Harvard Medical School/Harvard School of Dental Medicine
Curriculum Vitae

Date Prepared: **November 13, 2018**

Name: **Janey L. Wiggs**

Education

1976	BA	Biochemistry	University of California, Berkeley
1981	PhD	Biochemistry (Michael J. Chamberlin PhD, advisor)	University of California, Berkeley
1985	MD	Medicine	Harvard Medical School

Postdoctoral Training

1984*-1986	Intern/Resident	Medicine	Beth Israel Hospital, Boston
1986-1987	Fellow	Molecular Genetics (Ted Dryja MD, Advisor)	Mass Eye and Ear
1987-1990	Resident	Ophthalmology	Mass Eye and Ear
1990-1991	Fellow	Molecular Genetics (Ted Dryja MD, Advisor)	Mass Eye and Ear
1991-1992	Fellow	Glaucoma (David Epstein MD, Advisor)	Mass Eye and Ear
1994-1996	Fellow	Medical Genetics (Diana Bianchi MD, Advisor)	Tufts Medical School

*Started internship during 4th year of medical school

Faculty Academic Appointments

07/92-09/93	Instructor	Ophthalmology	Harvard Medical School
10/93-02/01	Assistant Professor	Ophthalmology and Genetics	Tufts Medical School
03/01-11/05	Assistant Professor	Ophthalmology	Harvard Medical School
12/05-12/11	Associate Professor	Ophthalmology	Harvard Medical School
01/12-04/15	Paul Austin Chandler Associate Professor	Ophthalmology	Harvard Medical School
05/15-	Paul Austin Chandler Professor	Ophthalmology	Harvard Medical School

Appointments at Hospitals/Affiliated Institutions

07/92-09/93	Assistant Surgeon	Ophthalmology	Massachusetts Eye and Ear
10/93-02/01	Assistant Professor	Ophthalmology	New England Medical Center
03/01-11/05	Assistant Surgeon	Ophthalmology	Massachusetts Eye and Ear
12/05-04/15	Associate Surgeon	Ophthalmology	Massachusetts Eye and Ear
05/15-	Surgeon	Ophthalmology	Massachusetts Eye and Ear

04/05-06/08	Clinical Associate	Surgical Service	Massachusetts General Hospital
07/08-	Clinical Associate	Ophthalmology Service	Massachusetts General Hospital

Other Professional Positions

2003-	Scientific Advisory Board	Glaucoma Research Foundation
2012-	Scientific Advisory Board	Glaucoma Foundation
2013-	Scientific Advisory Board	Research to Prevent Blindness
2016-	Associate Member	Broad Institute of Harvard and MIT

Major Administrative Leadership Positions

Local

1994	Symposium organizer, "Molecular Genetics of Ocular Disease"	Tufts Medical School
1995-2001	Co-director Gene 201-202, "Introduction to Genetics"	Tufts Medical School
2009-	Associate Director Howe Laboratory	Massachusetts Eye and Ear
2010-2014	Associate Chief for Clinical Research	Massachusetts Eye and Ear
2010-	Co-director, Glaucoma Center of Excellence	Massachusetts Eye and Ear
2011-	Associate Director of the Ocular Genomics Institute	Massachusetts Eye and Ear
2011-	Director, Ocular Genetics Diagnostics Laboratory	Massachusetts Eye and Ear
2014-	Associate Chief for Clinical Research (Ophthalmology)	Massachusetts Eye and Ear
2014-	Vice Chair for Clinical Research (Ophthalmology)	Harvard Medical School

National and International

2009-2010	Chair, PhenX Ocular Working Group	National Human Genomics Research Institute (NHGRI)
2005-2012	Chair, Genetics Cross Sectional Group	Association for Research in Vision and Ophthalmology (ARVO)
2002-2006	Scientific Advisory Council	National Eye Institute (NEI), National Institutes of Health (NIH)
2009-	US Chair, US-INDO JWG (joint working group)	National Eye Institute (NEI), Department of Biotechnology, India (DBT)

Committee Service

Local

1994-2001	Executive Committee, Graduate Program in Genetics	Tufts Medical School
1994-2001	Executive Committee: M.D.-Ph.D. Program	Tufts Medical School
1998-2001	Tufts University-wide Committee on Genetics	Tufts Medical School
2001-2003	MEEI/SERI Joint Clinical Research Center Joint Governance Committee	Massachusetts Eye and Ear
2002-	Glaucoma Fellowship Selection Committee	Massachusetts Eye and Ear
2003-	Human Studies Committee (Institutional Review Board)	Massachusetts Eye and Ear
2005-2012	Patent Committee	Massachusetts Eye and Ear
2005-	Residency Selection Committee	Massachusetts Eye and Ear
2007-	Research Leadership Committee	Massachusetts Eye and Ear
2007-	Medical Board	Massachusetts Eye and Ear
2007-	Admissions Committee	Harvard Medical School (HST)
2013-	Genomics Working Group	Partner's Health Care
2016-	Promotions and Reappointments	Harvard Medical School
2018-	Data & Tissue Sharing Committee	Partner's Health Care

National and International

1998	National Eye Institute 5-year planning panel	National Eye Institute (NIH)
2009	Genetics and Genomics strategic planning group	National Eye Institute (NIH)
2011	National Eye Institute 5-year planning panel	National Eye Institute (NIH)

Professional Societies

1986-	Association for Research in Vision and Ophthalmology (ARVO)	
	2005-2012	Chair, Genetics Cross-sectional group
	2004-2008	Member, Publications Committee
	2005-2012	Member, Program Planning Committee
1991-	American Academy of Ophthalmology	
1991-	American Society of Human Genetics	
1992-	Chandler Grant Society	
2010-	American Glaucoma Society	
2013-2016	Member, American Glaucoma Society Program Planning Committee	
2012-	New England Ophthalmological Society	
2013-	American Ophthalmological Society	Elected Member
2015-	Academia Ophthalmologica Internationalis (Chair LXXX)	Elected Member
2016-	Glaucoma Research Society	Elected Member
2018-	Glaucoma Research Society	Program Planning Committee
2018-	National Academy of Medicine (NAM)	Elected Member

Grant Review Activities

1995-200	American Health Assistance Foundation	Reviewer
1996-2002	NEI Special Emphasis Study section	Adhoc reviewer
1998-2002	NEI Visual Sciences "A" study section	Adhoc reviewer
1998-2002	NEI Training Grant Study Section	Reviewer
2010-2013	NEI P30 Special Emphasis	Adhoc reviewer
2011-2013	NHGRI Genomes Health and Disease	Adhoc reviewer
3/28/2013	NEI ZEY1-VSN-01	Member
3/18/2014	NEI ZRG1-BDCN-H-02	Member
8/1/2014	NEI ZEY1-VSN-02	Chair
11/5/2014	NEI ZRG1-BDCN-L-02	Member
1/26/2015	NIDDK ZDK1-GRB-S-M3	Member
3/27/2015	NEI ZEY1-VSN-03	Member
3/14/2016	NEI ZEY1-VSN-03	Chair
6/16/2016	NHGRI Genomes Health and Disease	Adhoc reviewer
11/2/2017	NHGRI Genomes Health and Disease	Adhoc reviewer

Editorial Activities

Adhoc reviewer: Science, Nature Genetics, American Journal of Human Genetics, Human Molecular Genetics, Journal of Clinical Investigation, Journal of Medical Genetics, Human Genetics, Ophthalmology, American Journal of Ophthalmology, British Journal of Ophthalmology, Molecular Vision, Clinical Genetics, Gene, PLoS Genetics, PLoS One, BMC Genetics, BMC Medical Genetics, Human Mutation, Genomics, Eye

Other Editorial Roles:

1995-1997	Investigational Ophthalmology and Visual Science (IOVS)	Editorial Board Member
1999-2001	Eyenet	Editorial Board Member
1999-	Journal of Glaucoma	Editorial Board Member
200-2004	Ophthalmic Genetics	Editorial Board Member
2005-2012	Archives of Ophthalmology, Ocular Genetics Section	Associate Editor
2008-	Investigational Ophthalmology and Visual Science (IOVS)	Editorial Board Member
2010-	Molecular Vision	Editorial Board Member
2013-2016	JAMA Ophthalmology	Editorial Board Member
2016-	JAMA Ophthalmology	Assoc. Deputy Editor
2013-	Annual Reviews in Visual Science	Editorial Board Member

Honors and Prizes

1983-1984	Pearl and Martin Silverstein scholar in Health Sciences and Technology	MIT/Harvard Division of Health Sciences and Technology (HST)	Academic Scholarship
1990-1992	Heed/Knapp Foundation Award	Heed Foundation	Academic
1990-1992	Fight for Sight Awards	Fight for Sight	Academic
1991-1993	Knights Templar Foundation Award	Knights Templar	Academic

1993	Teacher of the Year	Massachusetts Eye and Ear	Teaching
1994	Earl P. Charlton Research Award	New England Medical Center	Academic
1995	Miriam & Benedict Wolf Scholars Award	Research to Prevent Blindness	Academic
1997	Glaucoma Research Foundation Award	Glaucoma Research Foundation	Academic
2003	Honor Award	American Academy of Ophthalmology	Academic
2006	Lew Wassserman Merit Award	Research to Prevent Blindness	Academic
2009	Silver Fellow	Association for Research in Vision and Ophthalmology (ARVO)	Academic
2010	Gold Fellow	Association for Research in Vision and Ophthalmology (ARVO)	Academic
2013	Audacious Goal Winner	National Eye Institute (NEI), National Institutes of Health (NIH)	Academic
2013	Alcon Research Institute Award	Alcon Research Institute	Academic
2014	Member	American Ophthalmological Society	Academic
2015	Member	Academia Opthalmologica Internationalis	Academic
2016	Member	Glaucoma Research Society	Academic
2017	Translational Research Award	Asia ARVO Translational Research Summit	Academic
2018	David Epstein Award	ARVO Foundation	Academic
2018	Member	National Academy of Medicine	Academic

Report of Funded and Unfunded Projects

Funding Information

Past

1985-1986 Genetic Studies of Retinitis Pigmentosa
NEI/NRSA
PI
Supported post-doctoral studies designed to identify genes contributing to retinitis pigmentosa

1993-2008 Linkage Study of Juvenile Glaucoma
NEI/RO1 EY09847
PI
The goal of this study was to identify genes responsible for early-onset forms of glaucoma.

1995-2005 Familial Primary Open Angle Glaucoma
NEI/RO1 EY010886
PI
The overall goal of this study was to identify genetic loci that contribute to adult-onset primary open angle glaucoma using linkage approaches.

2001-2005 Genetic Studies of Pseudoexfoliation Glaucoma
NEI/RO1 EY013882
PI
The primary goal of this study was to identify genetic risk factors the pseudoexfoliation syndrome and the related glaucoma.

2005-2010 Genetic Etiologies of Primary Open Angle Glaucoma
NEI/R01 EY015872
PI
This study aimed to identify genetic risk factors for age-related primary open angle glaucoma using case/control association analyses.

2008-2010 Genes and Environment Initiative in Glaucoma
NHGRI/U01HG004728
Co-investigator
The overall goal of this study was to identify genetic risk factors for primary open angle glaucoma as well as investigate gene-environment interactions in primary open angle glaucoma.

2008-2010 LOXL1 and Pseudoexfoliation Glaucoma: Studies in Animal Models
NEI/R21 EY019161
PI
The goal of this study was to develop an animal model for pseudoexfoliation syndrome and the related glaucoma.

2009-2011 India US Genetic Study of Ocular Quantitative Traits
NEI/R21 EY018149
PI
In this study we used large consanguineous pedigrees from South India to map genetic loci for ocular quantitative traits.

2009 NEIGHBOR consortium GWAS for primary open angle glaucoma
NHGRI/X01 HG005259
PI
This grant provided support for genotyping through CDIR for over 6000 glaucoma cases and controls.

2013-2016 Genetic etiologies of congenital glaucoma
March of Dimes foundation ($281,819 total costs)
PI

Current

2012-2019 The NEIGHBORHOOD: POAG Heritable Overall Operational Database
NEI/R01 EY022305 ($714,074 annual total costs)
PI
The overall goal of this project is to perform genome-wide association studies to identify genetic risk factors for primary open angle glaucoma (POAG).

2011-2020 Genetic and Environmental Risk Factors for Exfoliation syndrome and Glaucoma
NEI/R01 EY020928 ($666,517 annual total costs)
PI
The goals of this project are to identify genetic and environmental risk factors for pseudoexfoliation (exfoliation) syndrome and the related glaucoma.

2016-2019 INDO US Study of Ocular Quantitative Traits Related to Glaucoma
NEI/R01 EY027129 ($424,375 annual total costs)

PI
This study aims to identify genetic risk factors that influence susceptibility to forms of glaucoma common in India and in the United States.

2014-2019 P30 Core grant for vision research
NEI/P30EY014104 ($608,957 annual total costs)
PI
This grant provides support for core services for investigators in Ophthalmology at Massachusetts Eye and Ear as well as the Department of Ophthalmology at Harvard Medical School

2004-2018 Gene-environment interactions in glaucoma
NEI/R01 EY015473 ($446,038 annual costs)
Co-investigator
The overall goal of this project is the identify gene-environment interactions in glaucoma as well as modifiable risk factors for glaucoma.

Current Unfunded Projects

Identification of genes responsible for pigmentary glaucoma
PI
I am supervising a fellow who is using whole exome sequencing to identify genes responsible for pigmentary glaucoma, a form of glaucoma that affects young adults.

Identification of novel genetic risk factors for primary optic atrophy
PI
I am supervising a post-doctoral fellow who is using whole exome sequencing and other genomic approaches to identify novel genes responsible for primary optic atrophy.

Report of Local Teaching and Training

Teaching of Students in Courses

1995-2001	Gene 201-202, "Introduction to Genetics"	Tufts Medical School
	Medical students, research fellows	3 one hour lectures/year
2002-2010	Fundamental Issues in Vision Research	National Eye Institute (Wood's Hole course)
	Post-doctoral fellows	2 one hour lecture every other year
2002-	HST 160	HMS
	Medical Students	Preceptor (8 hours/year)
2002-	Molecular Basis of Eye Disease	HMS
	Ophthalmology research fellows, residents	1 one hour lecture/year
2005-	Advanced Human Genetics	HMS
	Medical students, residents	1 one hour lecture/year
2013	HST 220	HMS
	Medical students	Preceptor (8hours/year)

Formal Teaching of Residents, Clinical Fellows and Research Fellows (post-docs)

1992-2005	Glaucoma Lecture Series	Massachusetts Eye and Ear
	Ophthalmology residents and fellows	2 one hour lectures/year
1992-2009	Lancaster course in Ophthalmology	Massachusetts Eye and ear

	Ophthalmology residents and fellows	3 one hour lectures/year
1994-2001	Glaucoma Lecture Series	New England Medical Center
	Ophthalmology residents and fellows	1 one hour lecture/year
1994	Basic Science Course in Ophthalmology	Boston University
	Ophthalmology residents and fellows	1 one hour lecture
1994-1996	Basic Science Course in Ophthalmology	Columbia University
	Ophthalmology residents and fellows	1 one hour lecture/year

Clinical Supervisory and Training Responsibilities

1992-1993	Ambulatory Ophthalmology Clinic Preceptor/Massachusetts Eye and Ear	2 full days/week
1992-1993	Surgical instruction/Massachusetts Eye Ear (Residents and Fellows)	One full day/week
1994-2001	Ambulatory Ophthalmology clinic Preceptor/New England Medical Center	One full day/week
1994-1998	Surgical instruction/New England Medical Center	One full day/week
2001-	Ambulatory Ophthalmology Clinic Preceptor/Massachusetts Eye and Ear Infirmary (Residents and Fellows)	One half day/month

Formally Supervised Trainees

1991-1992	Andrew Fine, MD, MPH/Assistant Professor of Pediatrics, Children's Hospital Boston, Harvard Medical School Published a manuscript on early-onset glaucoma genes.
1994-1995	Carolyn J. Sporn, MD/Emergency Medicine, Holy Cross Hospital, Silver Spring Maryland Published a manuscript on early-onset glaucoma genes.
1995-1999	Jeffrey C. Phillips, DVM,PhD/Assistant Professor of Oncology and Medical Genetics, College of Veterinary Medicine, University of Tennessee Published a manuscript (Am Journal of Human Genetics) on a new genetic locus for Rieger syndrome.
1996-2000	Jessica S. Andersen, PhD/ Freelance Writer Published a manuscript on a new genetic locus for pigmentary glaucoma.
1998-2000	Grant Yanagi, MD/Radiologist, Fayetteville, North Carolina Published two manuscripts as co-author on early-onset glaucoma genes.
1998-2001	Sean P. Lynch, MD/Gastroenterologist, Gastroenterology Professional Association, New Hampshire. Published two manuscripts on early-onset glaucoma genes.
1998-1999	Simone Finzi, MD, PhD/Assistant Professor of Ophthalmology, University of San Paulo, Brazil. Published a manuscript characterizing a chromosome deletion associated with congenital glaucoma.
2001-2002	Mary Lillian Tocyap, MD/Pediatrician, Marana, Arizona. Published a manuscript describing chromosomal deletions affecting corneal pathology related to glaucoma.
2001-2004	Dayse Figueiredo Sena, MD/Ora investigator, Andover, Massachusetts

	Published a manuscript on founder mutations in *CYP1B1* (J of Med Genet) and 3 additional manuscripts as co-author.
2004-2006	Maria Isabel Triana, MD/Ophthalmologist, San Antonio, Texas Published a manuscript as a co-author on risk factors for glaucoma.
2005-2006	Relief Jones, MD/Assistant Professor, Ophthalmology, University of Texas Health Science Center at San Antonio Published a manuscript as a co-author on risk factors for glaucoma.
2006-2008	Ambika Sud, MD/Glaucoma Fellow, University of Miami, Florida Published a manuscript on the fine mapping of a genetic locus for early-onset glaucoma and obtained fellowship funding from HHMI.
2007-2008	Hana Takusagawa, MD/Assistant Professor, Ophthalmology, Oregon Health Sciences University, Portland, Oregon Published a manuscript on risk factors for glaucoma.
2007-2011	Baojian Fan, MD, PhD/Instructor, Ophthalmology, Harvard Medical School Published 6 manuscripts as first author and 4 manuscripts as a co-author on genetic risk factors for common complex forms of glaucoma.
2010-2013	Danyi Wang, MD, PhD/Senior Technology, Genetic Diagnostic Laboratory, Mass Eye and Ear. Published 3 manuscripts as a co-author on genetic risk factors for glaucoma.
2011-2012	Xi Chen, MD,PhD/Resident in Ophthalmology, Massachusetts Eye and Ear Infirmary, Harvard Medical School Screened candidate genes for early-onset glaucoma.
2012-2015	Keri Allen, MD/Senior research fellow, Massachusetts Eye and Ear Infirmary, Harvard Medical School Exome sequencing and other genomic approaches to identify novel genes contributing to congenital glaucoma.
2013-2014	Ryan Wang/Medical Student, Tulane University Medical School Genomic analysis of patients with ocular developmental defects.
2013-2016	Isao Nakata, PhD/Senior research fellow, Massachusetts Eye and Ear Infirmary, Harvard Medical School Whole exome sequencing to identify novel genes causing primary optic atrophy.
2013-2015	Shirley Chen, MD/Senior research fellow, Massachusetts Eye and Ear Infirmary, Harvard Medical School Exome sequencing and other genomic approaches to identify novel genes contributing to early-onset glaucoma.
2014-2015	Alice Choi/Undergraduate Student, MIT Evaluation of candidate genes responsible for early onset glaucoma.
2016-	Shisong Rong MD PhD/Senior research fellow, Massachusetts Eye and Ear Infirmary, Harvard Medical School Whole genome sequencing of POAG loci
2016-	Ryan Collantes MD/Senior research fellow, Massachusetts Eye and Ear Infirmary, Harvard Medical School Whole exome sequencing of early-onset glaucoma families
2016-2018	Angela Gauthier/Medical Student, Yale Functional studies of early-onset glaucoma candidate genes
2018-	Paulo Svicki PhD/Senior research fellow, Massachusetts Eye and Ear Infirmary, Harvard Medical School Functional studies of early-onset glaucoma candidate genes

2018- Shiming Li, MD/Senior research fellow, Massachusetts Eye and Ear Infirmary, Harvard Medical School
Whole genome sequencing of POAG loci

2018- Youjin (Rachel) Oh/Medical Student, Queen's University Canada
Functional studies of early-onset glaucoma candidate genes

Formal Teaching of Peers (e.g., CME and other continuing education courses)

No presentations below were sponsored by outside entities.

2009-2011	Genetics of Macular Degeneration HMS CME	On-line
2013	Genetics and Genomics of Ocular Disease HMS CME	Course director

Local Invited Presentations

No presentations were sponsored by outside entities.

1995 Genetics Clinic Conference/Invited speaker
Massachusetts General Hospital

1996 New England Medical Center Celebration and Medical Symposium/Invited Professor
New England Medical Center

1996 Broadhurst Distinguished Lecture Series/Invited speaker
Schepens Eye Research Institute

2005 Update on Ophthalmology/Invited speaker
Department of Ophthalmology, HMS

2006 Schepens Eye Institute seminar series/Invited speaker

2007 Massachusetts Eye and Ear Infirmary/Grand rounds

2013 Genetic risk factors for glaucoma/Invited Faculty Speaker
Annual meeting, Mass Eye and Ear

2015 Mariana Mead Lecture, Mass Eye and Ear Annual meeting

Report of Regional, National and International Invited Teaching and Presentations

Invited Presentations and Courses

No presentations were sponsored by outside entities unless otherwise noted.

Regional

1994 Glaucoma Genes/Visiting Professor
Boston University School of Medicine, Boston MA

1995 Genetics Clinic Conference/Invited speaker
Massachusetts General Hospital
Berkshire Medical Center, University of Massachusetts.

1996 Glaucoma Genetics/Visiting Professor
Department of Ophthalmology, Yale University, New Haven CT

1996 Glaucoma Genetics/Visiting Professor
Jackson Laboratory Seminar Program, Bar Harbor ME

1999 Advances in glaucoma genetics/Invited symposium speaker
Massachusetts Society of Eye Physician and Surgeons

2003 Genetic Risk Factors for Glaucoma/Symposium speaker
New England Ophthalmological Society
2003 Three lessons learned from family studies/Invited speaker
Chandler Grant Society
2006 Genetics of Glaucoma/Invited speaker
Genetics Grand Rounds Tufts University School of Medicine
2011 Genetic Etiologies of Glaucoma/Invited speaker
New England Regional Genetics Group Annual Meeting, Portsmouth, New Hampshire
2016 Glaucoma Genetics: From the Lab to the Clinic/Morton Grant Lecture/Tufts University Department of Ophthalmology, Boston, Massachusetts
2017 Glaucoma Genetics: Bringing the Lab to the Clinic/Chandler Grant Lecturer/New England Ophthalmological Society/Boston Massachusetts

National

1994 Glaucoma Genetics/Invited Lecturer
New York Society of Clinical Ophthalmology, NY,NY
1994 Genetics of Glaucoma/Visiting Professor
University of Wisconsin-Madison
1995 Genetics and Glaucoma/Invited Lecturer
Glaucoma Research Foundation, San Francisco CA
1996 Genetics and Glaucoma/Visiting Professor
New York Hospital-Cornell Medical Center, NY, NY
1996 Genes and Glaucoma/Visiting Professor
Washington University, St. Louis, MO
1996 Advances in Glaucoma Genetics/Symposium speaker
American Academy of Ophthalmology, Chicago, Illinois
1996 Genetics and Glaucoma/Invited symposium speaker
Glaucoma Research Foundation, Chicago, Illinois
1997 Genetic studies in glaucoma/Visiting Professor
Chairman's Special Lecture Series, Duke Eye Center, Durham, NC
1997 Genetics of secondary glaucomas/Invited speaker
American Academy of Ophthalmology Subspecialty Day, San Francisco CA
1997 Glaucoma Genetics/Invited symposium speaker
New Jersey Academy of Ophthalmology, Eatontown, NJ
1997 Genetics of Glaucoma/Invited lecturer
American Glaucoma Society, Scottsdale AZ
1997 Genetic linkage studies in glaucoma/Invited speaker
National Science Writers Seminar Series, Research to Prevent Blindness
1998 Genetics and Glaucoma/Invited speaker
Milwaukee Ophthalmological Society, Milwaukee WI
1998 Update on glaucoma genetics/Visiting Professor
Cleveland Clinic Foundation, Cleveland OH
1998 Update on Glaucoma Genetics/Invited symposium speaker
American Academy of Ophthalmology, Subspecialty Day, New Orleans, LA
2000 Gene therapy for glaucoma/Invited symposium speaker
American Academy of Ophthalmology, Dallas, Texas
2000 Genetics of glaucoma/Invited symposium speaker
International Congress of Eye Research, Santa Fe, New Mexico

2001 Glaucoma: Genes and phenotypes/Invited symposium speaker
American Academy of Ophthalmology, New Orleans, LA

2002 Molecular genetics of glaucoma/Invited speaker
Pittsburgh Ophthalmological Society, Pittsburgh, Pennsylvania

2002 Glaucoma genetics: new genes/Invited speaker
American Academy of Ophthalmology, Subspecialty Day (Glaucoma), Orlando FL

2003 Glaucoma Genetics 2003: Vision for the Future/Invited symposium speaker
Cleveland Clinic, Cleveland Ohio

2004 Update Glaucoma genetics/Invited symposium speaker
American Society of Human Genetics, Toronto, Canada

2005 Genetics: Needs and Opportunities/Invited speaker and thought leader
US-INDO Workshop, National Eye Institute, Hyderabad, India

2005 Genetics in Glaucoma/Grand rounds speaker
Ophthalmology, University of Illinois, Chicago, Illinois

2005 Genetic studies in glaucoma/Invited symposium speaker
The Great Lakes Abrahamson Pediatric Eye Institute Vision Research Conference,
Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio

2007 Glaucoma genes and phenotypes/Invited speaker
eyeGENE Lecture Series, National Eye Institute, Bethesda Maryland

2007 Glaucoma: genes, phenotypes and future research/Grand rounds speaker
Ophthalmology, University of California, San Francisco

2007 Glaucoma genetics/Invited symposium speaker
Annual Alumni symposium, Ophthalmology, University of Pennsylvania

2007 Glaucoma genes and phenotypes/Grand rounds speaker
Visual Science Program, Columbia University, NY

2008 Glaucoma: Genes, phenotypes and new directions for therapy/Rich Lecturer
Ophthalmology, University of Alabama

2009 Glaucoma Genetics: Genes, phenotypes and new approaches/Invited symposium speaker
ARVO/Pfizer Ophthalmic Research Institute, Ft. Lauderdale, FL (Pfizer)

2009 Personalized Medicine for Ophthalmology/Invited symposium speaker
American Academy of Ophthalmology Subspecialty day (Retina), San Francisco, CA

2009 Glaucoma Genetics/Invited speaker
Gene-based diagnostic testing for glaucoma/Invited speaker
ARVO symposium on Genetics, American Academy of Optometry, Orlando, FL

2010 Human Glaucoma: Genes and genetic approaches/Invited symposium speaker
American Glaucoma Society, Los Angeles, CA

2010 NEIGHBOR study: Rational, study design and clinical variables/Invited symposium speaker
Association for Research in Vision and Ophthalmology (ARVO), Ft. Lauderdale, FL

2010 The who, what, where and why of genetic testing for glaucoma/Invited symposium speaker
American Academy of Ophthalmology, Chicago, IL

2010 Personalized medicine for ophthalmology/Invited symposium speaker
Ophthalmology, Vanderbilt University, Nashville, TN

2011 NEIGHBOR/GLAUGEN glaucoma GWAS/Invited symposium speaker
Association for Research in Vision and Ophthalmology (ARVO), Ft. Lauderdale, FL

2011 NEIGHBOR/GLAUGEN POAG GWAS/Invited speaker
eyeGENE biannual conference, National Eye Institute, Bethesda, Maryland

2011 NEIGHBOR/GLAUGEN POAG GWAS/Invited speaker

Yale Glaucoma Conference, New Haven, CT

2011 Update on Glaucoma Genetics/Invited speaker
Hawaiian Eye, Maui, Hawaii

2012 Toward the identification of genetic risk factors for glaucoma/Grand Rounds
University of Tennessee, Memphis, TN

2012 Genetic Testing for Glaucoma/Invited speaker
Clues to Neuroprotection from genetics/Invited speaker
Hawaiian Eye, Maui, Hawaii

2013 What genetic studies are teaching us about glaucoma/Invited speaker
Current genetic testing for glaucoma/Invited speaker
Hawaiian Eye, Hawaii, Hawaii

2013 Glaucoma genetics: new insights into a complex disease/Grand Rounds
Ophthalmology, Columbia University, NY, NY

2013 Vision BioBank/Invited speaker (presented winning entry)
Audacious Goals Meeting, National Eye Institute, Bethesda, MD

2013 Genetics in the Glaucoma Clinic/Grand Round
Ophthalmology, Indiana University Medical Center, Indianapolis, IA

2013 Genetic risk factors for Glaucoma/Plenary speaker
Fight for Sight Annual Meeting, NY, NY

2013 Glaucoma genetics: genetic testing and new approaches to therapy
Midwest Glaucoma Symposium/Pittsburgh PA

2013 Genetic Testing for glaucoma/Invited symposium speaker
American Academy of Ophthalmology Subspecialty Day

2013 Great Debate: genetic testing for glaucoma/Invited speaker
American Academy of Ophthalmology

2013 Genetics in the Glaucoma Clinic: Genetic Testing and New Ideas for Treatment/Kenneth Swan Lecturer, Devers Eye Clinic, Oregon Health Sciences University

2014 Glaucoma Genetics: Families and NEIGHBORs/Clinician Scientist Lecturer
American Glaucoma Society, Washington DC

2014 Genome-wide Association Studies in Glaucoma/Invited symposium speaker, Association for Research in Vision and Ophthalmology (ARVO), Orlando, FL

2014 Genetic Risk Factors for Glaucoma, Genetics in the Glaucoma clinic/Invited professor/Case Western Reserve Vision Research Symposium, Cleveland Ohio

2014 NEIGHBOR GWAS for glaucoma/Invited Faculty Member, Summer Research Institute, American Optometry Association, Ohio State University, Columbus, Ohio

2014 Genome-wide association studies in glaucoma/Invited symposium speaker, International Society of Eye Research (ISER), San Francisco, CA

2015 Genetic and epidemiologic risk factors for normal tension glaucoma (vs high tension glaucoma)/Invited symposium speaker, American Glaucoma Society, San Diego, CA

2015 Primary Open Angle Glaucoma Genes and New Insights into Disease Pathogenesis/Invited speaker, Alcon Research Symposium, Boston, MA

2015 Glaucoma genetics: What we know and what we need to know/Leopold Lecture, Mt. Sinai School of Medicine, NYC, NY

2015 Glaucoma genetics: Insights into a complex disease/Institutional Research Seminar, SUNY Downstate School of Medicine, Brooklyn, NY

2016 Genetics of exfoliation syndrome and glaucoma/Invited Symposium speaker, American Glaucoma Society, Ft. Lauderdale FL

2016 Mendelian Ocular Disease/Keynote speaker, Association for Vision and Research in

Ophthalmology (ARVO), Seattle, WA

2016 Glaucoma Genetics: From the Lab to the Clinic/Keynote Speaker/Gained in Translation Symposium/ University of Washington Department of Ophthalmology, Seattle, Washington

2016 Glaucoma Genetics: New insights into a complex disease/Invited speaker/Department Seminar/Molecular and Human Genetics/Baylor School of Medicine/Houston TX

2016 Precision Medicine and the Glaucoma Patient/Invited Speaker/New York Glaucoma Society/New York, New York

2016 Glaucoma Genetics: New insights into a complex disease/Invited Seminar Speaker/Vision Discovery Institute/Georgia Regents University/Augusta Georgia

2017 Putting together collaborative groups/Symposium speaker/Association of University Professors in Ophthalmology/San Diego California

2017 Using EMRs for Clinical Research: Opportunities and Challenges/Symposium speaker/Association of University Professors in Ophthalmology/San Diego California

2017 US INDO collaborations and Genetic Eye Disease/Symposium speaker/Association for Research in Vision and Ophthalmology/Baltimore, Maryland

2017 Primary open angle glaucoma: Insights into a complex disease/Phelps Lecture/ University of Iowa School of Medicine Department of Ophthalmology/Iowa City Iowa

2017 Primary open angle glaucoma: Insights into a complex disease/International Society of Eye Research Glaucoma Symposium/Keynote speaker/Atlanta Georgia

2017 Gene therapy for glaucoma/Glaucoma subspecialty day invited speaker/ American Academy of Ophthalmology, New Orleans, LA

2017 Glaucoma Genes and New Opportunities for Therapy /Robert N. Shaffer Lecture/American Academy of Ophthalmology, New Orleans, LA

2018 When it's not glaucoma/Symposium speaker/Hawaiian Eye/ Maui Hawaii

2018 Research Collaborations: Benefits and Effective Strategies/Invited speaker/American Glaucoma Society/New York, New York

2018 Glaucoma genes and new opportunities for therapy; Genetic Risk factors for exfoliations syndrome and glaucoma; Research collaborations: Benefits and Effective Strategies/Thorny Issues featured speaker/Devers Eye Institute/Portland Oregon

2018 Genetic and environmental risk factors for exfoliation syndrome/Glaucoma subspecialty day invited speaker/American Academy of Ophthalmology, Chicago, Illinois

2019 Drs. Henry and Frederick Sutro Memorial Lecture/Glaucoma 360 New Horizons Forum/San Francisco CA

International

1995 Genetic studies in glaucoma/APOS lecturer
Annual meeting of the Atlantic Provinces Ophthalmological Society (Held in Jamaica)

1996 Genetic studies in glaucoma/Visiting Professor
University Erlangen-Nürnberg, Erlanger, Germany

1996 Genetics of Glaucoma/Invited speaker
International Glaucoma Society, Wurzburg, Germany

1997 Genetic etiologies of glaucoma/Invited speaker
International Symposium on Experimental and Clinical Ocular Pharmacology and Pharmaceutics, Munich Germany

1997 Glaucoma genetics/Visiting Professor
Ophthalmology Course, Tel Aviv, Israel

1998 Genetics and Glaucoma/Invited symposium speaker

International Congress of Ophthalmology, Amsterdam, The Netherlands

1998 Molecular Genetics of Glaucoma/Symposium speaker
International Congress of Eye Research, Paris, France

1998 Genetics and Molecular Biology of Glaucoma/Invited symposium speaker
Glaucoma Research Foundation, San Juan, Puerto Rico

1999 Genes and Glaucoma/Invited symposium speaker
Glaucoma in the Next Millennium, Rome, Italy (Merck)

2000 Molecular genetics of glaucoma/Invited symposium speaker
International Ophthalmology Symposium, Oxford England

2001 Genetic Etiologies of Glaucoma/Invited symposium speaker
International Symposium on Glaucoma Research, Prague, Czechoslovakia

2008 Macular Degeneration: Update on genetics/Invited symposium speaker
World Ophthalmology Congress, Hong Kong, China

2008 Genetic approaches to glaucoma/Invited symposium speaker
International Society of Eye Research, Beijing, China

2009 Glaucoma genetics:/Invited symposium speaker
Asia ARVO, Hyderabad, India

2009 Pseudoexfoliation Genetics/Invited symposium speaker
World Glaucoma Congress, Boston, MA

2010 Glaucoma Genetics: Past, Present and Future/Champaulimaud Lecturer
LV Prasad Eye Institute, Hyderabad, India

2010 NEIGHBOR study: Rationale, study design and clinical variables/Invited symposium speaker
International Society of Eye Research, Montreal Canada

2010 NEIGHBOR study: Rationale, study design and clinical variables/Invited symposium speaker
Asia-Pacific Academy of Ophthalmology, Beijing, China

2011 NEIGHBOR GWAS for Glaucoma/Invited speaker
Sankara Nethralaya Vision Research, Chennai, India

2011 NEIGHBOR study: Rationale, study design and clinical variables/Invited symposium speaker
Asia ARVO, Singapore

2011 Clues to neuroprotection from genetics studies/Invited symposium speaker
Glaucoma GWAS/Invited symposium speaker
Genetic and environmental risk factors for pseudoexfoliation glaucoma/Invited symposium speaker
World Glaucoma Congress, Paris, France

2011 Genetic risk factors for optic nerve disease in glaucoma/Invited symposium speaker
Optic Nerve Symposium (sponsored by ARVO), Obergurl, Austria

2012 Genetic risk factors for glaucoma: results from the GLAUGEN and NEIGHBOR POAG GWAS/Invited symposium speaker
Asian-Pacific Academy of Ophthalmology, Busan, Korea

2013 Genetic Risk Factors for Primary Open Angle Glaucoma/Invited symposium speaker
Asian-Pacific Academy of Ophthalmology, Busan Korea

2013 Glaucoma: genetics perspective/Invited symposium speaker
Moorfields Eye Hospital Glaucoma Symposium, London, England (Merck)

2013 Primary Glaucoma Heritability: should we bother with family history?/Invited speaker
From the laboratory to the clinic/Invited speaker

Understanding the genetic basis of glaucoma/Organizer and speaker
Introduction to the International Glaucoma Genetics Consortium/Invited speaker
World Glaucoma Congress, Vancouver, Canada

2013 NEIGHBOR GWAS for Glaucoma/Organizer and speaker
GWAS results support a role for cerebrospinal fluid pressure in glaucoma/Organizer and speaker Asia ARVO, Delhi, India

2014 Genetic testing in Ophthalmology, Genetic research in early-onset glaucoma, Genetics in the Glaucoma Clinic/Invited speaker/Canadian Ophthalmological Society/Halifax Nova Scotia

2014 Genome-wide association studies in Glaucoma/Invited symposium speaker/Asian Pacific Glaucoma Society/Hong Kong

2015 Drug-induced secondary angle closure: risk factors/Asia-ARVO/Yokohama, Japan

2015 Recent advances in the genetics of POAG/Asia-Pacific Academy of Ophthalmology/Guangzhou, China

2015 Genetics of POAG/ Asia-Pacific Academy of Ophthalmology/Guangzhou, China

2015 Genetic testing in anterior segment dysgenesis/World Glaucoma Congress/ Hong Kong

2015 Collaborative phenotyping networks/World Glaucoma Congress/Hong Kong

2016 Genetic Testing for Glaucoma/Invited symposium speaker/World Ophthalmology Congress/Guadalajara, Mexico

2016 Genome-wide association studies for POAG/symposium speaker/Asia-Pacific Academy of Ophthalmology/Taipei, Taiwan

2016 Gene-environment interactions in Exfoliation syndrome/symposium speaker/Asia-Pacific Academy of Ophthalmology/Taipei, Taiwan

2016 Primary open angle glaucoma genetics: Insights into a complex disease/Invited speaker/Glaucoma Research Society/Seoul, Korea

2016 Research programs at the Harvard Department of Ophthalmology/Invited Speaker/Chinese Ophthalmological Society and Global Symposium/Suzhou, China

2016 Genome-wide association studies in Normal tension glaucoma/International Society of Eye Research/Symposium organizer and speaker/Tokyo, Japan

2017 Association of ANGPT1 with POAG/Translational Summit Speaker/Asia ARVO/Brisbane Australia

2017 Early-onset glaucoma genetics/Symposium speaker/Asian Pacific Academy of Ophthalmology/Singapore

2017 Primary open angle glaucoma: New insights into disease mechanisms and translational opportunities/Symposium speaker/Asian Pacific Academy of Ophthalmology/Singapore

2017 Genotype/phenotype correlations in Glaucoma/Symposium speaker/World Glaucoma Association/Helsinki Finland

2017 Genetics of Early-onset Glaucoma/Invited symposium speaker/World Congress of Pediatric Ophthalmology/Hyderabad India

2018 Phenome-wide association studies (PheWAS)/Asian Pacific Academy of Ophthalmology/Hong Kong

2018 Genetic testing for glaucoma/Asian Pacific Academy of Ophthalmology/Hong Kong

2018 Within 20 years Precision medicine will revolutionize glaucoma diagnosis and management (debate topic)/Asian Pacific Academy of Ophthalmology/Hong Kong

2018 The Role of Genomics and Family History in glaucoma/Invited speaker/World Ophthalmology Congress/Barcelona Spain

2018 Genetics of Adult Glaucoma/World Ophthalmology Congress/Barcelona Spain

2018 Updates on Primary Open Angle Glaucoma Genetics/Invited speaker/Glaucoma Research

	Society/Parma Italy
2018	Advances in Childhood Glaucoma Genetics/Invited speaker/International society for Eye Research (ISER)/Belfast Ireland
2018	Primary Open Angle Glaucoma Genetics: Insights into a complex disease/Invited symposium speaker/Philippine Academy of Ophthalmology/Manila Philippines

Report of Clinical Activities and Innovations

Current Licensure and Certification

1986	Massachusetts Medical License
1991	Board certified, American Board of Ophthalmology
1999	Board certified, American Board of Medical Genetics (Molecular Genetics)

Practice Activities

1992-1993	Ambulatory Care	Glaucoma Service Mass Eye and Ear	2 days/week
1992-1993	Surgery	Mass Eye and Ear	1 day/week
1994-2001	Ambulatory Care	Glaucoma Service New England Medical Center	1 day/week
1994-1998	Surgery	New England Medical Center	1 day/week
2001-	Ambulatory Care	Glaucoma Service Mass Eye and Ear	One half day/month
2005-	Clinical Laboratory	Director CLIA-certified Genetic Diagnostic Lab Mass Eye and Ear	1 day/week

Clinical Innovations

In collaboration with Eric Pierce MD, PhD, I have developed a gene panel test for diagnosis of inherited disease in patients with glaucoma, optic neuropathy and retinal degenerations. This single test makes it possible to identify mutation carriers and appropriate family members at risk for blinding eye disorders. This information has a significant impact on the diagnosis, prognosis and genetic counseling of affected individuals and family members.

Report of Education of Patients and Service to the Community

Activities

2010	Genetics of Glaucoma/speaker Glaucoma Foundation, Boston chapter
2010	Glaucoma, genes and India/speaker HMS Alumni Day Symposium
2011	Diseases of the Optic Nerve/invited speaker Perkins School for the Blind, Watertown, MA

Report of Scholarship

Publications

Peer reviewed publications in print or other media

Research Investigations:

1. Gonzales N, **Wiggs J**, and Chamberlin MJ. (1977) A simple procedure for resolution of Escherichia Coli RNA polymerase holoenzyme from core polymerase. Arch. Biochem. Biophys. 182: 404-408.

2. **Wiggs JL**, Bush JW, and Chamberlin MJ (1979) Utilization of promoter and terminator sites on bacteriophage T7 DNA by RNA polymerases from a variety of bacterial orders. Cell 16:97-109.

3. Jaehning JA, **Wiggs JL**, and Chamberlin MJ. (1979) Altered promoter selection by a novel form of Bacillus subtilis RNA polymerase. Proc. Nat. Acad. Sci. USA 76:5470-5474.

4. Chamberlin MJ, Nierman WC, **Wiggs JL**, Neff N. (1979) A quantitative assay for bacterial RNA polymerases. J. Biol. Chem. 254:10061-10069.

5. Ballou L, Grove RJ, Roon RH, **Wiggs J**, and Ballou CE. (1981) Temperature-Sensitive Glucosamine Auxotrophy of Saccharomyces cerevisiae. Molecular and Cellular Biology 1:9-12.

6. **Wiggs JL**, Gilman MZ and Chamberlin MJ. (1981) Heterogeneity of RNA polymerase in Bacillus subtilis: evidence for an additional sigma factor in vegetative cells. Proc. Nat. Acad. Sci. USA 78:2762-2766.

7. Gilman MZ, **Wiggs JL**, and Chamberlin MJ. (1981) Nucleotide sequences of two Bacillus subtilis promoters used by Bacillus subtilis sigma-28 RNA polymerase. Nucleic Acids Research 9:5991-6000.

8. Chamberlin M, Kingston R, Gilman M, **Wiggs JL**, and deVera A. (1983) Isolation of bacterial and bacteriophage RNA polymerases and their use in synthesis of RNA in vitro. Methods in Enzymology. 101:540-568.

9. **Wiggs J**, Nordenskjold M, Yandell D, Rapaport J, Grondin V, Janson M, Werelius B, Petersen R, Craft A, Riedel K, Liberfarb R, Walton D, Wilson W, and Dryja T. (1988). Prediction of the risk of hereditary retinoblastoma, using DNA polymorphisms within the retinoblastoma gene. New England Journal of Medicine 318:151-157.

10. **Wiggs JL** and Dryja TP (1988) Predicting the risk of hereditary retinoblastoma. American Journal of Ophthalmology 106:346-351.

11. Shiang R, Murray JC, **Wiggs J**, Dryja T. (1988) A TaqI RFLP identified at the retinoblastoma locus on chromosome 13. Nucleic Acids Research 16:9069.

12. Reichel E, **Wiggs JL**, Mukai S, D'Amico D. (1992) Oxycephaly, Bilateral Ectopia Lentis and Retinal Detachment. Annals of Ophthalmology 24:97-98.

13. **Wiggs JL**, Haines J, Paglinauan CM, Fine A, Sporn C, Lou D. (1994) Genetic linkage of autosomal dominant juvenile glaucoma to 1q21-q31 in three affected pedigrees. Genomics

21:299-303.

14. Paglinauan C, Haines JL, DelBono EA, Schuman J, Stawski S, **Wiggs JL**. (1995) Exclusion of chromosome 1q21-q31 from linkage to three pedigrees affected by the pigment dispersion syndrome. Am J Hum Genet 56:1240-1243.

15. **Wiggs JL**, DelBono EA, Schuman JS, Hutchinson BT, Walton DS. (1995) Clinical features of five pedigrees genetically linked to the juvenile glaucoma locus on chromosome 1q21-q31. Ophthalmology 102:1782-1789.

16. **Wiggs JL**, Damji KF, Haines JL, Pericak-Vance MA, Allingham RR. (1996) The distinction between juvenile and adult-onset primary open-angle glaucoma. Am J Hum Genet 58:243-244.

17. Phillips JC, DelBono EA, Haines JL, Pralea AM, Cohen JS, Greff LJ, **Wiggs JL**. (1996) A second locus for Rieger syndrome maps to chromosome 13q14. Am J Hum Genet 59:613-619.

18. Andersen JS, Pralea AM, DelBono EA, Haines JL, Gorin MB, Schuman JS, Mattox CG, **Wiggs JL**. (1997) A gene responsible for the pigment dispersion syndrome maps to chromosome 7q35-36. Archives of Ophthalmology 115:384-388.

19. Chang JT, Milligan S, Li Y, Chew CE, **Wiggs J**, Copeland NG, Jenkins NA, Campochiaro PA, Hyde DR, Zack DJ. (1997) Mammalian homolog of Drosophila retinal degeneration B rescues the mutant fly phenotype. J Neurosci 17:5881-5890.

20. Allingham RR, **Wiggs JL**, DeLaPaz MA, Vollrath D, Tallett DA, Broomer B, Jones KH, DelBono EA, Kern J, Patterson K, Haines JL, Pericak-Vance MA. (1998) Gln368STOP myocilin mutation in families with late-onset primary open-angle glaucoma. Invest Ophthalmol Vis Sci 39:2288-2295.

21. Allingham RR, **Wiggs JL**, Damji KF, Herndon L, Youn J, Tallett DA, Jones KH, DelBono EA, Reardon M, Haines JL, Pericak-Vance MA. (1998) Adult-onset primary open angle glaucoma does not localize to chromosome 2cen-q13 in North American families. Hum Hered 48:251-255.

22. **Wiggs JL**, Allingham RR, Vollrath D, Jones KH, DeLaPaz M, Kern J, Patterson K, Babb VL, DelBono EA, Broomer BW, Pericak-Vance MA, Haines JL. (1998) Prevalence of mutations in TIGR/Myocilin in patients with adult and juvenile primary open angle glaucoma. Am J Hum Genet 63:1549-1552.

23. Pacella R, McLellan J, Grice K, DelBono EA, **Wiggs JL**, Gwiazda J.(1999) Role of genetic factors in the etiology of juvenile-onset myopia based on a longitudinal study of refractive error. Optometry and Vision Science 76:381-386.

24. **Wiggs JL**, Allingham RR, Hossain A, Kern J, Auguste J, DelBono EA, Broomer B, Graham FL, Hauser M, Pericak-Vance M, Haines JL. (2000) Genome-wide scan for adult onset primary open angle glaucoma. Hum Mol Genet. 9:1109-1117.

25. Finzi S, Pinto CF, **Wiggs JL**. (2001) Molecular and clinical characterization of a patient with a chromosome 4p deletion, Wolf-Hirschhorn syndrome, and congenital glaucoma. Ophthalmic

Genetics 22:1710-1711.

26. **Wiggs JL**, Vollrath D. (2001) Molecular and clinical evaluation of a patient hemizygous for TIGR/MYOC. Arch Ophthalmol 119:1674-1678.

27. Andersen MG, Smith RS, Hawes NL, Zabaleta A, Chang B, **Wiggs JL**, John SW. (2001) Mutations in genes encoding melanosomal proteins cause pigmentary glaucoma in DBA/2J mice. Nature Genetics 30:81-85.

28. Lynch S, Yanagi G, DelBono E, **Wiggs JL**. (2002) DNA sequence variants in the tyrosinase-related protein 1 (TYRP1) gene are not associated with human pigmentary glaucoma. Molecular Vision 8:127-129.

29. **Wiggs JL**, Allingham, RR, Pericak-Vance M, Hauser M, Auguste J, Rodgers K, Broomer B, Del Bono E, Haines JL. (2003) Lack of association of mutations in optineurin with disease in patients with adult-onset primary open-angle glaucoma. Arch Ophthalmol. 121:1181-3.

30. Sena DF, Rogers K, DelBono E, **Wiggs JL**. (2004) CYP1B1 founder mutations in congenital glaucoma patients from the U.S. and Brazil. Journal of Medical Genetics 41:23-27.

31. **Wiggs JL**, Maselli M, Lynch S, Yanagi G, DelBono E, Haines JL. (2004) A genome-wide scan identifies novel early onset primary open angle glaucoma loci on 9q22 and 20p12. Am J Hum Genet 74:1314-1320.

32. Allingham RR, **Wiggs JL**, Hauser EA, Larocque-Abramson KR, Santiago-Turla C, Broomer B, Del Bono EA, Graham FL, Haines JL, Hauser MA, Pericak-Vance MA. (2005) Early adult-onset POAG linked to 15q11-13 using ordered subsets analysis. Invest Ophthalmol Vis Sci 46: 2002-2005.

33. Tocyap ML, Azar M, Chen T, **Wiggs JL**. (2006) Clinical and molecular characterization of a patient with an interstitial deletion of chromosome 12q15-q23 and peripheral corneal abnormalities. Am J Ophthalmol 141(3):566-567.

34. Hauser MA, Allingham RR , Linkroum K, Wang J, LaRocque-Abramson K, Figueiredo D, Santiago-Turla C, Del Bono EA, Haines JL, Pericak-Vance MA, **Wiggs JL**. (2006) Distribution of WDR36 DNA sequence variants in primary open angle glaucoma patients. Invest Ophthal Vis Sci 47(6):2542-6.

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36. Hewitt AW, Samples JR, Allingham RR, Jarvela I, Kitsos G, Krishnadas SR, Richards JE, Lichter PR, Petersen MB, Sundaresan P, **Wiggs JL**, Mackey DA, Wirtz MK. (2007) Investigation of founder effects for the Thr377Met Myocilin mutation in glaucoma families from differing ethnic backgrounds. Mol Vis. 13:487-92.

37. Liu Y, Schmidt S, Qin X, Gibson J, Munro D, **Wiggs JL**, Hauser MA, Allingham RR. (2007) No association between OPA1 polymorphisms and primary open-angle glaucoma in three different populations. Mol Vis. 13:2137-41.

38. Fan BJ, Pasquale L, Grosskreutz CL, Rhee D, Chen T, Deangelis MM, Kim I, Delbono E, Miller JW, Li T, Haines JL, **Wiggs JL.** (2008) DNA sequence variants in the LOXL1 gene are associated with pseudoexfoliation glaucoma in a U.S. clinic-based population with broad ethnic diversity. BMC Med Genet. 6;9(1):5.

39. Liu Y, Schmidt S, Qin X, Gibson J, Hutchins K, Santiago-Turla C, **Wiggs JL**, Budenz DL, Akafo S, Challa P, Herndon LW, Hauser MA, Allingham RR. (2008) Lack of association between LOXL1 variants and primary open-angle glaucoma in three different populations. Invest Ophthalmol Vis Sci. 49(8):3465-3468.

40. Sud A, Del Bono EA, Haines JL, **Wiggs JL**. (2008) Fine mapping of the GLC1K juvenile primary open-angle glaucoma locus and exclusion of candidate genes. Mol Vis. 21;14:1319-1326.

41. Fan BJ, Chen T, Grosskreutz C, Pasquale L, Rhee D, DelBono E, Haines JL, **Wiggs JL**. (2008) Lack of Association of Polymorphisms in Homocysteine Metabolism Genes with Pseudoexfoliation Syndrome and Glaucoma. Mol Vis. 14:2484-91.

42. Kang JH*, **Wiggs JL***, Rosner BA, Hankinson SE, Abdrabou W, Fan BJ, Haines JL, Pasquale LR (2010) The Relation between Endothelial Nitric Oxide Synthase Gene Variants and Primary Open-Angle Glaucoma: Interactions with Gender and Postmenopausal Hormone Use. Invest Ophthalmol Vis Sci Feb;51(2):971-9. *authors contributed equally.

43. Fan BJ, Figuieredo DR, Pasquale LR, Grosskreutz CL, Rhee DJ, Chen TC, DelBono EA, Haines, JL, **Wiggs JL.** (2010) Lack of Association of Polymorphisms in Elastin with Pseudoexfoliation Syndrome and Glaucoma. J Glaucoma. Sep;19(7):432-6.

44. Cornelis MC, Agrawal A, Cole JW, Hansel NN, Barnes KC, Beaty TH, Bennett SN, Bierut LJ, Boerwinkle E, Doheny KF, Feenstra B, Feingold E, Fornage M, Haiman CA, Harris EL, Hayes MG, Heit JA, Hu FB, Kang JH, Laurie CC, Ling H, Manolio TA, Marazita ML, Mathias RA, Mirel DB, Paschall J, Pasquale LR, Pugh EW, Rice JP, Udren J, van Dam RM, Wang X, **Wiggs JL**, Williams K, Yu K for the GENEVA Consortium. (2010) The Gene, Environment Association Studies Consortium (GENEVA): Maximizing the knowledge obtained from GWAS by collaboration across studies of multiple conditions. Genet Epidemiol. 2010 May;34(4):364-72.

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Pasquale LR, Olivier M, Haines JL, Fan BJ, **Wiggs JL** (2010) Distribution of COL8A2 and COL8A1 gene variants in Caucasian primary open angle glaucoma patients with thin central corneal thickness. Mol Vis. Oct 29;16:2185-91.

48. Fan BJ, Pasquale LR, Rhee D, Li T, Haines JL, **Wiggs JL**. (2011) LOXL1 promoter haplotypes are associated with exfoliation syndrome in a US Caucasian population. Invest Ophthalmol Vis Sci. Apr 12;52(5):2372-8.

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51. Kang JH, **Wiggs JL**, Rosner BR, Hankinson SE, Haines JL, Abdrabou W, Pasquale LR. (2011) Endothelial Nitric Oxide Synthase Gene Variants and Primary Open-Angle Glaucoma: Interactions with Hypertension, Alcohol, and Cigarette Smoking. Archives of Ophthalmology 129(6):773-80.

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53. Kang JH, Loomis S, **Wiggs JL**, Stein JD, Pasquale LR (2012). Demographic and Geographic Features of Exfoliation Glaucoma in Two United States-based Prospective Cohorts. Ophthalmology, Jan;119(1):27-35.

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Narrative Report

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As a clinician-scientist I have focused my research on the identification of risk factors for glaucoma, a leading cause of blindness world-wide. Working toward this overall goal my group has identified genetic loci associated with age-related glaucoma and has genetically mapped genes causing early-onset glaucoma. Clinically I treat glaucoma patients and direct our CLIA-certified genetic testing laboratory where we routinely screen glaucoma patients for mutations in disease-causing genes. My teaching is primarily on topics related to ocular genetics and the role of genetics in the clinical care of glaucoma patients.

Research:
I am currently investigating the genetics of different types of glaucoma including adult onset primary open angle glaucoma (POAG), pseudoexfoliation glaucoma, and early-onset (childhood) glaucoma. Through collaboration we also investigate glaucoma-related environmental risk factors and gene-environment interactions.

Primary open angle glaucoma (POAG): POAG is a common type of glaucoma with complex inheritance. I am the PI of the NEIGHBOR/NEIGHBORHOOD consortium of 45 investigators from 15 different academic institutions. In 2012 we completed the NEIGHBOR genome-wide association study (GWAS), identifying a novel genomic region contributing to POAG. With additional NEI funding we expanded the consortium (creating the NEIGHBORHOOD), and in 2016 published the largest GWAS for POAG identifying 3 novel disease-associated loci (Cooke Bailey et al., *Nature Genetics*, 2016 Feb;48(2):189-94.). NEIGHBORHOOD data has contributed to over 40 publications and the discovery of more than 30

loci in total for glaucoma.

Pseudoexfoliation syndrome and glaucoma: We have completed three GWAS for pseudoexfoliation glaucoma, another common age-related glaucoma with complex inheritance, identifying 6 of the 7 known genetic loci for this condition. Previously we found that residing in northern latitudes is an environmental risk factor for this condition. We have also characterized the phenotypic features of a mouse lacking *LOXL1*, a known genetic risk factor. Our current work is focusing on rare variants that may contribute to this condition using whole genome sequencing of families and cases with the extreme phenotype of early onset before age 50.

Genes associated with ocular quantitative traits that are risk factors for glaucoma: Using homozygosity mapping combined with whole exome analysis and large consanguineous pedigrees from southern India we are identifying genes contributing to ocular quantitative traits that are risk factors for glaucoma including intraocular pressure, central corneal thickness, and optic nerve parameters.

Early-onset forms of glaucoma: We have characterized over 500 families affected by different types of early-onset glaucoma and have discovered 4 novel genetic loci for this condition. We are currently using exome sequencing and other novel genomic approaches to identify causative genes in selected families.

Administrative and Institutional Service:

I have a number of administrative roles in the department of Ophthalmology at Harvard Medical School and the Massachusetts Eye and Ear (MEE). I have been the Associate Chief for Clinical Research (Ophthalmology, MEE) since 2010 and am now Vice Chair for Clinical Research (Ophthalmology, MEE). In this position I am responsible for the implementation and development of clinical research for the Harvard department of Ophthalmology. Additionally I am the Associate Director of the Ocular Genomics Institute (Ophthalmology, MEE), the Associate Director of the Howe Laboratory (Ophthalmology, MEE), and the Director of the Genetic Testing Laboratory (Mass Eye and Ear). As director of the Genetic Testing Laboratory I oversee all procedures, manage a staff of 5 and am responsible for quality control as well as genetic test reporting. I am a long-standing member of the Institutional Review Board at the Mass Eye and Ear where I am frequently called on to chair the committee and in addition to full-board reviews I complete many expedited reviews. I am a member of the Ophthalmology Research leadership committee and a member of the Ophthalmology Residency selection committee that works each year to select the incoming resident class. For Mass Eye and Ear I am on the Medical Board as a research member, and I am a member of the hospital Research committee and Chair of the Mass Eye and Ear BioBank committee. At Harvard Medical School I am a member of the Medical Student Admissions Committee and the committee for Promotions and Reappointments and for Partner's Health Care I am a member of the Partner's Genomic Working group and Data and Tissue Sharing committees.

Teaching:

My clinical teaching includes instruction of residents, fellows, and medical students in the glaucoma clinic. Didactic lectures, given in a variety of settings, include topics in basic human genetics, ophthalmic genetics and glaucoma genetics. Annually I teach in the HMS Advanced Human Genetics course (one hour lecture on inherited eye disease) and in the HMS Molecular Basis of Eye disease course (one hour lecture on glaucoma). I am also regular mentor for HST 160 (Molecular Biology and Genetics in Modern Medicine) and a preceptor for HST 220 (Introduction to the Care of Patients). For HST 160 a student is assigned to me to observe in the glaucoma clinic and to write a paper on a selected topic. For HST 220 a student also observes in my clinic and then meets with me to discuss training to become a physician-

scientist. Additionally I supervise the research activities of approximately 4 medical students and 4 post-doctoral fellows each year.

Clinical practice:
My clinical practice includes glaucoma patients as well as patients with other inherited ocular disorders. I am board-certified in both ophthalmology and in medical genetics, with a specialization in clinical molecular genetics. I direct our CLIA-certified genetic testing laboratory where we routinely screen glaucoma patients for mutations in disease-associated genes. We have recently developed a novel selective exon capture test (GEDi) to simultaneously screen over 300 genes known to cause inherited ocular disease (glaucoma, optic atrophy and retinal degenerations). As part of this activity I prepare a genetic test report and meet with patients and referring physicians to discuss the results.

My research investigations have yielded important risk factors for glaucoma. Clinically we have created gene-based tests that can be used to establish disease susceptibility and predisposition. Our current research aims to identify additional genetic and environmental risk factors for glaucoma that will provide new information about the molecular mechanisms contributing to disease pathogenesis.