

BIOGRAPHICAL SKETCH

Name: Edwin M. Stone, M.D., Ph.D

Position/Title: Professor , Department of Ophthalmology.
Investigator, Howard Hughes Medical Institute

Education/Training

<i>Institution & Location</i>	<i>Degree</i>	<i>Years</i>	<i>Field of Study</i>
Rice University	B.A.	1978	Biology and English
Baylor College of Medicine	Ph.D.	1983	Cell Biology
Baylor College of Medicine	M.D.	1985	Medicine

Research & Professional Experience

Intern (Transitional)	St. Joseph Hospital, Houston	7/85-6/86
Resident (Ophthalmology)	The University of Iowa	7/86-6/89
Fellow-Associate (Ophthalmology)	The University of Iowa	7/89-12/89
Fellow (Ophthalmology Research)	The University of Iowa	1/90-6/90
Fellow (Vitreoretinal Surgery)	The University of Iowa	7/90-2/92
Assistant Professor of Ophthalmology	The University of Iowa	7/90-7/94
Associate Professor of Ophthalmology	The University of Iowa	7/94-7/97
Professor of Ophthalmology	The University of Iowa	7/97-present
Investigator	Howard Hughes Medical Inst.	8/02-present

Honors

Sigma Xi Award for Ph.D. Thesis Excellence, 1983
The DeBakey Scholar, 1984
Alpha Omega Alpha, 1985
Outstanding Intern, Department of Medicine, St. Joseph Hospital, 1986
P. J. Leinfelder Award for Ophthalmology Resident Research, 1989
P. J. Leinfelder Award for Ophthalmology Fellow Research, 1990
Research to Prevent Blindness Dolly Green Scholar, 1993
Alcon Research Institute Award, 1995
The Chibret Award (Switzerland), 1995
Prix Porphyrogenis (Switzerland), 1996
Prix Tissières (Switzerland), 1996
Lew Wasserman Merit Award (Research to Prevent Blindness), 1997
Rosenthal Award (The Macula Society), 1998
Cogan Award (Association for Research in Vision and Ophthalmology, ARVO), 1998
Louis Rudin Glaucoma Prize (New York Academy of Sciences), 1999
The Gregg Lecture (The Royal Australian College of Ophthalmology), 1999
The Inaugural Davson Lecture (The Institute for Ophthalmology, London), 2000
The Doyne Lecture (The Oxford Congress), 2000
The Krill Lecture (The Chicago Ophthalmological Society), 2000

Papers & Chapters

- 1) Stone, E.M., Nichols, B.E., Streb, L.M., Kimura A.E., Sheffield, V.C., Genetic Linkage of Vitelliform Macular Degeneration (Best's Disease) to Chromosome 11q13, Nature Genetics, (1) 246-250, 1992.

- 2) Sheffield, V.C., Stone, E.M., Alward, W.L.M., Drack, A.V., Johnson, A.T., Streb, L.M., Nichols, B.E., Genetic Linkage of Familial Open Angle Glaucoma to Chromosome 1q21-q31, *Nature Genetics*, (4) 47-50, 1993.
- 3) Kwitek-Black, A.E., Carmi, R., Duyk, G.M., Buetow, K.H., Elbedour, K., Parvari, R., Yandava, C.N., Stone, E.M., Sheffield, V.C., Linkage of Bardet-Biedl Syndrome to Chromosome 16q and Evidence for Non-Allelic Genetic Heterogeneity, *Nature Genetics*, (5) 392-396, 1993.
- 4) Stone, E.M., Mathers, W.D., Rosenwasser, G., Holland, E., Folberg, R., Krachmer, J.H., Nichols, B.E., Gorevic, P.D., Taylor, C., Streb, L.M., Fishbaugh, J.A., Daley, T.E., Sucheski, B., Sheffield, V.C., Three Autosomal Dominant Corneal Dystrophies Map to Chromosome 5q, *Nature Genetics*, (6) 47-51, 1994.
- 5) Héon, E., Mathers, W.D., Alward, W.L.M., Weisenthal, R., Sunden, S.L.F., Fishbaugh, J., Taylor, C.M., Krachmer, J.H., Sheffield, V.C., Stone, E.M., Linkage of Posterior Polymorphous Corneal Dystrophy to 20q11, *Human Molecular Genetics*, (4) 485-488, 1995.
- 6) Brown, D.M., Graemiger, R.A., Hergersber, M., Schinzel, A., Messmer, E.P., Niemeyer, G., Schneeberger, S.A., Streb, L.M., Taylor, C.M., Kimura, A.E., Weingeist, T.A., Sheffield, V.C., Stone, E.M., Genetic Linkage of Wagner Disease and Erosive Vitreoretinopathy to Chromosome 5q13-14, *Archives of Ophthalmology*, (113) 671-675, 1995.
- 7) Jacobson, S.G., Cideciyan, A.V., Regunath, G., Rodriguez, F.J., Vandenberg, K., Sheffield, V.C., Stone, E.M., Night Blindness in Sorsby's Fundus Dystrophy TIMP-3-associated is Reversed by Vitamin A Therapy, *Nature Genetics*, (11) 27-32, 1995.
- 8) Héon, E., Piguet, B., Munier, F., Sneed, S.R., Morgan, C.M., Forni, S., Schorderet, D., Taylor, C.M., Streb, L.M., Wiles, C.D., Nishimura, D.Y., Sheffield, V.C., Stone, E.M., Linkage of Autosomal Dominant Radial Drusen (Malattia Leventinese) to Chromosome 2p16-21, *Archives of Ophthalmology*, (114) 193-198, 1996.
- 9) Sheffield, V.C., Kraiem, Z., Beck, J.C., Nishimura, D., Stone, E.M., Salameh, M., Sadeh, O., Glaser, B., Pendred Syndrome Maps to Chromosome 7q21-34 and Is Caused by an Intrinsic Defect in Thyroid Iodine Organification, *Nature Genetics*, (12) 424-426, 1996.
- 10) Scott, D.A., Carmi, R., Elbedour, K., Stone, E.M., Sheffield, V.C., An Autosomal Recessive Nonsyndromic-Hearing-Loss Locus Identified by DNA Pooling Using Two Inbred Bedouin Kindreds, *American Journal of Human Genetics*, (59) 385-391, 1996.
- 11) Stone, E.M., Fingert, J.H., Alward, W.L.M., Nguyen, T.D., Polansky, J.R., Sunden, S.L.F., Nishimura, D., Clark, A.F., Nystuen, A., Nichols, B.E., Mackey, D.A., Ritch, R., Kalenak, J.W., Craven, E.R., Sheffield, V.C., Identification of a Gene Causing Primary Open Angle Glaucoma (GLC1A), *Science*, (275) 668-670, 1997.
- 12) Freund, C.L., Wang, Q.-L., Chen, S., Muskat, B.L., Wiles, C.D., Sheffield, V.C., Jacobson, S.G., McInnes, R.R., Zack, D.J., Stone, E.M., De Novo Mutations In The CRX Homeobox Gene Associated With Leber Congenital Amaurosis, *Nature Genetics*, (18) 311-312, 1998.
- 13) Scott, D.A., Kraft, M.L., Stone, E.M., Sheffield, V.C., Smith, R.J., Connexin Mutations and Hearing Loss, *Nature*, (391) 32, 1998.

- 14) Alward, W.L.M., Fingert, J.H., Coote, M.A., Johnson, A.T., Lerner, S.F., Junua, D., Durcan, F.J., McCartney, P.J., Mackey, D.A., Sheffield, V.C., Stone, E.M., Clinical Features Associated with Mutations in the Chromosome 1 Open-Angle Glaucoma Gene (GLC1A), *The New England Journal of Medicine*, (338) 1022-1027, 1998.
- 15) Nishimura, D.Y., Swiderski, R.E., Alward, W.L.M., Searby, C.C., Patil, S.R., Bennet, S.R., Kanis, A.B., Gastier, J.M., Stone, E.M., Sheffield, V.C., The Forkhead Transcription Factor Gene FKHL7 is Responsible for Glaucoma Phenotypes Which Map to 6p25, *Nature Genetics*, (19) 140-147, 1998.
- 16) Stone, E.M., Webster, A.R., Vandeburgh, K., Streb, L.M., Hockey, R.R., Lotery, A.J., Sheffield, V.C., Allelic Variation in ABCR Associated with Stargardt Disease but not with Age-Related Macular Degeneration, *Nature Genetics*, (20) 328-329, 1998.
- 17) Fingert, J.H., Héon, E., Liebmann, J.M., Yamamoto, T, Craig, J.E., Rait, J., Kawase, K., Hoh, S., Buys, Y.M., Dickinson, J., Williams-Lyn, D., Trope, G., Kitazawa, Y., Ritch, R., Mackey, D.A., Alward, W.L.M., Sheffield, V.C., Stone, E.M., Analysis of Myocilin Mutations in 1703 Glaucoma Patients from Five Different Populations, *Human Molecular Genetics*, (8) 899-905, 1999.
- 18) Stone, E.M., Lotery, A.J., Munier, F.L., Héon, E., Piguet, B., Guymer, R.H., Vandeburgh, K., Cousin, P., Nishimura, D., Swiderski, R.E., Silvestri, G., Mackey, D.A., Hageman, G.S., Bird, A.C., Sheffield, V.C., Schorderet, D.F., A Single EFEMP1 Mutation Associated with both Malattia Leventinese and Doyme Honeycomb Retinal Dystrophy, *Nature Genetics*, (22) 199-202, 1999.
- 19) ElShanti, H., AlSalem, M., EINajjar, M., Ajlouni, K., Beck, J., Sheffield, V.C., Stone, E.M., A Nonsense Mutation in the Retinal Specific Guanylate Cyclase Gene is the Cause of Leber Congenital Amaurosis in a Large Inbred Kindred from Jordan, *Journal of Medical Genetics*, (36) 862-865, 1999.
- 20) Haider, N.B., Jacobson, S.G., Cideciyan, A.V., Swiderski, R., Streb, L.M., Searby, C., Beck, G., Hockey, R., Hanna, D.B., Gorman, S., Duhl, D., Carmi, R., Bennett, J., Weleber, R.G., Fishman, G.A., Wright, A.F., Stone, E.M., Sheffield, V.C., Mutation of a Nuclear Receptor Gene, NR2E3 Causes Enhanced S Cone Syndrome, A Disorder of Retinal Cell Fate, *Nature Genetics*, (24) 127-131, 2000.
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- 22) Slavotinek, A.M., Stone, E.M., Mykvtyn, K., Heckenlively, J.R., Green, J.S., Héon E., Musarella, M.A., Parfrey, P.S., Sheffield, V.C., Biesecker, L.G., Mutations in MKKS cause Bardet-Biedl Syndrome, *Nature Genetics*, (26) 14-16, 2000.
- 23) Lotery, A.J., Jacobson, S.G., Fishman, G.A., Weleber, R.G., Fulton, A.B., Namperumalsamy, P., Heon, E., Levin A.V., Grover, S., Rosenow, J.R., Kopp, K.K., Sheffield V.C., Stone, E.M., Mutations in the CRB1 Gene Cause Leber Congenital Amaurosis, *Archives of Ophthalmology*, (119) 415-420, 2001.
- 24) Nishimura, D.Y., Searby, C.C., Carmi, R., Elbedour, K., Van Maldergem, L., Fulton, A.B., Lam, B.L., Powell, B.R., Swiderski, R.E., Bugge, K.E., Haider, N.B., Kwitek-Black, A.E., Ying, L., Duhl, D.M., Gorman, S.W., Héon, E., Iannaccone, A., Bonneau, D., Biesecker, L.G., Jacobson, S.G., Stone, E.M., Sheffield, V.C., Positional Cloning of a Novel Gene on Chromosome 16q causing Bardet-Biedl Syndrome (BBS2), *Human Molecular Genetics*, (10) 865-874, 2001.

- 25) Lotery A.J., Derksen, T.A., Russell, S.R., Mullins, R.F., Sauter, A., Affatigato, L.M., Stone, E.M., Davidson, B.L. Gene Transfer to the Nonhuman Primate Retina with Recombinant Feline Immunodeficiency Virus Vectors Human Gene Therapy (13) 689-696, 2002.
- 26) Milam, A.H., Rose, L., Cideciyan, A.V., Barakat, M.R., Tang, W.-X., Gupta, N., Aleman, T.S., Wright, A.F., Stone, E.M., Sheffield, V.C., Jacobson, S.G., The Nuclear Receptor NR2E3 Plays a Role in Human Retinal Photoreceptor Differentiation and Degeneration, Proceedings of the National Academy of Sciences, (1) 473-478, 2002.
- 27) Mykytyn, K., Nishimura, D.Y., Searby, C.C., Shastri, M., Yen, H.J., Beck, J.S., Braun, J.S., Streb, L.M., Cornier, A.S., Cox, G.F., Fulton, A.B., Carmi, R., Luleci, G., Chandrasekharappa, S.C., Colins, F.S., Jacobson, S.G., Heckenlively, J.R., Weleber, R.G., Stone, E.M., Sheffield, V.C., Identification of the Gene (BBS1) Most Commonly Involved in Bardet-Biedl Syndrome, a Complex Human Obesity Syndrome, Nature Genetics, (4) 435-438, 2002.
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- 29) Lotery, A.J., Yang, G.S., Mullins, R.F., Russell, S.R., Schmidt, M., Stone, E.M., Lindbloom, J.D., Chiorini, J.A., Kotin, R.M., Davidson, B.L., Adeno-associated Virus Type 5: Transduction Efficiency in Cell-type Specificity in the Primate Retina, Human Gene Therapy (14), 1663-1671, 2003.
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- 31) Chiang, A.P., Nishimura, D., Searby, C., Elbedour, K., Carmi, R., Ferguson, A.L., Secrist, J., Braun, T., Casavant, T., Stone, E.M., Sheffield, V.C., Comparative Genomic Analysis Identifies an ADP-Ribosylation Factor-like Gene as the Cause of Bardet-Biedl Syndrome (BBS3), American Journal of Human Genetics, (75) 475-84, 2004.