilkinson CJ, Nigg EA, Shou C, Lillo C, Williams D, Hoppe B, Kemper M, Neuhaus T, Petry M, Kispert A, Zou Y, Gloy J, Ganner A, Walz G, Zhu X, Goldman D, Nurnberg P, **Swaroop A**, Leroux MR, Hildebrandt F: The centrosomal protein nephrocystin-6 is mutated in Joubert syndrome and activates transcription factor ATF4. *Nat Genet.* 38:674-681, 2006.
20. Khanna H, Akimoto M, Siffroi-Fernandez S, Friedman JS, Hicks D, **Swaroop A**: Retinoic acid regulates the expression of photoreceptor transcription factor NRL. *J Biol Chem.* 281:27327-27334, 2006.
21. Cheng H, Aleman TS, Cideciyan AV, Khanna R, Jacobson SG, **Swaroop A**: *In vivo* function of the orphan nuclear receptor NR2E3 in establishing photoreceptor identity during mammalian retinal development. *Hum Mol Genet.* 15:2588-2602, 2006.
22. Li M, Atmaca-Sonmez P, Othman M, Branham KEH, Khanna R, Wade MS, Li Y, Liang L, Zarepari S, **Swaroop A\***, Abecasis GR\*: CFH haplotypes without Y402H coding variant show strong association with susceptibility to age-related macular degeneration. *Nature Genet.* 38:1049-1054, 2006 (\*co-corresponding authors)
23. MacLaren RE, Pearson RA, MacNeil A, Douglas RH, Salt TE, Akimoto M, **Swaroop A\***, Sowden JC, Ali RR\*: Retinal repair by transplantation of photoreceptor precursors. *Nature* 444:203-207, 2006 (\*co-corresponding authors). [Cover]
24. Friedman JS, Chang B, Kannabiran C, Chakarova C, Singh HP, Jalali S, Hawes NL, Branham K, Othman M, Filippova E, Thompson DA, Webster A, Andreasson S, Jacobson SG, Bhattacharya SS\*, Heckenlively JR, **Swaroop A\***: Premature truncation of a novel protein, RD3, exhibiting sub-nuclear localization is associated with retinal degeneration. *Am J Hum Genet.* 79:1059-1070, 2006 (\*co-corresponding authors).
25. Oh ECT, Khan N, Novelli E, Khanna H, Strettoi E, **Swaroop A**: Transformation of cone precursors to functional rod photoreceptors by bZIP transcription factor NRL. *Proc Natl Acad Sci USA* 104:1679-1684, 2007. [Cover]
26. Kanda A, Friedman JF, Nishiguchi KM, **Swaroop A**: Retinopathy mutations in the bZIP protein NRL alter phosphorylation and transcriptional activity. *Human Mutation.* In press.

**ACTIVE RESEARCH SUPPORT**

RO1 EY11115-10

12/01/04 – 11/30/08

NIH

"Molecular Mechanisms of Retina-Specific Gene Expression"

The goals of this study are to delineate the molecular mechanisms of gene regulation in the retina. A major focus is on the bZIP transcription factor, NRL, its interacting proteins, and target genes.

RO1 EY016862-01

03/01/06 – 02/28/11

NIH.

"Genetic Variations in Age-related Macular Degeneration

The goals of this study are to identify genetic susceptibility variants associated with age-related macular degeneration by genome-wide and targeted association studies.

The Foundation Fighting Blindness

07/01/04 – 06/30/07

"Resource Facility for X-linked Retinitis Pigmentosa and Inherited Retinal and Molecular Dystrophies"

The primary goal of the project is to continue to collect families with X-linked retinal dystrophies, other retinopathies and AMD, and collaborate with other research centers working on these diseases. Additional goals are to perform mutation screening of X-linked retinal dystrophy families.

The Foundation Fighting Blindness

07/01/05 – 06/30/10

Clinical Research Center for the Study of Retinal Degenerative Diseases at the University of Michigan

Role: Co-PI with Dr. John Heckenlively

(Multiple modules)

Module III—"Animal Models"

The focus of this module is on delineating pathways leading to retinal degeneration using mouse models.

Module VI—"Molecular Mechanisms of Pathogenesis of X-Linked Retinal Dystrophies"

The primary goal of this project is to generate mouse models for RP2 and CHM and identify the COD2/RP24 gene(s).

Elmer and Sylvia Sramek Charitable Foundation

09/01/06 – 08/31/09

"Interactive and Integrated Genetic Databases for the Study of

Age-related Macular Degeneration (AMD): The Gladiator 2 project"

The goal of this project is to design and develop databases for integrating clinical information with genetic and microarray data in order to facilitate AMD research.

George M. O'Brien Kidney Research Center,

08/01/05 – 07/31/07

University of Michigan Medical School.

Pilot/ Feasibility Project "Function of ciliary protein RPGR in renal epithelial cells: Possible implications for renal-retinal diseases."

The goal of this project is to determine the function of RPGR in the kidney.

National Science Foundation, DBI 0543272.

10/01/06 – 04/30/08

"Integrated Biological Sequence Data Management."

[P.I. Dr. Jignesh Patel, EECS]