

Curriculum Vitae

Brian P. Brooks, M.D., Ph.D.

Ophthalmic Genetics & Visual Function Branch, National Eye Institute
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Employment

- 2008- present Tenure-track investigator, National Eye Institute
- 2009-present Adjunct Appointment, National Human Genome Research Institute
- 2004-2008 Staff Clinician, National Eye Institute
NEI Physician-Scientist Development Program
- 2004 - present Volunteer Staff, Children's National Medical Center,
Department of Ophthalmology, Director, Ophthalmic Genetics Clinic
- 2005-2007 Founding Director, National Ophthalmic Genotyping Network (eyeGENE)
- 2005-present Adjunct Assistant Professor, Dpt. of Biochemistry and Molecular & Cellular
Biology, Georgetown University
- 2008-present Director, Ophthalmic Genetics Fellowship Training Program, NEI
- 2007-present Director, Ophthalmic Genetics Rotation, NHGRI

Education and Training

- 2005 Diplomate, American Board of Medical Genetics (specialty boards)
- 2002-2004 Fellow, Medical Genetics, National Human Genome Research Institute, NIH
- 2002 Diplomate, American Board of Ophthalmology (specialty boards)
- 2001-2002 Fellow, Pediatric Ophthalmology, University of Michigan
- 1998 – 2001 House Officer, Dpt. Ophthalmology, University of Michigan
Chief resident, 2000-2001
- 1997-1998 House Officer, Dpt. Internal Medicine, Allegheny University, Graduate
Division; Philadelphia, PA
- 1989 - 1997 University of Pennsylvania School of Medicine, Philadelphia, PA
 - ◆ Doctor of Medicine (M.D.)
 - ◆ Doctor of Philosophy (Ph.D.)
Thesis : *Characterization of a Normal and Expanded-Repeat Androgen
Receptor in Neuronal Cells* (Advisor: Dr. Kenneth Fischbeck)
 - ◆ Funded by the Medical Scientist Training Program
- 1985 - 1989 University of Maryland, College Park
 - ◆ B.S., Biochemistry
 - ◆ *summa cum laude* (4.0 GPA)
 - ◆ With General Honors (Thesis : *Love as a Theme in the Epistles of John*)
 - ◆ With Honors in Zoology (Thesis : *Characterization of Receptors for [³H]
Opipramol in Rat Brain*, Advisor: Dr. Solomon Snyder)

Awards and Honors

- Camras Translational Research Award, American Association for Research in Vision and Ophthalmology (2011)
- University of Michigan Department of Ophthalmology, Distinguished Alumnus Award (2011)
- Presidential Early Career Award for Scientists and Engineers (2010)—*the nation's highest award for early career scientists and engineers*
- NEI Director's Award (2010)—“in recognition for his outstanding contributions to basic, clinical, and multidisciplinary research which personifies the highest standards of NIH Intramural Research Program.”
- NEI Director's Award (2009) –two separate awards for eyeGENE and for IC service
- NIH Director's Award (2008)
- Young Investigator Award, American Association of Pediatric Ophthalmology & Strabismus (2007)
- Joint National Eye Institute/National Human Genome Research Institute Physician-Scientist Development Program (2005)
- Heed Foundation Fellowship (2001-2002)
- Resident research award, 3rd place, University of Michigan (2001)
- Michigan Ophthalmologic Society Resident Research Award (2000)
- The George Slocum Resident Research Award, Department of Ophthalmology (2000)
- The Walter R. Parker Resident Teaching Award, 3rd place, awarded twice (2000, 2001)
- Dr. O. H. Perry Pepper Prize for Clinical and Research Excellence (1997)
- Board of Directors, Philadelphia Medical Society (1994-1996)
- Alpha Omega Alpha Medical Honor Society (elected as a junior, 1994)
- National Eye Institute Woods Hole summer course scholarship (1994)
- Society of Nuclear Medicine Research Award (1991)
- Eagle Scout, silver palm, Boy Scouts of America
--Vigil Honor member of Order of the Arrow

Grants/Funding

- 2011 NIH Director's Challenge Award
- 2009-2010 Blind Children's Center, \$16,850 for pre-clinical work on treatments for albinism
- 2009 NIH Director's Special Project Award \$850,000
- Intramural program funding, National Eye Institute

Patents

- Nitissinone (NTBC) for the treatment of albinism and to increase pigmentation (2010, patent pending)

Student Mentoring

- Lan Chang, M.D., Clinical Research Training Program (2004-2005), now ophthalmology resident, Duke University Medical Center
- Mohit Mehtani, M.D., Georgetown University Medical Student (2005), now ophthalmology resident, George Washington University School of Medicine
- Jacob D. Brown, Ph.D., Georgetown University Physician-Scientist Training Program (2005-2008), Ph.D. advisor, completing medical school, Georgetown University, accepted, resident in radiology, University of California, San Francisco
- Amana Akhtar, M.D., Howard Hughes Medical Institute Training Program (2006-2007), currently, radiology resident, University of Pennsylvania
- Elizabeth Garabedian (2008), NIH Summer Internship Program, currently undergraduate, University of Notre Dame
- Lorenzo Nichols, II (2008-2009), NIH Undergraduate Scholarship Program for Disadvantaged Individuals, currently optometry student, University of Puerto Rico
- Anand Bhat (2009, 2010, 2011), NIH Summer Internship Program, currently undergraduate, Harvard University
- Miguel Enongene (2009-present), Bowie State University undergraduate
- Holly Babcock (2009-2011), Intramural research training associate, applying to genetic counseling programs
- Shahila Sriskanda (2010-2011), Intramural research training associate, applying to graduate schools
- Athenais Lapeyre (2010-present), high school student

Service/Consulting

- (2008-present) Board of Senior Consultants for the NIH-wide Undiagnosed Diseases Initiative
- (2008) Molecular Genetics Testing Subcommittee of the NIH Medical Executive Committee
- (2008) Education Subcommittee, American Association of Pediatric Ophthalmology & Strabismus
- (2005-present) Clinical advisor, GeneDx, Inc., Gaithersburg, MD (unpaid)

Invited Lectures

- Visiting professor, University of Iowa, Scheduled February 2012
- Visiting professor, University of California, San Francisco, Schedule January 2012
- The Genetics of Uveal Coloboma (University of Michigan, Distinguished Alumnus Talk, September 2011)
- Genetic testing in optometric practice (American Academy of Optometry, November 2010, San Francisco, CA)
- The Genetics of Uveal Coloboma (American Society of Human Genetics, November 2010, Washington, DC)
- Visiting professor, Boston Children's Hospital/Harvard Medical School, May 2010
- Genetic Testing and Genetic Counseling in Inherited Corneal Disease, World Cornea Symposium (April 2010, Boston, MA) The Genetics of Uveal Coloboma, Medical College of Wisconsin (February 3, 2010, Milwaukee, WI)
- Clinical and Basic Science Studies of Albinism at the NEI (Vision of Children Symposium, San Diego, CA, May 2009)
- How Can New Genetics Techniques Aid in the Diagnosis and Treatment of Inherited Corneal Disease? (Fifth Annual ARVO/Pfizer Ophthalmics Research Institute: Corneal Dystrophies: Molecular Genetics to Therapeutic Intervention., May 2009)
- Genetics of Uveal Coloboma (Genetics & Genomics of Vision Symposium, National Eye Institute's 40th Anniversary Celebration, April 2009)
- Genetics of Ocular Malformations (American Academy of Ophthalmology and AAPOS Meeting, November 2008, Symposium on the Genetic Basis of Pediatric Eye Disease)
- Genetics of Uveal Coloboma and Molecular Diagnosis in Ophthalmology (University of Alberta, Edmonton, CA, September 2008)
- Molecular Diagnosis in Ophthalmology (University of Cincinnati, May 28, 2008)
- Phenotype-Genotype and Beyond (Association for Vision Research and Ophthalmology Meeting, Symposium on Frontiers in Genetics, April 27, 2008)
- The Genetics of Uveal Coloboma (University of California, Berkeley, April 12, 2008)
- Molecular Diagnosis in Ophthalmology, (Retina Subspecialty Day, American Academy of Ophthalmology, November 2007)
- American Association of Pediatric Ophthalmology & Strabismus, Young Investigator Award Lecture (April 2007)
- Xeroderma Pigmentosum & Other Disease of Human Premature Aging & DNA Repair: From Patients To Molecules (September 2006)
- The Genetics of Uveal Coloboma (Georgetown University, April 2006)
- Ocular abnormalities in *Rab38* Mutant Mice (The Vision of Children Foundation Symposium, New York, New York, October 2004)
- European School of Genetics, Course on Human Genetics, Tunisia, Tunis (November 2004)

Teaching

- Pediatric ophthalmology fellows and ophthalmology residents (George Washington University, Georgetown University) at Children's National Medical Center, Washington, D.C. (July 2005-present)
- Ophthalmology fellows, National Eye Institute (2005-present)
- Ophthalmology residents, Howard University (2008-present)
- Medical genetics fellows, NHGRI (2005-present)
- Instructor, Joint Johns Hopkins-NHGRI Genetic Counseling Program (2004)

- Instructor, Foundation for Advanced Education in the Sciences, Introduction to Medical Genetics (2002-present)
- Lecturer, Medical Student Ophthalmology Rotation, University of Michigan (1999-2002)
- Instructor, Cardiovascular Pharmacology, University of Pennsylvania (1994-6)

Publications

ORIGINAL ARTICLES IN PEER-REVIEWED JOURNALS

[Listed in chronological order.]

Battaglia G, **Brooks BP**, Kulsakdinun C, and DeSouza EB. Pharmacological profile of MDMA (3,4-methylenedioxymethamphetamine) at various brain recognition sites. *Eur. J. Pharmacol.* 149:159-163, 1988.

Ferris CD, Hirsch DJ, **Brooks BP**, Snowman AM, Snyder SH. [³H]Opipramol labels a novel binding sites and sigma receptors in rat brain membranes. *Mol. Pharmacol.* 39:199-204, 1991.

Ferris CD, Hirsch DJ, **Brooks BP**, Snyder SH. Sigma receptors: from molecule to man. *J. Neurochem.* 57:729-737, 1991.

Mackler SA, **Brooks BP**, Eberwine JH. Stimulus-induced coordinate changes in mRNA abundance in single postsynaptic hippocampal CA1 neurons. *Neuron.* 9:539-548, 1992.

Chumpradit S, Kung M P, Panyachotipun C, Prapansiri V, Foulon C, **Brooks BP**, Szabo SA, Tejani-Butt S, Frazer A., Kung HF. Iodinated tomoxetine derivatives as selective ligands for serotonin and norepinephrine uptake sites. *J Med Chem.* 35: 4492-7, 1992.

Brooks BP, Paulson HF, Merry DE, Salazar-Grueso EF, Wilson EM, Fischbeck KH. Characterization of an expanded-glutamine repeat androgen receptor in a neuronal cell culture model system. *Neurobiol. Dis.* 4:313-323, 1997.

Brooks BP, Merry DE, Paulson HF, Lieberman AP, Kolson DL, Fischbeck KH. A cell culture model for androgen effects in motor neurons. *J. Neurochem.* 70:1054-1060, 1998.

Brooks BP, Simpson JL, Leber SM, Robertson PL, Archer SM. Infantile spasms as an acquired cause of perinatal visual loss. *J. AAPOS* 6:385-8, 2002.

Kriederman BM, Myloyde TL, Witte MH, Dagenais SL, Witte CL, Rennels M, Bernas MJ, Lynch MT, Erickson RP, Caulder MS, Miura N, Jackson D., **Brooks BP**, Glover TW. *Foxc2* haploinsufficient mice are a model for human autosomal dominant lymphedema-distichiasis syndrome. *Hum Mol Genet* 12: 1179-1185, 2003.

Brooks BP, Dagenais SL, Nelson CC, Glynn MW, Caulder MS, Downs C., Glover TG. Mutations in the *FOXC2* gene in familial distichiasis. *J-AAPOS.* 7:354-357, 2003.

Brooks BP, Moroi SE, Downs CA, Wiltse S, Othman MI, Semina EV, Richards JE. A novel mutation in the *PITX2* gene in a family with Axenfeld-Rieger syndrome. *Ophthalmic Genetics.* 25:57-62, 2004.

Brooks BP, Kleta R, Caruso RC, Stuart C, Ludlow J, Stratakis CA. Triple-A syndrome with prominent ophthalmic features and a novel mutation in the *AAAS* gene: a case report. *BMC Ophthalmology.* 4:7, 2004.

Espinoza MH, Ganga M, Vadlamudi U, Martin DM, **Brooks BP**, Semina EV, Murray JC, Amendt BA. Protein kinase C phosphorylation modulates N- and C-terminal regulatory activities of the PITX2 homoeodomain protein. *Biochemistry*. 44:3942-3954. 2005.

Brooks BP, Kleta R, Stuart C, Tuchman M, Jeong A, Stergiopoulos S, Bei T, Bjornson B, Russel L, Chanoine JP, Tsagarakis S, Kalsner LR, Stratakis CA. Genetic heterogeneity and clinical phenotype in triple-A syndrome: a review of the NIH experience 2000-2005. *Clin Genetics*. 68:215-221. 2005.

Brooks BP, Meck JC, Bendavid C, Haddad B, Blain D, Toretsky JA. Unbalanced 16p;13q translocation in a patient with factor VII deficiency and developmental abnormalities. *BMC Medical Genetics*. 7:2 (2006)

Tang J, Gokhale PA, Brooks SE, Blain D, **Brooks BP**. Increased corneal thickness in patients with ocular coloboma. *J-AAPOS*. 10:175-7 (2006).

Brooks BP, Larson DM, Chan CC, Kjellstron S, Smith RS, Crawford MA, Lamoreux L, Huizing M, Hess R, Jiao X, Hejtmancik FJ, Maminishkis A, John SWM, Bush R, Pavan WJ. Analysis of ocular hypopigmentation in Rab38^{cht/cht} mice. *Inv Ophthalm Vis Sci*. 48:3905-13 (2007).

Doherty ES, Lachawan F, Hadley DW, Brewer C, Zalewski C, Kim HJ, Glass P, Solomon B, Rosenbaum K, Domingo DL, Hart TC, **Brooks BP**, Immken L, Lowry RB, Kimonis V, Shanske AL, Knightly C, McDonald-McGinn Dm Zackai EH, Muenke M. Muenke syndrome (FGFR3-related-craniosynostosis). Expansion of the phenotype and literature review. *Am J Med Genet*. 143:3204-15 (2007)

Goodwin H, **Brooks BP**, Porter FD. Acute postnatal cataract formation in Smith-Lemli-Opitz Syndrome. *Am J Med Genet*. 146:208-11 (2008)

Meredith M, Gordon L, Clauss S, Sachdev V, Smith A, Perry M, Brewer C, Zalewski C, Kim J, Solomon B, **Brooks BP**, Gerber L, Turner M, Domingo D, Hart TC, Graf J, Reynolds J, Gropman A, Yanovski J, Collins F, Nabel E, Cannon R, Gahl W, Introne W. Comprehensive evaluation of patients with Hutchinson-Gilford progeria syndrome. *NEJM*. 358:592-604 (2008).

Mahindra P, DiGiovanna JJ, Tamura D, Brahim JS, Hornyak TJ, Stern JB, Lee CC, Khan SG, **Brooks BP**, Smith JA, Driscoll BP, Montemarano AD, Sugarman K, Kraemer KH. Skin cancers, blindness, and anterior tongue mass in African brothers. *J Am Acad Dermatol*. 59:8881-6 (2008)

Alur RR, Cox TA, Crawford MA, Gong X, **Brooks BP**. Optic nerve axon number in mouse is regulated by Pax2 in mouse. *J-AAPOS*. 12:117-21 (2008).

Priya M, John JD, Tamura D, Brahim JS, Hornyak TJ, Stern JB, Lee CR, Khan SG, **Brooks BP**, Smith JA, Driscoll BP, Montemarano AD, Sugarman K, Kraemer KH. Skin cancers, blindness, and anterior tongue mass in African brothers. *J Acad Derm*. 59:881-6 (2008).

Asai-Coakwell M, French CR, Ye M, Garcha K, Bigot K, Perena AG, Staehling-Hampton K, Mema S, Chanda B, Mushegian A, Bamforth S, Doschak MR, Li G, Dobbs MB, Giampietro PF, **Brooks BP**, Vijayalakshmi P, Sauvei Y, Abitbol M, Sundaresan P, van Heyningen V, Pourquie O, Underhill TM, Waskiewicz AJ, Lehmann OJ. Incomplete penetrance and phenotypic variability characterize Gdf6-attributable oculo-skeletal phenotype. *Hum Molec Genetics*. 15:1110-21 (2009)

Brown JD, Duuta S, Bharti K, Bonner RF, Munson PJ, Dawid IB, Akhtar AL, Onojafe IF, Alur RP, Gross JM, Hejtmancik JF, Jiao X, Chan WY, **Brooks BP**. Expression profiling during ocular development

identifies two *Nlz* genes with a critical role in optic fissure closure. *Proc Natl Acad Sci U S A*. 106:1462-7 (2009).

Solomon BD, Pineda-Alvarez DE, Balog JZ, Hadley D, Gropman AL, Nandagopal R, Han JC, Hahn JS, Blain D, **Brooks B**, Muenke M. Compound heterozygosity for mutations in *PAX6* in a patient with complex brain anomaly, neonatal diabetes mellitus, and microphthalmia. *Am J Med Genet*. 149A:2543-6 (2009)

Salchow DJ, Kohlhase J, Miller M, Kadon N, FitzGibbon EJ, Caruso RC, **Brooks BP**. Absent optic chiasm presenting with horizontal nystagmus. *J Pediatr Ophthalmol Strabismus*. 47: 187-91 (2010)

Alur RP, Camasamudram V, Brown JD, Mehtani M, Onojafe IF, Sergeev YV, Boobalan E, Jone MP, Tang K, Liu H, Chun-hong X, Gong X, **Brooks BP**. Papillorenal syndrome-causing missense mutations in *PAX2/Pax2* result in hypomorphic alleles in mouse and human. *PloS Genet* 6(3):e1000870. Doi 10.1371/journal.pgen.100870. (2010)

Nichols L, Alur RP, Boobalan E, Caruso RC, Stone EM, Swaroop A, Johnson MA, **Brooks BP**. Molecular characterization of two novel *CRX* mutations causing autosomal dominant Leber congenital amaurosis. *Human Mutation*. 31:E1472-83 (2010)

Zhou X, Khan SG; Tamura D, Patronas NJ, Zein WM, **Brooks BP**, Kraemer KH. Brittle hair, developmental delay, neurologic abnormalities and photosensitivities in a 4 year old girl. *J. Amer.Acad of Dermatology*. 32:323-328 (2010).

Weiss JS & the **Fifth ARVO/Pfizer Ophthalmics Research Institute Conference Working Group**. Corneal dystrophies: Molecular genetics to therapeutic intervention—Fifth ARVO/Pfizer Ophthalmics Research Institute Conference. *Inv Ophthalmol Vis Sci*. 51:5391-5402 (2010).

Zeeberg BR, Liu H, Kahn AB, Ahler M, Rajapakse VN, Bonner RF, Brown JD, **Brooks BP**, Larionov VL, reinhold W, Weinstein JN, Pommier YG. RedundancyMiner: De-replication of redundant GO categories in microarray and proteomics analysis. *BMC Bioinformatics*. 12:52 (2011). [Epub ahead of print.]

Feuillan PP, Ng D, Han JC, Sapp JC, Wetsch K, Spaulding E, Zheng YC, Caruso RC, **Brooks BP**, Johnston JJ, Yanovski JA, Biesecker LG. Patients with Bardet-Biedl syndrome have hyperleptinemia suggestive of leptin resistance. *J. Clin Endocrinol Metab*. 96:E528-35.

Skeens HM, **Brooks BP**, Holland EJ. Congenital aniridia variant: minimally-abnormal irides with severe limbal stem cell deficiency. *Ophthalmology*, 2011 March 2 [Epub ahead of print]

Ehler M, Rajapakse VN, Zeeberg BR, **Brooks BP**, Brown J, Czaja K, Bonner RF. Nonlinear gene cluster analysis with labeling for microarray gene expression data in organ development. *BMC Proc*. 28:5 (2011)

Ramkumar HL, **Brooks BP**, Cao X, Tamura D, Digiovanna JJ, Kraemer KH, Chan CC. Ophthalmic manifestations and histopathology of xeroderma pigmentosum: two clinicopathological cases and a review of the literature. *Surv. Ophthalmol*. 56:348-61 (2011).

Brooks BP, Thompson AH, Clayton JA, Chan CC, Tamura D, Zein WM, Blain D, Hadsall C, Rowan J, Bowles KE, Khan SG, Ueda T, Bowle J, Oh KS, DiGiovanna JJ, Kraemer KH. The ocular manifestations of trichothiodystrophy. *Ophthalmology*, 2011 Sep 27. [Epub ahead of print]

Onojafe IF, Adams DR, Simeonov DR, Zhang J, Chan CC, Bernardini IM, Sergeev YV, Dolinska MB, Alur RP, Brilliant MH, Gahl WA, **Brooks BP**. Nitisinone improves eye and skin pigmentation defects in a mouse model of oculocutaneous albinism. *J Clin Invest*, 2011 Sep 26 [Epub ahead of print]

Tamura D, Meredith M, Digiovanna JJ, Zhou X, Tucker MA, Goldstein AM, **Brooks BP**, Khan SG, Oh KS, Ueda T, Boyle J, Mosiehi R, Kraemer KH. High-risk pregnancy and neonatal complications in the DNA repair and transcription disorder trichothiodystrophy: report of 27 affected pregnancies. *Prenat Diag*. 2011 31:1046-53.

Pindea-Alvarez DE, Solomon BD, Roessier E, Balog JZ, Hadley DW, Zein WM, Hadsall CK, **Brooks BP**, Muenke M. A broad range of ophthalmic anomalies is part of the holoprosencephaly spectrum. *Am J Med Genet A*. 2011. 155:2713-20.

INVITED EDITORIALS

Brooks BP. *Albinism: Making Progress*. *J-AAPOS*. 15:1-2 (2011)

American Association of Pediatric Ophthalmology and Strabismus: Position Paper: "On the Use Of 'Stem Cells' For Optic Nerve Hypoplasia." *Ophthalmology*. 118:795-6 (2011). **Served as chair of the writing committee.**

REVIEW ARTICLES AND BOOK CHAPTERS

Brooks BP and Fischbeck KH. Spinal and bulbar muscular atrophy: a trinucleotide repeat expansion neurodegenerative disease. *Trends Neurosci*. 18:459-461, 1995.

Brooks BP, Richards JE, Lichter PR. "Heredity and Glaucoma" In: Tasman W, Jaeger EA, eds. *Duane's Foundations of Clinical Ophthalmology*, Philadelphia: Lippincott Williams & Wilkins, 2002; Vol 3, Ch 5, 1-16.

Brooks BP and Sugar A. "Fabry's Disease" in *The Eye Pathologist* (Website), ed. G. Klintworth. www.eyepathologist.org. 2004.

Huizing M, **Brooks BP**, Anikster Y. Optic atrophies in metabolic disorders. *Mol Genet Metab*. 86:51-60 (2005)

Huizing M, **Brooks BP**, Anikster Y. Optic neuropathies in inherited metabolic disorders. *Ped Endocrin Rev*. 3:97-103(2005).

Brooks BP and Traboulsi EI. "Congenital Malformations of the Eye." In: Tasman W, Jaeger EA, eds. *Duane's Foundations of Clinical Ophthalmology*, Philadelphia: Lippincott Williams & Wilkins, 2005; Vol 1, Ch 40, 1-28.

Alur RP, **Brooks BP**. Clinical and genetic analysis of coloboma: a review. *Asian J Exp Sci*. 20:1-15. (2006)

Brooks BP and Traboulsi EI. "Congenital Optic Nerve Anomalies" In: Tasman W, Jaeger EA, eds. *Duane's Foundations of Clinical Ophthalmology*, Philadelphia: Lippincott Williams & Wilkins, 2006.

Chang L, Blain D, Bertuzzi S, **Brooks BP**. Uveal coloboma: clinical and basic science update. *Current Opinions in Ophthalmology*. 17:447-70 (2006).

Blain D and **Brooks BP**. Molecular testing and genetic counseling in ophthalmology. *Archives of Ophthalmology*. 125:196-203 (2007).

Kraemer KH, Patronas NJ, Schiffmann R, **Brooks BP**, Tamura D, Digiovanna JJ. Xeroderma pigmentosum, trichothiodystrophy, and Cockayne syndrome: A complex genotype-phenotype relationship. *Neuroscience*. 145:1388-96 (2007).

MacDonald IM, **Brooks BP**, Sieving PA. Eyeing a new network. *Science*. 318:1068 (2007)

Brooks BP, MacDonald IM, Tumminia SJ, Smaoui N, Blain D, Nezhuvungal AA, Sieving PA. Genomics in the Era of Molecular Ophthalmology: Reflections on the National Ophthalmic Disease Genotyping Network (eyeGENE™). *Arch Ophthalmol*. 126:424-5 (2008).

Brooks BP and Traboulsi EI. “Anophthalmia, Microphthalmia and Optic Fissure Closure Defects.” *Oxford Guide to Inherited Eye Disorders*. In press 2011.

Brooks BP and Traboulsi EI. “Congenital Optic Nerve Anomalies.” *Oxford Guide to Inherited Eye Disorders*. In press 2011.

Zein WM, Wang X, **Brooks BP**. “Congenital Optic Nerve Anomalies.” *Pediatric Ophthalmology and Strabismus*. Ed. Kenneth Wright. In press 2011.

Brooks BP. “Congenital Ocular Abnormalities” and “Optic Nerve Disorders.” *Principles and Practice of Medical Genetics*. Ed. Emery, Rimoin & Korf. In press 2011.

Clinical Research Protocols

Principal investigator, Open

- Generation of induced pluripotent stem (iPS) cell lines from somatic cells of Best disease, late onset retinal degeneration (L-ORD) and age-related macular degeneration (AMD) patients (11-EI-0245)
- The National Ophthalmic Disease Genotyping and Phenotyping Network, Phase II (10-EI-N164)
- National Ophthalmic Disease Genotyping Network, Stage 1—Creation of DNA repository for inherited ophthalmic diseases (06-EI-0236)
- Family studies of uveal coloboma (06-EI-0230)
- Ophthalmic Genetics & Visual Function Branch/NEI Repository (08-EI-N014)

Principal Investigator, Now Closed

- Correlation of gene abnormalities with clinical manifestations in aniridia (06-EI-0044)

Associate Investigator, Open

- Genetics and clinical characteristics of Bardet-Biedl syndrome (04-HG-0123)

- Genetic and clinical study of patients with xeroderma pigmentosum, Cockayne syndrome, or trichothiodystrophy (NCI-99-C-0099)
- Clinical study of Muenke syndrome (FGFR3-related craniosynostosis) (05-HG-0131)
- Clinical and laboratory study of methylmalonic aciduria (04-HG-0127)
- Clinical and Genetic Studies on Holoprosencephaly (04-HG-0093)
- WAGR Syndrome and Other 11p Contiguous Gene Deletions: Clinical Characterization and Correlation with Genotype (08-CH-0213)
- Clinical, Cellular and Molecular Investigations into Oculocutaneous Albinism (09-HG-0035)
- Investigation of Neurodegeneration in Glycosphingolipid Storage Disorders (02-DK-0107)
- Immunopathology and Ocular Pathology of Ocular Disease in Humans (92-EI-0013).
- Study of Proteus syndrome and related congenital disorders (94-HG-0132)