COLLEGE OF MEDICINE CURRICULUM VITAE
Val C. Sheffield, M.D., Ph.D.
April 2018

I. EDUCATIONAL AND PROFESSIONAL HISTORY

A. List of institutions attended:

1974	B.S. Zoology	Brigham Young University, Provo, Utah
1977	M.S. Devel. Biol	Brigham Young University, Provo, Utah
1983	Ph.D. Devel. Biol	University of Chicago, Chicago, Illinois
1985	M.D. with Honors	University of Chicago, Chicago, Illinois
1985-87	Pediatric Resident	University of California, San Francisco
1987-90	Fellowship	Medical Genetics, Univ of Calif, San Francisco

Certification and licensure:

1990	Permanent Iowa license to practice medicine and surgery
1990	Diplomate of the National Board of Medical Examiners
1990	Board certified in Clinical Medical Genetics
2010-2019	Board certified in Clinical Molecular Genetics

B. Professional and academic positions held:

1990-1994	Assistant Professor	University of Iowa, College of Medicine, Department of Pediatrics
1994-1998	Associate Professor	University of Iowa, College of Medicine, Department of Pediatrics
1997-present	Director	University of Iowa, Interdepartmental Research Program in Human Molecular Genetics
1997-present	Director	University of Iowa, Department of Pediatrics Division of Medical Genetics
1998-2003	Assoc. Investigator	Howard Hughes Medical Institute
1998-present	Professor	University of Iowa, College of Medicine Department of Pediatrics
2003-2015	Investigator	Howard Hughes Medical Institute
2006-2015	Chair	Martin and Ruth Carver Chair in Genetics Carver
2015-present	Chair	Roy J. Carver Chair in Molecular Genetics, College of Medicine, University of Iowa

C. Honors, Awards, Recognitions, Outstanding Achievements:

1985	Recipient of the F. Howell Wright Award for outstanding performance in Pediatrics from the University of Chicago
1985	Graduated with Honors from the University of Chicago Pritzker School of Medicine
1988	Recipient of Young Alumnus Achievement Award from Brigham Young University
1988	Recipient of Carver Clinician Scientist Award from University of Iowa
1990	Recipient of Melvin Grumback Research Award for Outstanding Pediatric Research from University of California, San Francisco
1997	Brigham Young University Service to Family Award
1998	Rudin Award for Outstanding Research in Glaucoma
2003	E. Mead-Johnson Award for Pediatric Research
2005	Elected to the Institute of Medicine of the National Academies

II. TEACHING

A. Teaching assignments on semester by semester basis:

1976	Teaching Assistant in Genetics, Brigham Young University
1980, 1981	Teaching Assistant in Development Biology, University of Chicago
1987, 1988, 1989	Teaching Assistant in Genetics, University of California, San Francisco
1994, 1996, 1998	Human Molecular Genetics Course, University of Iowa
1995 – present	Lecturer, Medical Genetics Course, University of Iowa
2001 – 2010	Course Director, M1 Medical Genetics Course, University of Iowa

B. Graduate Students Supervised:

Anne Kwitek-Black, Ph.D.
Daryl Scott, M.D., Ph.D.
Arne Nystuen, Ph.D.
Neena Haider, Ph.D.
Terry Braun, Ph.D.
Hsan-Jan Yen, Ph.D.
Marwan Tayeh, Ph.D.
Roger Davis
Annie Chiang, Ph.D.
Kwang-Youn Kim, Ph.D.
Pamela Pretorious, Ph.D.
Yan Zhang, Ph.D.
Xitiz Chamling, Ph.D.
Calvin Carter, Ph.D.
Katie Weihbrecht, Ph.D.
Xiaolei Lin
Ying Hsu
Wes Goar
Matt Cring
Sunny Huang
Thomas Pak

C. Other contributions to institutional programs:

1993-1996	Member of the Executive Committee for the University of Iowa Genetics PhD Program
1993	Member of Interim Planning Committee for the Genetics PhD Program
1993	Chairman of the Admissions Committee for the Genetics PhD Program
1994-1998	Member of University of Iowa College of Medicine Research Review Committee
1995-present	Member of the University of Iowa Research Advisory Committee
1995-present	Transgenic Animal Facility Advisory Committee
1995-present	Member of University of Iowa College of Medicine Research Advisory Committee
1995-present	University of Iowa Howard Hughes Medical Institute Grant Planning and Supervisory Committee
2000-present	Member of Association of American Physicians
2000-present	Member of American Pediatric Society
2000-present	Member of Society of Pediatric Research

III. SCHOLARSHIP

A. Publications or creative works:

1. **Sheffield VC.** Seegmiller RE. Impaired energy metabolism as an initial step in the mechanism for 6-aminonicotinamide induced limb malformation. *J Embryol Exp Morph* 1980 59:217-222
2. Vuorio E, Sandell L, Kravis D, **Sheffield VC**, Vuorio T, Dorfman A, Upholt WB. Construction and partial characterization of two recombinant cDNA clones for procollagen from chicken cartilage. *Nucleic Acids Res* 1982 25;10(4):1175-92
3. Upholt W, Kravis D, Sandell L, **Sheffield VC**, Dorfman A. Gene expression during chick limb cartilage differentiation. *Limb Development and Regeneration* 1982 110 Pt B:175-82
4. **Sheffield VC,** Upholt WB. Copy number of the chicken type II procollagen gene. *Coll Relat Res* 1985 5(1):1-8
5. **Sheffield VC**, Cox DR, Lerman LS, Myers RM. Attachment of a 40-base-pair G+C-rich sequence (GC-clamp) to genomic DNA fragments by the polymerase chain reaction results in improved detection of single-base changes. *Proc Natl Acad Sci USA* 1989 89:232-36
6. **Sheffield VC**, Fishman GA, Beck JS, Kimura AE, Stone EM. Identification of novel rhodopsin mutations associated with retinitis pigmentosa by GC-clamped denaturing gradient gel electrophoresis. *Am J Hum Genet* 1991 49:699-706
7. Metzger A, **Sheffield VC**, Duyk G, Daneshvar L, Edwards MS, Cogen PH. Identification of a germline mutation in the p53 gene in a patient with an intracranial ependymoma. *Proc Natl Acad Sci USA* 1991 88:7825-7829
8. Stone EM, Fishman GA, Kimura AE, Khadivi P, **Sheffield VC.** Regional distribution of retinal degeneration in patients with the proline to histidine mutation in codon 23 of the rhodopsin gene. *phthalmology* 1991 98:1806-1813
9. Fishman GA, Stone EM, Gilbert LD, Kenna P, **Sheffield VC**. Ocular findings associated with a rhodopsin gene codon 58 transversion mutation in autosomal dominant retinitis pigmentosa. *Arch Ophthalmology* 1991 109:1387-1393
10. Fishman GA. Stone EM, **Sheffield VC**, Gilbert LD, Kimura AE. Ocular findings associated with rhodopsin gene codon 17 and codon 182 transition mutations in dominant retinitis pigmentosa. *Arch Ophthalmology* 1992 110:54-62
11. **Sheffield VC**, Beck J, Nichols B, Cousineau A, Lidral AC, Stone EM. Detection of multi-allele polymorphisms within gene sequences by GC-clamped denaturing gradient gel electrophoresis. *Am J Hum Genet* 1992 50:567-575
12. **Sheffield VC**, Beck JS, Stone EM, Myers RM. A simple and efficient method for attachment of a 40 base pair GC-rich sequence to PCR-amplified DNA. *Biotechniques* 1992 12(3):386-388
13. Cogen PH, Daneshvar L, Metzger AK, Duyk G, Edwards MSB, **Sheffield VC.** Involvement of multiple chromosome 17p loci in medulloblastoma tumorigenesis. *Am J Hum Genet* 1992 50:584-589
14. Fishman GA, Stone EM, Gilbert LD, **Sheffield VC**. Ocular findings associated with a rhodopsin gene codon 106 mutation: Glycine–to-Arginine change in autosomal dominant retinitis pigmentosa. *Arch Ophthalmology* 1992 110:646-653
15. Stone E, Nichols B, Streb L, Kimura A, **Sheffield VC.** Genetic linkage of vitelliform macular degeneration (Best Disease) to chromosome 11q13. *Nature Genet* 1992 1:246-250
16. Ferrell L, Schmidt K, **Sheffield V**, Packman S. Neonatal hemochromatosis: Genetic counseling based on retrospective pathologic diagnosis. *Am J Med Genet* 1992 44:429-433
17. Brown DM, Nichols B, Weingeist T, **Sheffield V**, Kimura A, Stone E. Procollagen II gene mutation in Stickler syndrome. *Arch Ophthalmology* 1992 110:1589-1593
18. Stone EM, Kimura AE, Folk JC, Bennett SR, Nichols BE, Streb LM, **Sheffield VC.** Genetic linkage of autosomal dominant neovascular inflammatory vitreoretinopathy to chromosome 11q13. *Hum Mol Genet* 1992 1(9):685-689
19. Freneaux E, **Sheffield VC**, Molin L, Bedell K, Kutschke W, Reece C, Shires A, Rhead W. Glutaric acidemia type II: Heterogeneity in β-oxidation flux, polypeptide synthesis and complementary DNA mutations in the α-subunit of electron transfer flavoprotein in eight patients. *J Clin Invest* 1992 90:1679-1886

20. Buetow KH, **Sheffield VC**, Zhu M, Zhou T, Shen F, Hino O, Smith M, McMahon BJ, Lanier AP, London WT, Redeker AG, Govindarajan S. Low frequency of p53 mutations observed in diverse collection of primary hepatocellular carcinomas. *Proc Natl Acad Sci* 1992 89(20):9622-9626
21. Fishman GA, Vandenburgh K, Stone EM, Gilbert LD, Alexander KR, **Sheffield VC**. Ocular findings associated with rhodopsin gene codon 267 and codon 190 mutations in dominant retinitis pigmentosa. *Arch Ophthalmology* 1993 110:1582-1588
22. **Sheffield VC.** Stone EM, Alward WLM, Drack AV, Johnson AT, Streb LM, Nichols BE. Genetic linkage of familial open angle glaucoma to chromosome 1q21-q31. *Nature Genet* 1993 4:47-50
23. Beck JS, Kwitek AE, Cogen P, Metzker AK, Duyk G, **Sheffield VC**. A denaturing gradient gel assay for sensitive detection of p53 mutations. *Hum Genet* 1993 91:25-30
24. Nichols BE, **Sheffield VC**, Vandenburgh K, Drack AV, Kimura AE, Stone EM. Butterfly-shaped pigment dystrophy of the fovea caused by a point mutation in codon 167 of the RDS gene. *Nature Genet* 1993 3:202-207
25. **Sheffield VC.** Beck JS, Kwitek AE, Sandstrom DW, Stone EM. The sensitivity of single strand conformation polymorphism analysis for the detection of single base substitutions. *Genomics* 1993 16:325-332
26. Nichols BE, Drack AV, Vandenburgh K, Kimura AE, **Sheffield VC**, Stone EM. A 2 base pair deletion in the RDS gene associated with butterfly-shaped pigment dystrophy of the fovea. *Hum Mol Genet* 1993 2(5):601-603
27. Weleber RG, Carr RE, Murphey WH, **Sheffield VC**, Stone EM. Phenotypic variation including retinitis pigmentosa, pattern dystrophy, and fundus flavimaculatus in a single family with a deletion of codon 153 or 154 of the peripherin/RDS gene. *Arch Ophthalmol* 1993 111:1531-1542
28. Ibraghimov-Beskrovnaya O, **Sheffield VC**, Campbell KP. Single base polymorphism in the DAG1 gene detected by DGGE and mismatch PCR. *Hum Mol Genet* 1993 2(11)1983
29. Kwitek-Black AE, Carmi R, Duyk GM, Buetow KH, Elbedour K, Parvari R, Yandava CN, Stone EM, **Sheffield VC.** Linkage of Bardet-Biedl syndrome to chromosome 16q and evidence for non-allelic genetic heterogeneity. *Nature Genet* 1993 5(4):392-396
30. Nichols BE, **Sheffield VC**, Stone EM. A user-friendly hypercard interface for human linkage analysis. *Comput Appl Biosci* 1993 9(6):757-759
31. Stone EM, Mathers WD, Rosenwasser G, Holland E, Folberg R, Krachmer JH, Nichols BE, Gorevic PD, Taylor C, Streb LM, Fishbaugh JA, Daley TE, Sucheski B, **Sheffield VC.** Three autosomal dominant corneal dystrophies map to chromosome 5q. *Nature Genet* 1993 6:47-51
32. Nichols BE, Bascom R, Litt M, McInnes R, **Sheffield VC**, Stone EM. Refining the locus for Best's vitelliform macular dystrophy and mutation analysis of the candidate gene ROM1. *Am J Hum Genet* 1994 54:95-103
33. Cousineau AJ, Lauer RM, Pierpont ME, Burns TL, Ardinger RH, Patil SR, **Sheffield VC.** Linkage analysis of autosomal dominant atrioventricular canal defects: Exclusion of chromosome 21. *Human Genet* 1994 93:103-108
34. Buetow KH, Weber JL Ludwigsen S, Scherpbier-Heddema T, Duyk G, **Sheffield VC**, Wang Z, Murray JC. Integrated human genome-wide maps constructed using the CEPH reference panel. *Nature Genet* 1994 6(4):391-393
35. Stone EM, Nichols BE, Kimura AE Weingeist TA, Drack A, **Sheffield VC.** Clinical features of a Stargardt-like dominant progressive macular dystrophy with genetic linkage to chromosome 6q. *Arch Ophthalmol* 1994 112:765-772
36. Folberg R, Stone EM, **Sheffield VC**, Mathers WD. The relationship between granular, lattice type 1, and avellino corneal dystrophies: A histopathologic study. *Arch Ophthalmol* 1994 112:1080-1085
37. **Sheffield VC**, Carmi R, Kwitek-Black A, Rokhlina T, Nishimura D, Duyk GM, Elbedour K, Sunden SL, Stone EM. Identification of a Bardet-Biedl syndrome locus on chromosome 3 and evaluation of an efficient approach to homozygosity mapping. *Hum Mol Genet* 1994 3(8):1331-1335
38. Murray, JC, Buetow KH, Weber JL, Ludwigsen S, Scherpbier-Heddema T, Manion F, Quillen J, **Sheffield VC**, Sunden S, Duyk GM, Weissenbach J, Gyapay G, Dib C, Morrissette J, Lathrop GM, Vignal A, White R, Matsunami N, Gerken S, Melis R, Albertsen H, Plaetke R, Odelberg S, Ward D, Dausset J, Cohen D, Cann H. A comprehensive human linkage map with centimorgan density. *Science* 1994 265:2049-2054

39. Kemp CM, Jacobson SG, Cideciyan AV, Kimura AE, **Sheffield VC**, Stone EM. RDS gene mutations causing retinitis pigmentosa or macular degeneration lead to the same abnormality in photoreceptor function. *Invest Ophth Vis Sci* 1994 35:3154-3162
40. Buetow KH, Ludwigsen S, Scherpbier-Heddema T, Quillen J, Murray JC, **Sheffield VC**, Duyk GM, Weber JL, Weissenbach J, Gyapay G, Dib C, Vignal A, Morrissette J, Lathrop GM, White R, Matsunami N, Gerken S, Mells R, Albertsen H, Ward K, Plaetke R, Odelberg S, Ward D, Bray-Ward P, Menninger A, Lieman L, Desai T, Banks A. Human genetic map. Genome maps V. Wall chart. *Science* 1994 30;265(5181):2055-70
41. Fishman GA, Stone E, Gilbert LD, Vandenburgh K, **Sheffield VC**, Heckenlively JR. Clinical features of a previously undescribed codon 216 (proline to serine) mutation in the peripherin/RDS gene in autosomal dominant retinitis pigmentosa. *Ophthalmology* 1994 101:1409-1421
42. Carmi R, Rokhlina T, Kwitek-Black AE, Elbedour K, Nishimura D, Stone EM, **Sheffield VC.** Use of a DNA pooling strategy to identify a human obesity syndrome locus on chromosome 15. *Hum Mol Genet* 1995 4(1):9-13
43. Lam BL, Vandenburgh K, **Sheffield VC**, Stone EM. Retinitis pigmentosa associated with a dominant mutation in codon 46 of the peripherin/RDS gene (Arginine-46-Stop) *Am J Ophthalmol* 1995 119:65-71
44. Brown DM, Vandenburgh K, Kimura AE, Weingeist TA, **Sheffield VC**, Stone EM. Novel frameshift mutations in the procollagen 2 gene (COL2A1) associated with Stickler syndrome (hereditary arthro-ophthalmopathy). *Hum Mol Genet* 1995 4:141-142
45. Heon E, Mathers W, Alward WLM, Weisenthal R, Sunden S, Fishbaugh J, Taylor CM, Krachmer JH, **Sheffield VC**, Stone EM.Linkage of posterior polymorphous corneal dystrophy to 20q11. *Hum Mol Genet* 1995 4(3):485-488
46. Dubovsky J, **Sheffield VC**, Duyk GM, Weber JL. Sets of short tandem repeat polymorphisms for efficient linkage screening of the human genome. *Hum Mol Genet* 1995 4(3):449-452
47. **Sheffield VC.** Nishimura DY, Stone EM. Novel approaches to linkage mapping. *Cur Opin Genet Dev* 1995 5:335-341
48. Brown DM, Graemiger RA, Hergersberg M, Schinzel A, Messmer EP, Niemeyer G, Schneeberger SA, Streb LM, Taylor CM, Kimura AE, Weingeist TA, **Sheffield VC**, Stone EM. Genetic linkage of Wagner disease and erosive vitreoretinopathy to chromosome 5q13-14. *Arch Opththalmol* 1995 113:671-675
49. Heon E, Sheth BP, Kalenak JW, Sunden SLF, Streb LM, Taylor CM, Alward WLM, **Sheffield VC**, Stone EM. Linkage of autosomal dominant iris hypoplasia to the region of the Rieger syndrome locus (4q25). *Hum Mol Genet* 1995 4(8):1435-1439
50. Terrell RB, Wille AH, Cheville JC, Nystuen AM, Cohen MB, **Sheffield VC.** Microsatellite instability in adenocarcinoma of the prostate. *Am J Pathol* 1995 147(3):799-805
51. Scott DA, Carmi R, Elbedour K, Duyk GM, Stone EM, **Sheffield VC.** Non-syndromic autosomal recessive deafness is linked to the DFNB1 Locus in a large inbred Bedouin family from Israel. *Am J Hum Genet* 1995 57:965-968
52. **Sheffield VC,** Weber JL, Buetow KH, Murray JC, Even DA, Wiles K, Gastier JM, Pulido JC, Yandava C, Sunden SL, Mattes G, Businga T, McClain A, Beck J, Scherpier T, Gilliam J, Zhong J, Duyk G. A collection of tri- and tetranucleotide repeat markers used to generate high quality, high resolution human genome-wide linkage maps. *Hum Mol Genet* 1995 4(10):1837-1844
53. Gorin MB, Jackson KE, Ferrell RE, **Sheffield VC**, Jacobson SG, Gass JDM, Mitchell E, Stone EM. A peripherin/RDS mutation (Pro-210-Arg) associated with macular and peripheral retinal degeneration. *Ophthalmology* 1995 102:246-255
54. Jacobson SG, Cideciyan AV, Regunath G, Rodriguez FJ, Yandenburgh K, **Sheffield VC**, Stone EM. Night blindness in Sorsby's fundus dystrophy reversed by vitamin A. *Nature Genet* 1995 11:27-32
55. El-Sayed NM, Alarcon CM, Beck JC, **Sheffield VC**, Donelson JE. cDNA expressed sequence tags of Typanosoma brucei rhodesiense provide new insights into the biology of the parasite. *Mol Biochem Parasitol* 1995 73:75-90
56. Gastier JM. Pulido JC, Sunden S, Brody T, Buetow KH, Murray JC, Weber JL, Hudson TJ, **Sheffield VC**, Duyk GM. Survey of trinucleotide repeats in the human genome: assessment of their utility as genetic markers. *Hum Mol Genet* 1995 4(10):1829-1836
57. Zittergruen MM, Murray JC, Lauer RM, Burns TL, **Sheffield VC.** Molecular analysis of nondisjunction in Down syndrome patients with and without atrioventricular septal defects. *Circulation* 1995 92(10):2803-10

58. Willert JR, Daneshvar L, **Sheffield VC**, Cogen PH. Deletion of chromosome arm 17p DNA sequences in pediatric high-grade and juvenile pilocytic astrocytomas. *Genes Chromosomes Cancer* 1995 12(3):165-72
59. Carmi R, Elbedour K, Stone EM, **Sheffield VC.** Phenotypic differences among patients with Bardet Biedl syndrome linked to three different chromosome loci. *Am J Med Genet* 1995 59:199-203
60. Alward WLM, Johnson AT, Nishimura DY, **Sheffield VC**, Stone EM. Molecular Genetics of Glaucoma: Current Status. *J Glaucoma* 1996 5:276-284
61. Nystuen AM, Benke P, Merren J, Stone EM, **Sheffield VC**. A cerebellar ataxia locus identified by DNA pooling to search for linkage disequilibrium in an isolated population from the Cayman Islands. *Hum Mol Genet* 1996 5(4):525-531
62. **Sheffield VC**, Kraiem Z, Beck JC, Nishimura D, Stone EM, 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 Genet* 1996 12:424-426
63. Scott DA, Carmi R, Elbedour K, Yosefsberg S, Stone EM, **Sheffield VC**. An autosomal recessive nonsyndromic-hearing-loss locus identified by DNA pooling using two inbred Bedouin kindreds. *Am J Hum Genet* 1996 59(2):385-91
64. Jacobson SG. Cideciyan AV, Kemp CM, **Sheffield VC**, Stone EM. Photoreceptor function in heterozygotes with insertion or deletion mutations in the RDS gene. *Inv Ophthalmol Vis Sci* 1996 37(8):1662-1674
65. Heon E, Piguet B, Munier F, Sneed SR, Morgan CM, Forni S, Schordert D, Taylor CM, Streb LM, Wiles CD, Nishimura DY, **Sheffield VC**, Stone EM. Linkage of autosomal dominant radial drusen (Mallatia Leventinese) to chromsome 2p16-21. *Arch Ophthalmology* 1996 114:193-198
66. Ionasescu V, Searby C, **Sheffield VC**, Rokhlina T, Nishimura DY, Ionasescu R. Autosomal dominant Charcot-Marie-Tooth axonal neuropathy mapped on chromosome 7p (CMT2D) *Hum Mol Genet* 1996 5(9):1373-1375
67. Sunden SLF. Alward WLM, Nichols BE, Rokhlina TR, Nystuen A, Stone EM, **Sheffield VC.** Fine mapping of the autosomal dominant juvenile open angle glaucoma (GLC1A) region and evaluation of candidate genes. *Genome Res* 1996 6(9):862-869
68. Wayne S, Der Kaloustian VM, Schloss M, Polomeno R, Scott DA, Hejtmancik JF, **Sheffield VC**, Smith RJH. Localization of the Usher syndrome type ID gene (Ush1D) to chromosome 10. *Hum Mol Gen* 1996 5(10):1689-1692
69. Wille AH, Terrell RB, Cheville JC, **Sheffield VC**, Cohen MB. Focal microsatellite mutations in relatives with prostatic adenocarcinoma. *Anticancer Res* 1996 16:3883-3886
70. Jacobson SG, Cideciyan AV, Maguire AM, Bennett J, **Sheffield VC**, Stone EM. Preferential rod and cone photoreceptor abnormalities in heterozygotes with point mutations in the RDS gene. *Exp Eye Res* 1996 63:603-608
71. Sunden SLF, Businga T, Beck J, McClain A, Gastier JM, Pulido JC, Yandava CN, Brody T, Ghazizadeh J, Weber JL, Duyk GM, Murray JC, Buetow KH, **Sheffield VC.** Chromosomal assignment of 2900 tri- and tetranucleotide repeat markers using NIGMs somatic cell hybrid panel 2. *Genomics* 1996 32:15-20
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73. Gastier JM, Brody T, Pulido JC, Businga T, Sunden D, Hu X, Maitra S, Buetow KH, Murray JC, **Sheffield VC**, Boguski M, Duyk GM, Hudson TJ. Development of a screening set for new (CAG/CTG)n dynamic mutations. *Genomics* 1996 15;32(1):75-85
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75. Lam BL, Fingert JH, Schutte BC, Singleton EM, Merin LM, Brown HH, **Sheffield VC**, Stone EM. Clinical and molecular characterization of a family affected with X-linked ocular albinism (OA1). *Ophthalmic Genet* 1997 18:175-84
76. Yuan B, Vaske D, Weber JL, Beck J, **Sheffield VC**. Improved set of short-tandem-repeat polymorphisms for screening the human genome. *Am J Hum Genet* 1997 60:459-460

77. Chen A, Wayne S, Bell A, Ramesh A, Srisailapathy CRS, Scott DA, **Sheffield VC**, Van Hauwe P, Zbar RIS, Ashley J, Lovett M, Van Camp G, Smith RJH. New gene for autosomal recessive non-syndromic hearing loss maps to either chromosome 3q or 19p. *Am J Med Genet*1997 5;71(4):467-471
78. Stone EM, Fingert JH, Alward WLM, Nguyen TD, Polansky JR, Sunden SLF, Nishimura D, Clark AF, Nystuen A, Nichols BE, Mackey DA, Ritch R, Kalenak JW, Craven ER, **Sheffield VC**. Identification of a gene that causes primary open angle glaucoma (GLC1A). *Science* 1997 275:668-670
79. **Sheffield VC**, Pierpoint ME, Nishimura D, Beck JS, Burns T, Berg MA, Stone EM, Patil SR, Lauer RM. 7Identification of a complex congenital heart defect susceptibility locus by using DNA pooling and shared segment analysis. *Hum Mol Gen* 1997 6(1) 117-121
80. Nystuen A, Costefff H, Elpeleg ON, Apter N, Bonne-Tamir B, Mohrenweiser H, Haider N, Stone EM, **Sheffield VC.** Iraqi-Jewish kindreds with optic atrophy plus (3-methylglutaconic aciduria type 3) demonstrate linkage disequilibrium with the CTG repeat in the 3' untranslated region of the myotonic dystrophy protein kinase gene. *Hum Mol Genet* 1997 6(4):563-569
81. Arbour NC. Zlotogora J, Knowlton RG, Merin S, Rosenmann A, Kanis AB, Rokhlina T, Stone EM, **Sheffield VC**. Homozygostiy mapping of achromatopsia to chromosome 2 using DNA pooling. *Hum Mol Genet* 1997 6(5):689-694
82. Yandava CN, Gastier JM, Pulido JC, Brody T, **Sheffield VC**, Murray J, Buetow K, Duyk GM. (Characterization of Alu repeats that are associated with trinucleotide and tetranucleotide repeat microsatellites. *Genome Res* 1997 7:716-724
83. Walder RY. Shalev H, Brennan TMH, Carmi R, Elbedour K, Scott DA, Hanauer A, Mark AL, Patil S, Stone EM, **Sheffield VC.** Familial hypomagnesemia maps to chromosome 9q, not to the X chromosome: genetic linkage mapping and analysis of a balanced translocation breakpoint. *Hum Mol Genet* 1997 6(9):1491-1497
84. Brown JW, Fingert JH, Taylor CM, Lake M, **Sheffield VC**, Stone EM. Clinical and genetic analysis of a family affected with dominant optic atrophy (OPA1). *Arch Ophthalmol* 1997 115:95-99
85. Vassileva P, Gieser S, West S, Cholakova T, Vitale S, **Sheffield V**. Prevalence of blindness and visual impairment due to cataract—Sofia Eye Survey. *Dev Ophthalmol* 1997 27:19-24
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87. Bonne-Tamir B, Nystuen A, Seroussi E, Kalinsky H, Kwitek-Black AE, Korostishevsky M, Adato A, **Sheffield VC.** Usher syndrome in the Samaritans: strengths and limitations of using inbred isolated populations to identify genes causing recessive disorders. *Am J Phys Anthropol* 1997 104(2):193-200
88. Weleber RG, Butler NS, Murphey WH, **Sheffield VC**, Stone EM. X-linked retinitis pigmentosa associated with a 2-base pair insertion in codon 99 of the RP3 gene RPGR. *Arch Ophthalmol* 1997 115(11):1429-35
89. Greinwald JH Jr, Scott DA, Marietta JR, Carmi R, Manaligod J, Ramesh A, Zbar RIS, Kraft ML, Elbedour K, Yairi Y, Musy M, Skvoark AB, Van Camp G, Srisailapathy CR, Lovett M, Morton CC, **Sheffield VC**, Smith RJH. Construction of P1-derived artificial chromosome and yeast artificial chromosome contigs encompassing the DFNB7 and DFNB11 region of chromosome 9q13-21. *Genome Res* 1997 7(9):879-86
90. Pannain, S, Weiss RE, Jackson CE, Dian D, Beck JC, **Sheffield VC**, Cox N, Refetoff S. Two different mutations in the thyroid peroxidase gene of a large inbred Amish kindred: power and limits of homozygosity mapping. *J Clin Endocrinol Metab* 1998 84(3):1061-71
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92. Walder RY, Garrett MR, McClain AM, Beck GE, Brennan TMH, Kramer NA, Kanis AB, Mark AL, Rapp JP, **Sheffield VC.** Short tandem repeat polymorphic markers for the rat genome from marker-selected libraries. *Mamm Genome* 1998 9(12):1013-21
93. Stone EM, Webster AR, Vandenburgh K, Streb LM, Hockey RR, Lotery AJ, **Sheffield VC** Allelic variation in ABCR associated with Stargardt disease but not age-related macular degeneration. *Nature Genet* 1998 20(4):328-9
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213. Davis, L.K., K.J. Meyer, E.I. Schindler, J.S. Beck, D.S, Rudd, A.J. Grundstad, T.E. Scheetz, T.A. Braun, J.H. Fingert, J.C. Folk, S.R. Russell, T.H. Wassink, **V.C. Sheffield**, E.M. Stone. A large scale study of copy number variation implicates the gene *DMXL1* and *TULP3* in the etiology of primary open angle glaucoma. ASHG 299, 2009.
214. Schindler, E.I., A.C. Ko, A.V. Drack, A.V. Cideciyan, T.S. Aleman, S.G. Jacobson, R.G. Weleber, G.A. Fishman, **V.C. Sheffield**, E.M. Stone. Assessment of the pathogenic contribution of individual recessive disease alleles in ABCA4-Assicated retinal degeneration. ASHG 448, 2009
215. Meyer, K.J., L.K. Davis, E.I. Schindler, J. Beck, D.S. Rudd, J.A. Grundstad, T.E. Scheetz, T.A. Braun, J.H. Fingert, J.C. Folk, S.R. Russell, T.H. Wassink, E.M. Stone, **V.C. Sheffield**. Genome-wide analysis of copy number variation implicates CHL1 in the etiology of age-related macular degeneration. ASHG 748, 2009
216. Wang, K., T.E. Scheetz, A.J. Grundstad, J.S. Beck, T.L. Casavant, T.A. Braun, J.C. Folk, J.H. Fingert, E.M. Stone. **V.C. Sheffield**. A novel efficient genome-wide associated study design: Application to glaucoma and age-related macular degeneration. ASHG 1013, 2009.
217. Parvari, R., E. Muhammd, G. Harel Levy, A. Levitas, V. Chasa, Caspi, E. Manor, J.C. Beck, A. Saada, **V.C. Sheffield**. Familial isolated cardiomyopathy caused by a mutation in the flavoprotein subunit of succinate dehydrogenase. ASHG 2316, 2009.
218. Vogel, T., D.Y. Nishimura, C.C. Searby, R.E. Swiderski, R.F. Mullins, P.R. Pretorius, S. Seo, Q. Zhang, K. Bugge, D.R. Thedens, M.D. Cassell, J.A. Wemmie, **V.C. Sheffield**. Novel BBsome independent phenotypes in a mouse model of Bardet-Biedl syndrome 3. ASHG 2394, 2009
219. Pretorius, P.R., L.M. Baye, R.F. Mullins, C.C. Searby, D.Y. Nishimura, K. Bugge, B. Yang, E.M. Stone, D.C. Slusarski, **V.C. Sheffield**. A newly identified Bardet-Biedl syndrome 3 (ARL6) long isolform has a vision specific function. ASHG 2436, 2009.

220. Scheetz, T.E., K. Cribben, T.L. Casavant, T.A. Braun, J.H. Fingert, E.M. Stone, **V.C. Sheffield**. A computational system to facilitate efficient discovery of disease genes. ASHG 2697, 2009.
221. Thompson S, Nylen EL, East JS, Kardon RH, Pinto LH, **Sheffield VC**, Stasheff SF, Mullins RF, Stone EM. Different bipolar cell input pathways for negative masking and the pupil light reflex in mice. Pgrm Nr: 669/D733 Association for Research in Vision and Ophthalmology Annual Meeting (May 2-6, 2010 in Fort Lauderdale, FL)
222. Fingert JH, Robin AL, Stone JL, Scheetz TE, Casavant TL, Wassink TH, Alward WLM, **Sheffield VC**, Stone EM. Identification of a novel glaucoma locus. Pgrm Nr: 2159/A258 Association for Research in Vision and Ophthalmology Annual Meeting (May 2-6, 2010 in Fort Lauderdale, FL)
223. Vogel T, Zhang Q, Moninger T, Nishimura D, Searby C, Beck G, Bugge K, Thedens D, Swiderski R, Howard M, **Sheffield V**. Capase mediated apoptosis in neural progenitor cells in mouse models of Bardet-Biedl syndrome reveal novel mechanisms and treatment strategies for neonatal hydrocephalus. ASHG, 19, 2010
224. Muhammad E, Levitas A, Chalifa Caspi V, Manor E, Beck JC, **Sheffield VC**, Parvari R. Identification of a locus for recessive dilated-cardiomyopathy on chromosome 2 by linkage analysis in a Bedouin family. ASHG 679, 2010
225. Nimmakayalu M, Major H, Qian Q, Shchelochkov OA, Darbro B, Van Rheeden R, Hulseberg D, **Sheffield VC**, Nagy PL, Patil SR. Utilization of high density oligonucleotide array to analyze patients with developmental delay and congenital anomalies. ASHG 1273, 2010
226. Parvari R, Muhammad E, Levnthal N, Chalifa-Caspi V, Beck JC, **Sheffield VC**, Hershkovitz E. Identification of a chromosomal locus associated with recessive dysfunction of sodium homeostasis in a Bedouin family. ASHG 2185, 2010
227. Fingert JH, Stone JL, Robin AL, ROos B, Davis LK, Scheetz TE, Alward WLM, Kwon YH, Wassink TH, **Sheffield VC**, Stone EM. Identification of a novel duplication of chromosome 12q14 and the TBK1 gene in normal tension glaucoma patients. ASHG, 2182, 2010
228. Meyer KJ, Nishimura DY, Beck JS, Davis LK, Stone EM, Wassink TH, **Sheffield VC**. Copy number variations unmask recessive mutations and identify novel candidates for Bardet-Biedl syndrome. ASHG, 1800, 2010
229. Seo S, Bugge K, Searby CC, Nachury MV, **Sheffield VC**. A novel protein Lxtfl1 regulates ciliary trafficking of the BBSome. ASHG, 167, 2010
230. Baye LM, Patrinostro X, Beck JS, Swaminathan S, Zhang Y, Stone E, **Sheffield VC**, Slusarski DC. Structural-functional analysis of cep290 in zebrafish vision. ASHG, 1613, 2010
231. Yang Y, Mendiratta MS, Balazs A, Willis A, Potocki L, **Sheffield VC**, Karaviti LP, Eng CM. *POMC* gene mutation analysis and genotype-phenotype correlations in two patients with early-onset obesity. ASHG, 868, 2010
232. Pretorius PR, Aldahmesh MA, Zhang Q, Searby CC, Nishimura DY, Alkuraya FS, Stone EM, Rahmouni K, Slusarski DC, **Sheffield VC**. Functional analysis of BBS3 and BBS3L, a novel BBS3 transcript, in syndromic and non-syndromic retinal degeneration in animal models and humans. ASHG, 1590, 2010
233. Mazor M, Alkrinawi S, Caspi V, Manor E, Beck JC, **Sheffield V**, Aviram M, Parvari R. Identification of a chromosomal locus associated with recessive primary ciliary dyskinesia in a Bedouin family. ASHG, 2268, 2010
234. Zode GS, Bugge K, Searby CC, Stone EM, Kuehn MH, **Sheffield VC**. Development of a mouse model of primary open angle glaucoma reveals a disease mechanism. ASHG, 2606, 2010
235. Chamling X, Seo S, **Sheffield VC**. A novel BBSome interacting protein is an ADP/ATP translocase, SLC25A31. ASHG, 2011
236. Tucker BA, Scheetz TE, Mullins RF, DeLuca AP, Hoffmann JM, Johnston RM, Jacobsen SG, **Sheffield VC**, Stone EM. Exome sequencing and analysis of induced pluripotent stem cells identify the cilia-related gene MAK as a cause of retinitis pigmentosa. ASHG 2011
237. Muhammad E, Levitas A, **Sheffield VC**, Parvari R. Identification of a new chromosomal locus for mutation causing left ventricular non-compaction with ventricular tachycardia cardio-pathology. ASHG 2011
238. Zhang Y, Zhang Q, Searby C, Seo S, **Sheffield VC**. Cep290 function in localization of cilia proteins resulting in phenotypic heterogeneity of CEP290-associated disease. ASHG 2011

239. Zode G, Kuehn M, Searby C, Mohan K, Grozdanic S, Nishimura D, Bugge K, Anderson M, Clark A, Stone E, **Sheffield V**. Development and pharmacological rescue of a murine model of primary open angle glaucoma. ASHG 2011
240. Gottlieb RL, Nishimura D, Searby C, Bugge K, **Sheffield VC**. Strain-specific differences in perinatal viability support genetic modifiers in Bardet-Biedl syndrome. ASHG 2011
241. Scheetz TE, DeLuca AP, Braun TA, Streb LM, Affatigato LM, **Sheffield VC**, Stone EM. The importance of variation databases in interpretation of exome sequencing. ASHG 2011
242. Parvari R, Mazor M, Alkrinawi S, Shlifa-Caspi V, Manor E, **Sheffield VC**, Aviram M. Primary ciliary dyskinesia caused by homozygous mutation in DNAL1 encoding dynein light chain 1. ASHG 2011.
243. Mahajan VB, Folk JC, Fingert JH, Skeie JM, Kinnick TR, Scheetz TC, Bassuk AG, Manak JR, **Sheffield VC**, Stone EM. Genetic analysis and phenotypic staging of autosomal dominant neovascular inflammatory vitreoretinopathy. Pgrm Nr: 62/A175 Association for Research in Vision and Ophthalmology Annual Meeting (May 1-5, 2011 in Fort Lauderdale, FL)
244. Zhang Y, Baye LM, Zhang Q, Beck J, Patrinostro X, Swaminathan S, Slusarski DC, **Sheffield V**, Stone EM. A CEP290 N-terminal protein fragment rescues a zebrafish model of Cep290 Leber's congenital amaurosis. Pgrm Nr: 1400/A549 Association for Research in Vision and Ophthalmology Annual Meeting (May 1-5, 2011 in Fort Lauderdale, FL)
245. Stone EM, DeLuca A, Scheetz TE, Braun TA, Affatigato LM, Daggett HT, Johnston RM, Streb MR, **Sheffield VC**. Analysis of 200 human exomes for improved mutation detection specificity. Pgrm Nr: 3314/A562 Association for Research in Vision and Ophthalmology Annual Meeting (May 1-5, 2011 in Fort Lauderdale, FL)
246. Zode GS, Nishimura DY, Ding Q, Searby CC, Mohan K, Grozdanic SD, Stone EM, **Sheffield VC**, Kuehn MH. A chemical chaperone rescues glaucoma by reducing ER stress in a novel murine model of primary open angle glaucoma. Pgrm Nr: 5917 Association for Research in Vision and Ophthalmology Annual Meeting (May 1-5, 2011 in Fort Lauderdale, FL)
247. Weihbrecht K, Humbert M, **Sheffield V**, Seo S. NPHP10 (SDCCAG8) interacts with components of the multi-aminoacyl-tRNA synthetase complex. ASHG 2012
248. Zode GS, Bugge KE, Stone EM, **Sheffield VC**. Topical ocular sodium 4-phenylbutyrate rescues glaucoma in a mouse model of primary open angle glaucoma. ASHG 2012
249. Muhammad E, Levitas A, **Sheffield V**, Parvari R. Identification of a new chromosomal locus for a mutation causing left ventricular non-compaction with ventricular tachycardia cardio-pathology. ASHG 2012
250. Seo S, Humbert MC, Weihbrecht K, Searby CC, Li Y, Pope RM, **Sheffield VC**. ARL13B, INPP5E, PDE6D and CEP164 form a functional network involved in Joubert syndrome and Nephronophthisis. ASHG 2012.
251. Zhang Y, Bugge K, Searby CC, Mullins RF, Seo S, **Sheffield VC**. Physical and genetic interactions between BBS genes and CEP290. ASHG 2012
252. Parvari R, Mazor M, Alkrinawi S, **Sheffield VC**, Aviram M. A novel chromosomal locus in a family with primary ciliary dyskinesia. ASHG 2012
253. Chamling X, Seo S, Bugge K, Searby CC, Drack AV, Rahmouni K, **Sheffield VC**. Transgenic rescue of BBS phenotypes in Bbs4 null mice. ASHG 2012
254. Mahajan VB, Skeie JM, Bassuk AG, Fingert JH, Braun TA, Daggett HT, Folk JC, **Sheffield VC.** Calpain-5 causes autoimmune uveitis, retinal neovascularization and photoreceptor degeneration. ASHG 2012
255. Zode GS, Bugge KE, Grozdanic SD, Kardon RH, Anderson MG, Stone EM, Sheffield VC. Topical ocular sodium 4-phenybutyrate rescues glaucoma in a myocilin mouse model of primary open angle glaucoma. Pgrm Nr: 4678 Association for Research in Vision and Ophthalmology Annual Meeting (May 6-9, 2012 in Fort Lauderdale, FL)
256. Drack AV, Bhattarai S, Seo S, Gratie D, Stone EM, Mullins R, Sheffield V. Subretinal gene therapy in Bbs1mice. Pgrm Nr: 6566/A568 Association for Research in Vision and Ophthalmology Annual Meeting (May 6-9, 2012 in Fort Lauderdale, FL)
257. Lin X, Zode G, Searby CC, **Sheffield VC**. Defects in the autophagy pathway contribute to glaucoma caused by mutant myocilin accumulation. PgmNr 2009W: Presented at The 63th Annual Meeting of The American Society of Human Genetics, (October 22-26, 2013 in Boston, MA).

258. Weihbrecht K, Humbert M, **Sheffield V**, Seo S. NPHP10 (SDCCAG8) interacts with components of the multi-aminoacyl-tRNA syntehtase complex. PgmNr 2915W: Presented at The 63th Annual Meeting of The American Society of Human Genetics, (October 22-26, 2013 in Boston, MA).
259. Zode G, Sharma A, Lin X, Searby C, Bugge K, Clark A, **Sheffield V**. Genetic and pharmacological reduction of ER stress rescues glaucoma in a murine model of glucocorticoid induced glaucoma. PgmNr 2281T: Presented at The 63th Annual Meeting of The American Society of Human Genetics, (October 22-26, 2013 in Boston, MA).
252. Chamling X, Seo S, Searby CC, Kim GH, Slusarski DC, **Sheffield VC**. The centriolar satellite protein AZ11 interacts with BBS4 and is involved in ciliary trafficking of the BBSome. PgmNr 2914F: Presented at The 63th Annual Meeting of The American Society of Human Genetics, (October 22-26, 2013 in Boston, MA).
253. Muhammad E, Reish, O, Ohno Y, Scheetz T, DeLuca A, Searby C, Regev M, Benyamini L, Fellig Y, Kihara A, **Sheffield VC**, Parvari R. Identification of a novel locus for a recessive congenital myopathy by linkage analysis in an Israeli Bedouin family. PgmNr 2944F: Presented at The 63th Annual Meeting of The American Society of Human Genetics, (October 22-26, 2013 in Boston, MA).
254. Nuangchamnong N, Carter CS, Zhang Q, Vogel T, **Sheffield VC**. A primary ciliopathy protein plans an extra-ciliary role in neurodevelopmental disease. PgmNr 3130S: Presented at The 64th Annual Meeting of The American Society of Human Genetics, (October 18-22, 2014 in San Diego, CA).
255. Weihbrecht K, **Sheffield V**, Seo S. Characterizing the Nphp10 (Sdccag8Tn(sb-Tyr)2161B.CA1Cove) mouse model. PgmNr 921T: Presented at The 64th Annual Meeting of The American Society of Human Genetics, (October 18-22, 2014 in San Diego, CA).
256. Carter C, Drack A, Zhang Q, Nuangchamnong N, Searby C, **Sheffield VC**. Primary cilia mediate retinal development and photoreceptor homeostasis. PgmNr 389: Presented at The 64th Annual Meeting of The American Society of Human Genetics, (October 18-22, 2014 in San Diego, CA).
257. Hsu Y, Garrison JE, Nishimura DY, Bugge KE, Searby CC, **Sheffield VC**. The role of the BBSome in visual fuction during eye development and adulthood. PgmNr 2544:Presented at The 65th Annual Meeting of The American Society of Human Genetics, (October 6-10, 2015 in Baltimore, MD).
258. Weihbrecht K, Goar WA, Searby CC, Scheetz TE, **Sheffield VC**, Seo S. A mouse modifier study using the Nphp10 (Sdccag8 Tn(sb-Tyr)2161B.Ca1Cove) model to identify a modifier locus of ciliopathy-related phenotyeps. PgmNr 2801: Presented at The 65th Annual Meeting of The American Society of Human Genetics, (October 6-10, 2015 in Baltimore, MD).
259. Garrison JE, Hsu Y, Nishimura DY, Bugge KE, Searby CC, **Sheffield VC**. Postnatal excision of a Bardet-Biedl syndrome gene results in leptin resistance, leading to obesity. PgmNr 2818: Presented at The 65th Annual Meeting of The American Society of Human Genetics, (October 6-10, 2015 in Baltimore, MD).
260. Cring M, Garrison JE, **Sheffield VC**. CRISPR-Cas9 as a potential therapeutic tool for the Bardet-Biedl syndrome (BBS) M290R mutation in vitro and in vivo. PgmNr 488: Presented at The 65th Annual Meeting of The American Society of Human Genetics, (October 6-10, 2015 in Baltimore, MD).
261. Jain A, Zode G, Bugge K, Seary C, Zhang F, Clark A, **Sheffield V**. CRISPR-Cas9 Mediated genome editing of myocilin in hereditary glaucoma. PgmNr 494: Presented at The 65th Annual Meeting of The American Society of Human Genetics, (October 6-10, 2015 in Baltimore, MD).
262. Cring M, Searby C, Carter C, **Sheffield V**. CRISPR/Cas9 as a therapeutic tool for the germline correction of the BBS1 M390R mutation. (Abstract/Program # 263F) Presented at the 66th Annual Meeting of The American Society of Human Genetics, October 19, 2016, Vancouver, Canada.
263. Weihbrecht K, Searby C, Goar W, **Sheffield VC**, Seo S. SDCCAG8 affects AIMP2 localization and alters its downstream target, p53. (Abstract/Program # 2595F) Presented at the 66th Annual Meeting of The American Society of Human Genetics, October 19, 2016, Vancouver, Canada
264. DeLuca AP, Hiatt S, Bowling K, Andorf JL, Gray D, Finnila C, Thompson M, Drack AV, Cooper G, Scheetz TE, **Sheffield VC**, Stone EM. Phenotype-driven analysis of whole genome sequencing to find missing alleles in retinal disease. (Abstract/Program # 915F) Presented at the 66th Annual Meeting of The American Society of Human Genetics, October 19, 2016, Vancouver, Canada.
265. Goar WA, Majdalani P, Searby CC, Whitmore SS, DeLuca AP, Imtirat A, Stone EM, Pavari R, Scheetz TE, **Sheffield VC**. (Abstract/Program # 1710F) Presented at the 66th Annual Meeting of The American Society of Human Genetics, October 19, 2016, Vancouver, Canada.

266. Singh M, **Sheffield V**. Global loss of BBS proteins is associated with increased levels of reactive astrocytes and pro-inflammatory cytokines, IL1b, IL6, IL15 and TNFa in brains of moues models of Bardet Biedl Syndrome. (Abstract/Program # 2246T) Presented at the 66th Annual Meeting of The American Society of Human Genetics, October 19, 2016, Vancouver, Canada)
267. Hsu Y, Garrison J, Kim G, Nishimura D, Searby C, Schmitz A, Datta P, Seo S, **Sheffield V**. (Abstract/Program # 106) Presented at the 66th Annual Meeting of The American Society of Human Genetics, October 19, 2016, Vancouver, Canada)
268. Cring M, Drack A, Sheffield V. Genetic therapeutic strategies for Bardet-Biedl syndrome type 1 (Abstract/Program # 1087) Presented at the 67h Annual Meeting of the American Society of Huam Genetics, October 17, 2017, Orlando, Florida)
269. Aspit L, Reish O, Benyamin L, Sheffield V, Parvari R. A novel homozygous mutation in MMP21 is associated with heterotaqxia and cardiac defects. (Abstract/Program # 1147) Presented at the 67h Annual Meeting of the American Society of Huam Genetics, October 17, 2017, Orlando, Florida)
270. Pak T, Carter C, Gupta S, Wemmie J, Huang S, Nuangchamnong N, Searby C, Abbott P, Stevens H, Sheffield V. BBS1 M390R/M390R mice have impaired anxiety-like behavior. (Abstract/Program # 2137) Presented at the 67h Annual Meeting of the American Society of Huam Genetics, October 17, 2017, Orlando, Florida)

B. Areas of Research Interest and Current Projects

Molecular genetics of glaucoma
Molecular biology of hereditary macular degeneration
Molecular genetics of hypertension and obesity
Molecular biology of syndromic retinitis pigmentosa
Molecular biology of hydrocephalus
Treatment of human genetic diseases using iPS cells and genome editing

C. Previous Support

Title	Project Period	Direct Costs
NIH P30 HD27748 Child Health Research Center: Identification of Polymorphisms within Developmental Genes Principal Investigator	10/1/90 - 6/30/91	$30,000
Retinitis Pigmentosa and George Gund Foundation: Clinical Features and Molecular Biology of Dominant and Recessive Retinitis Pigmentosa. Principal Investigator	7/1/91 - 6/30/94	$250,000
NIH RO1 HG00457 Identification of Polymorphisms in Sequenced Tagged Sites Principal Investigator	9/30/91 - 08/31/94	$772,680
NIH P50 HG00835 Cooperative Human Linkage Center. Principal Investigator Project 2 Principal Investigator Molecular Analysis Core	9/25/92 - 8/31/96	$1,194,014 $1,853,548

NIH PO1 CA40737 Molecular Epidemiology of Primary Hepatocellular Carcinoma Co-Investigator	2/1/93 - 11/31/98	$939,670
Carver Charitable Trust An International Center for Ophthalmic Molecular Diagnosis and Technology Development Co-Principal Investigator	10/1/93 - 9/30/96	$450,000
Grousbeck Foundation Molecular Genetics of Leber's Congenital Amaurosis and Allied Conditions Co-Principal Investigator	1/1/94 - 12/31/94	$25,000
NIH RO1 EY10539 Molecular Genetics of Macular Degeneration Co-Principal Investigator	6/1/94 - 5/31/99	$1,065,849
NIH RO1 HG00457 Improved Mutation Detection and High Throughput Genotyping Principal Investigator	7/1/94 - 6/30/97	$897,657
Retinitis Pigmentosa Foundation Molecular Biology of Leber's Congenital Amaurosis and Autosomal Dominant Retinitis Pigmentosa Co-Principal Investigator	7/1/94 - 6/30/97	$300,000
NIH RO1 EY10564 The Molecular Genetics of Hereditary Glaucoma Principal Investigator	9/30/94 - 9/29/99	$1,212,906
NIH EY-11298 Molecular Biology of Syndromic Retinal Degeneration Principal Investigator	12/1/95-11/30/99	$705,263
NIH NHLBI RO1 HL-59789 A Program for Rat Gene Discovery and Mapping Principal Investigator	9/30/97 – 9/30/99	$2,529,201
NIH RO1 EY10564 The Molecular Genetics of Hereditary Glaucoma Principal Investigator	12/1/99 – 11/30/05	$1,681,115
NIH NHLBI RO1 HL-59789 A Program for Rat Gene Discovery and Mapping Principal Investigator	9/30/99 – 8/31/03	$6,754,254
NHLBI P50 HL-62178 SCOR in Pediatric Cardiovascular Disease Molecular Genetic Epidemiology of Three Cardiac Defects Co-Principal Investigator (PI: Ronald Lauer)	1/1/99 – 12/31/03	$4,759,574

NIH NIMH R10 MH-55284 A Collaborative Linkage Study of Autism Co-Investigator (PI: Joseph Piven)	7/15/99 – 6/30/03	$835,196
NIH/NHLBI P50-HL 55006 SCOR Molecular Genetics in Hypertension PI Genotyping Core (PI Curt Sigmund)	2/1/01-1/31/06	$1,129,260
NIH EY-016822 Fibulin-Associated Age-Related Macular Degeneration (PI Edwin Stone; Co-PI Val Sheffield)	9/15/05-5/31/10	$1,607,000
NIH MH-9907 Mouse Mutagenesis: Phentype-driven neuroscience screen Co-Investigator (subcontract with Northwestern University)	1/1/01-3/31/06	$810,839
NIH RO1 EY10564 The Molecular Genetics of Hereditary Glaucoma Principal Investigator	4/1/06 – 3/30/11	$1,741,646
NIH EY-11298 Molecular Biology of Syndromic Retinal Degeneration Principal Investigator	7/1/02-6/30/07	$1,799,831
NIH EY-11298 Evaluation of Complex Inheritance of Retinal Degeneration in Bardet-Biedl Syndrome Principal Investigator	8/1/07-7/31/12	$1,990,372
NIH EY-017168 Interdisciplinary Approach to Retinal Disease Gene Identification Principal Investigator	6/1/07-5/31/12	$1,874,061
Howard Hughes Medical Institute	4/1/97-8/31/15	
Virginia and L.E. Simmons Excellence Fund	6/24/13-5/31/16	
Binational Science Foundation Genes in left-right body asymmetry Co-Principal Investigator	10/1/12-9/30/16	$180,000
NIH EY-022616-01 Molecular Pathophysiology of Retinal Degeneration in Bardet-Biedl Syndrome Co-Investigator	9/1/12-8/31/16	$335,404
NIH Unfolded Protein Response in Glaucoma Pathogenesis Co-Investigator	9/1/13-8/31/16	$250,000

NIH/NHLBI HL-084207 Genetic & Signaling Mechanism in the Central Regulation of Blood Pressure Project 3 The Role of Brain Bardet-Biedl Syndrome Genes in Metabolic and Cardiovascular Regulation Co-Investigator	4/1/13-3/31/18	$322,230
NIH EY011298-15 Treatment of a Complex Retinal Degeneration Syndrome Principal Investigator	4/1/13-3/31/18	$289,568

D. Current Grant Support

Title	Project Period	Direct Costs
Carver Chair Molecular Medicine	8/1/14-6/31/18	$ 200,000
NIH EY024259 Novel Glaucoma Treatment Using Genome Editing Principal Investigator	4/1/14-3/31/19	$411,770
NIH NS-083543-01 The Role of Neural Progenitor Cells in the Development of Neonatal Hydrocephalus Principal Investigator	7/1/13-6/31/18	$218,750
NIH P30 EY025580 (PI: Val Sheffield) Multidisciplinary Investigations in Visual Science Principal Investigator	9/01/16-8/31/21	$351,157
NIH 2RO1-DE016886 (Paul Romitti, PI) Nonsyndromic Craniosynostosis: Phenotype/Genotype Study Principal Investigator	9/1/14-6/30/19	$ 73,227

E. Invited lectures, Conference presentations, visiting professorships

1990
ASHG Annual Meeting
Cincinnati, OH
"Identification of a germline mutation in the p53 gene by using GC-clamped denaturing gradient gel electrophoresis"

1991
p53 Workshop
Princeton University
Princeton, NJ

1992
ASHG Annual Meeting
San Francisco, CA
"The gene for autosomal dominant neovascular inflammatory vitreoretinopathy maps to 11q13"

1993
Third International Chromosome 11 Workshop
La Jolla, CA
"Genetic linkage of two forms of hereditary blindness to markers on chromosome 11"

Gordon Research Conferences
Salve-Regina University Rhode Island
“Mutations in the RDS/Peripherin gene cause widely different phenotypes between and within affected families”

ASHG Annual Meeting
New Orleans, LA

1994
ASHG Annual Meeting
Montreal, Quebec, Canada
“Evaluation of an efficient approach for identifying genetic disease loci”

World Health Organization
Rio de Janerio, Brazil

Israel
“Introduction to molecular biology for ophthalmologists”

Institute of Human Genetics
University of Minnesota
Minneapolis, MN

1996
Genetics and Glaucoma II (POAG)
Consortium Meeting
Lake Bluff, IL

National Institute of Child Health and Human Development
Rockville MD
“Novel methods of linkage analysis”

Weinstein Congenital Heart Disease Conference
Philadelphia, PA

Cold Spring Harbor,
Cold Spring Harbor, NY
“Use of isolated populations, a DNA pooling strategy, and linkage disequilibrium to map genetic disease loci”

ARVO
Fort Lauderdale, FL
“High density mapping of the juvenile open angle glaucoma region”

XIth Symposium of the International Society for Genetic Eye Disease and the VIIIth Symposium of the Retinoblastoma Society
Hobart, Tasmania, Australia
“Efficient identification of genetic disease loci”

1997
Ben Gurion University of the Negev
Division of Pediatrics and Ophthalmology Department
Beer-Sheva, Israel
“The use of genomic resources to identify a gene causing primary open angle glaucoma”

Hadassah Medical Center, Hebrew University
Department of Human Genetics
Jerusalem, Israel
"The molecular genetics of primary open angle glaucoma"

University of Tel-Aviv
Department of Human Genetics
Tel-Aviv, Israel
"The use of population isolates for the study of human genetic disease"

Rambam Medical Center
Genetics Department
Haifa, Israel
"The Human Genome Project and the search for disease genes"

American Heart Association Meeting
Orlando, Florida
"Screening for genetic markers of cardiovascular disease: When? How? Why?"

ASHG Annual Meeting
Baltimore, Maryland
"The molecular genetics of glaucoma"

20th Biennial Cornea Research Conference
Boston, Massachusetts
"The molecular genetics of hereditary blindness"

Third Great Basin Visual Science Symposium and
Western Eye Research Conference
Salt Lake City, Utah
"The molecular genetics of glaucoma"

ARVO
Fort Lauderdale, Florida
"The molecular genetics of glaucoma"

American Heart Association
San Diego, California
"Genetic approaches to the study of congenital heart disease"

1998

Mt. Sinai Hospital
Toronto Western Eye Research
Toronto, Ontario, Canada
"A comprehensive approach to identifying human hereditary disease genes using human populations"

Harvard Medical School
Department of Genetics
Boston, Massachusetts
"The search for genetic disease causing genes using human populations"

Twentieth Wellcome Trust Summer School Human Genome Analysis: From Genome to Function
London, England
"Strategies for identification of genes causing human hereditary diseases"

Marine Biological Laboratory
Wood Hole, Massachusetts
"The molecular biology of hereditary blindness"

Ohio State University
Cancer Education and Development
Comprehensive Cancer Center Grand Rounds
Columbus, Ohio
"Use of inbred populations in the identification of disease genes"

1999
Baylor College of Medicine
Department of Molecular and Human Genetics Seminar
Houston, Texas
"Genetic approaches to identifying genes causing human genetic eye diseases"

University of Tennessee
Department of Ophthamology
Memphis, Tennessee
"Genetic approaches for identifying genes causing hereditary blindness"

HHMI Genes & Genomics scientific meeting
Chevy Chase, Maryland
"The search for genes causing hereditary blindness and deafness using isolated human populations"

University of Michigan
Department of Pediatrics and Communicable Diseases/Pediatric Research Symposium
Ann Arbor, Michigan
"Molecular Genetic Studies of Hereditary Blindness and Deafness"

Cold Spring Harbor Laboratory
Physiological Genomics and Rat Models Conference
Cold Spring Harbor, New York
"Application of Serial Subtraction of Normalized cDNA Libraries to a Large Scale Rat Gene Discovery and Mapping Project"

2000
Keystone Symposium
Molecular and Cell Biology
Snowbird, Utah
"The Search for Genes Involved in Congenital Heart Defects"

University of Washington
Seattle, Washington
"Molecular Biology of Hereditary Deafness and Blindness" and "Gene Discovery and Mapping in the Rat"

HHMI Scientific Meeting
Chevy Chase, Maryland
"Use of Isolated Population to Identify Genes Causing Human Hereditary Blindness"

Kyoto University
Japan
Kyoto, Japan
"Molecular Biology of Glaucoma"

Great Thinkers Lecture
Utah Valley State College
Provo, Utah
"The Human Genome Project"

2001
Utah Valley State College
Provo, Utah
"Identification of Human Disease Genes using Isolated Populations in Israel"

Duke University Medical Center
Department of Genetics
Durham, North Carolina
"The Molecular Genetics of Hereditary Blindness"

Brigham Young University
Department of Zoology
"The Human Genome"

HHMI Scientific Meeting
Mammalian Genetics and Physiological Genomics
Chevy Chase, Maryland
"The Molecular Genetics of a Human Obesity Syndrome (BBS)"

Creighton University School of Medicine
The Great Plains States Society for Molecular Biology and Genetics Program
Omaha, Nebraska
"The Search for Human Disease Genes Using Isolated Inbred Populations"

The Neurogenetic Institute
Keck School of Medicine
The University of Southern California
"The Molecular Genetics of a Human Obesity Syndrome"

HHMI-NIH Research Scholars Program
Bethesda, Maryland
"The Molecular Genetics of Bardet-Biedl syndrome"

2002
University of California
Department of Neurosciences
San Diego, California
"The Molecular Genetics of Bardet-Biedl syndrome"

McDermott Center, Excellence in Human Genetics
University of Texas, Southwestern
Dallas, Texas
"Rare Insights into Common Disease: The Molecular Genetics of a Human Obesity Syndrome"

2003
University of Michigan
Training Program in Genomic Science
Ann Arbor, Michigan
"The Molecular Genetics of a Human Obesity Syndrome"

University of Iowa
Department of Medicine
Grand Rounds
Iowa City, Iowa
"The Role and Relevance of Genetics in Genomics in Medical Practice"

Howard Hughed Medical Institute
Science Meeting
Bethesda, Maryland
"Individuals and Populations: The Search for an Elusive Human Obesity Syndrome Gene"

NHGRI Symposium
From Double Helix to Human Sequence-and Beyond!
Bethesda, Maryland
"Impact of the Human Genome Project on Hereditary Blindness, Simple and Complex"

Pediatrics Academic Society
Annual Meeting
Seattle, Washington
"Disease Gene Identification Utilizing Isolated Human Populations"

Human Medical Genetics Seminar Series
University of Colorado
Denver, Colorado
"Rare Insights into Common Disease: The Molecular Genetics of a Human Obesity Syndrome"

HHMI/CSIS
Capitol Hill
Washington, DC
"The Study of Human Genetic Diseases Utilizing Human Subjects"

American Diabetes Association
Case Study Session
New Orleans, Louisiana
"The Molecular Genetics of Bardet-Biedl Syndrome"

Symposium for Rising Stars
University of Chicago
Chicago Illinois
"Genetic Diseases among the Bedouin: A Model for the Study and Management of Human Genetic Disease"

2004
Genetics Seminar
Huntsville Alabama
"The Study of a Complex Human Obesity Syndrome: From Human Populations to Animal Models"

Lovett/Genetics Seminar
Washington University
St Louis Missouri
"Study of a Complex Human Obesity Syndrome: From Isolated Populations to Animal Models"

Biotechnology Conference
Ben Gurion University of the Negev
Beer Sheva Israel
"From Human Populations to Animal Models: The BBS Story"

HHMI Science Meeting
Chevy Chase Maryland
"Molecular Genetics of Bardet-Biedl Syndrome"

Nebraska Medical Center
Omaha Nebraska
"Molecular Genetics of a Complex Human Obesity Syndrome: Bardet-Biedl Syndrome"

2005
HHMI Medical Fellows Meeting
Keynote Speaker
Chevy Chase Maryland

2006
Biochemistry Department, University of Iowa
Iowa City Iowa

Genentix/San Francisco California
"The Molecular Genetics of a Complex Human Obesity Syndrome"

Ponce School of Medicine
Cagua Puerto Rico
"The Molecular Genetics of a Syndromic Form of Retinitis Pigmentosa"

HHMI Scientific Meeting
Chevy Chase MD

American Society of Human Genetics
New Orleans Louisiana
"Demonstration of Genetic Interactions betweenTRIM32 and other Bardet-Biedl Syndrome Genes"

2007
GeNeSIS Investigators Meeting
Paris, France, 2007
Molecular Genetics of Bardet-Biedl Syndrome"

LDSLSRS Conference
Snowbird Mountain Resort
Snowbird, Utah 2007

2008
HHMI Scientific Meeting
Chevy Chase, MD
"The Molecular Pathophysiology of Bardet-Biedl Syndrome"

Mouse Genetics & Genomics: Development & Disease
Cold Spring Harbor, NY
"Mouse Models of the Human Obesity Disorder, Bardet-Biedl Syndrome"

Symposium on the Human Obesity Syndrome, Bardet-Biedl Syndrome, and Cilia
University of Iowa, Iowa City, Iowa
"Bardet-Biedl Syndrome Historical Perspectives"

2009
Clinical Nutrition Research Center
University of Alabama, Birmingham, Alabama
"Molecular Physiology of a Human Obesity Syndrome"

HHMI Scientific Meeting
Chevy Chase, MD
"Molecular Complexes and the Pathophysiology of Bardet-Biedl Syndrome"

Brigham Young University
Salt Lake City, UT
"The Molecular Genetics of a Complex Human Mendelian Disease: The Blind Leading the Obese"

UCSF Seminars in Biomedical Sciences
University of California San Francisco, San Francisco, CA
"The molecular genetics of a human obesity syndrome"

Genetics Seminar Series
Emory University School of Medicine, Atlanta GA
"The Molecular Genetics of a Human Obesity Syndrome"

NEI 40th Anniversary Symposia Series on Genetics and Genomics in Vision
NIH Bethesday Campus
"The Blind leading the Obese: The Molecular Pathophysiology of Bardet-Biedl Syndrome"

CVG Symposium Seminar
Cornell University, Ithaca NY
"The Molecular Genetics of a Human Obesity Syndrome"

College of Veterinary Medicine Seminar Series
Iowa State University, Ames, IA
"Animal Models and Bardet-Biedl Syndrome"

Symposium on The Human Model: Genetics as Two-Way Information
The 49th Annual Meeting of the American Society for Cell Biology
"Animal Models, Molecular Complexes: The Pathogenesis of a Human Obesity Syndrome"

2010
Lois and David Rich Lecture Series in Visual Science
University of Alabama at Birmingham, Birmingham AL
"Genetics Testing for Ophthalmologic Diseases"-Resident Lecture
"Molecular Genetics of a Human Retinopathy Syndrome, Bardet-Biedl Syndrome"-Scientific Lecture

Cilia, Signaling and Human Disease
Keystone Symposium on Molecular and Cellular Biology
Monteray, California

International Congress of Endocrinology
Kyoto, Japan

HHMI Scientific Meeting
Chevy Chase, MD
"The Molecular Pathophysiology of Bardet-Biedl Syndrome"

2011
Florida State University
The Inaugural 2011 FSU Life Sciences Symposium/From Molecules to Medicine
"Human Genetics of Bardet-Biedl Syndrome (BBS)"
Tallahassee, Florida

The Hospital for Sick Children
Program in Genetics and Genome Biology's Molecular Medicine Distinguished Lecturer Series
"Molecular Mechanisms of Bardet-Biedl Syndrome, a Human Obesity Syndrome"
Toronto, Canada

Human Genetics Center Seminar in Genetics and Population Biology
School of Public Health, The University of Texas
"Molecular Mechanisms of Bardet-Biedl Syndrome, a Human Obesity Syndrome"
Houston, Texas

Society for Endocrinology
"Primary Cilia Dysfunction Causes Impaired Leptin Signaling and Obesity
Birmingham, England

2012
University of Colorado, Denver
Human Medical Genetics Program
"The Blind Leading the Obese: The Molecular Pathophysiology of a Human Obesity Syndrome"

HHMI Scientific Meeting
Chevy Chase, MD

Hyperphagia Conference
Baton Rouge, LA
"Animal Models of Bardet-Biedl Syndrome and Hyperphagia"

2013

HHMI Renewal
Chevy Chase, MD

American Society of Nephrology
Atlanta GA

2014

University of Iowa
Anatomy and Cell Biology
"Diabetes and Obesity"

University of Iowa
Department of Pediatrics
"Novel Treatments of Common Disease"

Beer Sheva, Israel
"Progress Towards Treatment of Common Genetics Disorders"

Beer Sheva. Israel
"Molecular Basis of BBS, a Human Obesity Disorder"

HHMI Scientific Meeting
Chevy Chase, MD

2015

Univerity of North Texas Health Science Center
North Texas Eye Research Center
"Studies of the pathophysiology and intervention in a human syndromic blinding disorder"

Stony Brook School of Medicine
Vanderbilt University
Stony Brook, NY
"The Complex Pathophysiology of a Human Obesity Syndrome"

Sanford Childrens Health Research Center
University of South Dakota
Annual Center Symposium
"Molecular pathophysiology of a human obesity syndrome: Relationship to hydrocephalus"

Growth, Development, and Disabilities Training Program (GDDTP)
University of Chicago,
Chicago, IL
"Molecular pathophysiology and progress towards treatment of a complex human obesity syndrome"

2016

Association for Research in Vision and Ophthalmology
Seattle, Washington
"CRISPR-based treatment of MYOC-associated glaucoma"

The Jackson Laboratory for Genomic Medicine
Farmington, CT
"Molecular pathophysiology and progress towards treatment of a complex human obesity syndrome"

American Society of Human Genetics
Vancouver, Canada
"Functional validation and genetic intervention for retinal disease genes."

2017
Biology and Therapy of the Ciliated Senses
Gainesville, Florida
"Comprehensive Approaches to the Treatment of Cilia-Related Retinal Disease"

2018
Ground Round-Internal Medicine (Faculty and Residents)
University of Illinois, Colege of Medicine
Peoria, Illinois
"The Blind Leading the Obese: The Pathophysiology of a Human Obesity Syndrome"

Research Day (Students, Residents, Faculty)
University of Illinois, Colege of Medicine
Peoria, Illinois
"Molecular Pathophysiology and Progress Towards Treatment of Genetic Blinding Disorders"

Living Healthy Series (Open to Public)
University of Illinois, Colege of Medicine
Peoria, Illinois
"Improving Personal Health using Genome Information—Fact and Future"

IV. SERVICE

A. Offices held in professional organizations

1999-2002	Council Member of Society for Pediatric Research
2000-2002	Program Committee for American Society of Human Genetics
2002-2004	Board of Directors of the American Society of Human Genetics

National Committees:

1995-2000	Member of NIH Genetic Determinants of High Blood Pressure Data Safety Monitoring Board
1993-1999	Ad hoc member of NIH study sections for NCHGR, NHLBI, NCI, and NEI
1993	Retinitis Pigmentosa Foundation Grant Review Study Section (Co-Chairman)
1994-1999	Member of NIH Marshfield Clinic Genotyping Center Review Committee
1997-2002	Member of NIH Center for Inherited Disease Research grant review panel
1999-2002	Council Member of Society for Pediatric Research
1999-2003	Member NIH Genome Research Review Committee
1999	Member of Special NIH Panel to review sequencing centers
2000	NHGRI Blue Ribbon Review Panel
2002	NHGRI Genetics Disease Research Branch Review Panel
2006-2010	National Eye Institute Advisory Council
2010-present	NIH Undiagnosed Diseases Program Advisory Panel
2012-present	NIH NHGRI H3Africa Advisory Panel
2014-present	NIH NHGRI Council

Reviewer
American Journal of Human Genetics
American Journal of Medical Genetics
Genome Research
Genomics
Human Genetics
Human Molecular Genetics
Nature
Nature Genetics
Science

Professional Affiliations
American Association for the Advancement of Science
American Pediatric Society
American Society of Human Genetics

Association for Research in Vision and Ophthalmology
Association of American Physicians
Association of Professors of Human Medical Genetics
Society for Pediatric Research
Institute of Medicine of the National Academies

B. **Clinical assignments**

University of Iowa Hospitals and Clinics, Genetics clinic
University of Iowa Hospitals and Clinics, Metabolic clinic
Regional Genetics Consultation Service Clinics
University of Iowa Hospitals and Clinics, Genetics Consultation Service