

## Olof H. Sundin, Ph.D.

Dr. Sundin is interested in the processes that regulate growth and optical function of the eye. He is trained in biochemistry, molecular virology and developmental biology. His laboratory has recently identified genes underlying defects of color vision, as well as the first gene that appears to function primarily in determining axial length, a key refractive property of the human eye. Currently, he is also pursuing collaborative studies with the Cornea Division on the human genetics of corneal dystrophy.

### Education

Yale University, New Haven, CT , BS 1976  
Massachusetts Institute of Technology, Ph.D. 1981  
Cold Spring Harbor Laboratory, New York. Postdoctoral Fellow, 1981-1984.  
MRC Laboratory of Molecular Biology, Cambridge, UK. Postdoctoral Fellow, 1985-1987.  
Dept. Physiology, Harvard Medical School, Postdoctoral Fellow, 1987-1991.  
Dept. Biochemistry, Baylor college of Medicine, Research Associate, 1991-1992.  
Wilmer Ophthalmological Institute, Assistant Professor 1992.

### Appointments

Assistant Professor, Department of Ophthalmology  
Joint Appointment, Department of Molecular Biology and Genetics

### Teaching

*Principles of Developmental Biology*, first year medical student curriculum. Core faculty.

### Honors, Awards

<i>magna cum laude</i> , Belknap Prize in Biology,	Yale University
NSF Graduate Fellow,	National Science Foundation
Jane Coffin Childs Fellow,	Jane Coffin Childs Fund for Medical Research
Career Development Award,	Research to Prevent Blindness
Lew Wasserman Merit Award,	Research to Prevent Blindness

### Advisory Committees, Review Groups

<i>Ophthalmic Genetics</i> , Editorial Board member	1996-2005
Foundation for Retinal Research, Scientific Advisory Board	1998-present
NIH VisA Study Section, <i>ad hoc</i> reviewer.	2002
NIH VisA/AED Study Section, regular member	2002 - 2003

### Recent Publications:

1. Li, H-S. , Yang, J-M., Jacobson, R., Pasko, D. and Sundin, O. (1994). Pax-6 is first expressed in a region of ectoderm anterior to the early neural plate: Implications for stepwise determination of the lens. *Developmental Biology* **162**: 181-19.
2. Tomarev, S. , Sundin, O., Banerjee-Basu, S., Duncan, M.K., Yang, J-M., Piatigorsky, J. (1996) Chicken homeobox gene Prox 1, related to *Drosophila prospero*, is expressed in the developing lens and retina. *Developmental Dynamics* **206**: 354-367.

3. Koroma, B. M., Yang, J-M., and Sundin, O. (1997). The Pax-6 homeobox gene is expressed throughout the corneal and conjunctival epithelia. *Investigative Ophthalmology and Visual Science* **38**: 108-120.
4. Belecky-Adams, T., Tomarev, S., Li, H-S., Ploder, L., McInnes, R., Sundin, O and Adler, R. Prox-1, Pax-6 and CHX10 Homeobox Gene Expression Correlate with Phenotypic Fate of Retinal Precursor Cells. *Investigative Ophthalmology and Visual Science* **38**, 1293-1303.
5. Sundin, OH. (1998). Embryology of the eye and the role of developmental genes. Chapter 1 in Genetic Diseases of the Eye, a Textbook and Atlas. edited by Elias Traboulsi. **Oxford University Press. Oxford University Press, New York.**
6. Toy, J., Yang, J-M., Leppert, G., and Sundin, O. (1998). The *Optx2* homeobox gene is expressed in early precursors of the eye and activates retina-specific genes. *Proceedings of the National Academy of Sciences, USA* **95**: 10643-10648.
7. Leppert, G. Yang, J-M., and Sundin, O. (1999) Sequence and location of *SIX3*, a homeobox gene expressed in the human eye. *Ophthalmic Genetics* **20**: 1-15.
8. Toy, J. and Sundin, O. (1999). Expression of the *Optx2* homeobox gene during mouse development. *Mechanisms of Development* **83**: 183-186.
9. Silva, E.D., Yang, J.M., Li, Y., Dharmaraj, S., Sundin, O., and Maumenee, I.H. (2000). A Cone-Rod Homeobox (CRX) null mutation is associated with both Leber's congenital amaurosis and a normal ocular phenotype. *Invest Ophthalmol Vis Sci.* **41**: 2076-9.
10. Sundin, O.H., Yang, J.-M., Li, Y., Zhu, D., Hurd, J.N., Mitchell, T.N., Silva, E.D., Maumenee, I.H. (2000). Genetic basis of total colourblindness among the Pingelapese islanders. *Nature Genetics.* **25**: 289-293.
11. Mutational analysis and clinical correlation in Leber Congenital Amaurosis. (2000). Dharmaraj, S., Silva, E., Pina, A.L., Li, Y., Yang, J.M., Carter, R.C., Loyer, M., El-Hilali, H., Traboulsi, E.I., Sundin, O.H., Zhu, D., Koenekoop, R.K., Maumenee, I.H. *Ophthalmic Genetics* **21**: 135 - 150.
12. Parsa CF, Silva ED, Sundin OH, Goldberg MF, DeJong MR, Sunness JS, Zeimer R, Hunter DG. (2001). Redefining papillorenal syndrome: an undiagnosed cause of ocular and renal morbidity. *Ophthalmology* **108**: 738-749.
13. Brodsky MC, Atreides SP, Fowlkes JL, Sundin OH. (2004) Optic nerve aplasia in an infant with congenital hypopituitarism and posterior pituitary ectopia. *Archives of Ophthalmology* **122**: 125-126.
14. Silva E, Dharmaraj S, Li YY, Pina AL, Carter RC, Loyer M, Traboulsi E, Theodossiadis G, Koenekoop R, Sundin O, Maumenee I. (2004) A missense mutation in *GUCY2D* acts as a genetic modifier in RPE65-related Leber Congenital Amaurosis. *Ophthalmic Genetics* **25**: 205-217.
15. Sinha D, Hose S, Zhang C, Neal R, Ghosh M, O'brien TP, Sundin O, Goldberg MF, Gerald Robison W Jr, Russell P, Lo WK, Samuel Zigler J Jr. (2005) A spontaneous mutation affects programmed cell death during development of the rat eye. *Experimental Eye Research* **80**: 323-335.
16. Posterior polar cataract: genetic analysis of a large family. (2005) Finzi S, Li Y, Sallum JMF, Mitchell TN, Farr A, Sundin O, Maumenee IH. *Ophthalmic Genetics* **26**: 125-130.
17. Gottsch JD, Sundin OH, Liu SH, Jun AS, Broman KW, Stark WJ, Vito EC, Narang AK, Thompson JM, Magovern M. (2005) Inheritance of a novel *COL8A2* mutation defines a

- distinct early-onset subtype of Fuchs corneal dystrophy. *Invest Ophthalmol Vis Sci.* **46**:1934-1939.
18. Sundin OH, Leppert GS, Silva ED, Yang JM, Dharmaraj S, Maumenee IH, Santos LC, Parsa CF, Traboulsi EI, Broman KW, Dibernardo C, Sunness JS, Toy J, Weinberg EM. (2005) Extreme hyperopia is the result of null mutations in MFRP, which encodes a Frizzled-related protein. *Proc Natl Acad Sci USA.* **102**: 9553-9558.
  19. Gottsch JD, Zhang C, Sundin OH, Bell WR, Stark WJ, Green WR. (2005). Fuchs corneal dystrophy: aberrant collagen distribution in an L450W mutant of the COL8A2 gene. *Invest Ophthalmol Vis Sci.* **46**: 4504-4511.
  20. Sundin OH. (2005) The Mouse's Eye and Mfrp: Not Quite Human. *Ophthalmic Genet.* **26**: 153-5.
  21. Sundin OH, Jun AS, Broman KW, Liu SH, Sheehan SE, Vito EC, Stark WJ, Gottsch JD. (2006) Linkage of Late-Onset Fuchs Corneal Dystrophy to a Novel Locus at 13pTel-13q12.13. *Invest Ophthalmol Vis Sci.* **47**: 140-145.
  22. Gottsch JD, Sundin OH, Rencs EV, Emmert DG, Stark WJ, Cheng CJ, Schmidt GW. (2006) Analysis and documentation of progression of Fuchs corneal dystrophy with retroillumination photography. *Cornea* (in press).
  23. Abouzeid H, Li Y, Dharmaraj S, Sundin OH, Maumenee IH. (2006) A G1103R mutation in *CRB1* is co-inherited with high hyperopia and Leber congenital amaurosis. *Ophthalmic Genet.* (in press).