Harvard Medical School Curriculum Vitae

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Name: Eric A. Pierce

Education

1981	A.B. Magna cum laude	Biochemistry High Distinction	Dartmouth College
1986	Ph.D.	Biochemistry (Hector F. DeLuca)	University of Wisconsin-Madison
1990	M.D.	Medicine	Harvard Medical School and Massachusetts Institute of Technology, Health Sciences and Technology Division

Postdoctoral Training

07/90–06/91	Intern	Medicine	Massachusetts General Hospital
07/91–06/94	Resident	Ophthalmology	Massachusetts Eye and Ear Infirmary, Harvard Medical School
07/94–06/95	Research Fellow	Ophthalmology	Children's Hospital, Harvard Medical School
07/95–06/96	Clinical Fellow	Ophthalmology	Children's Hospital, Harvard Medical School

Faculty Academic Appointments

06/96–03/98	Instructor	Ophthalmology	Harvard Medical School
04/98–06/99	Assistant Professor	Ophthalmology	Harvard Medical School
07/99–06/07	Assistant Professor	Ophthalmology	University of Pennsylvania School of Medicine
07/07–08/11	Associate Professor with Tenure	Ophthalmology	University of Pennsylvania School of Medicine
09/11–06/12	Lecturer	Ophthalmology	Harvard Medical School
06/12	Associate Professor	Ophthalmology	Harvard Medical School

02/13-5/17	Solman and Libe Friedman Associate Professor	Ophthalmology	Harvard Medical School
06/17	Solman and Libe Friedman Professor	Ophthalmology	Harvard Medical School
07/17	William F. Chatlos Professor	Ophthalmology	Harvard Medical School

Appointments at Hospitals/Affiliated Institutions

07/96–06/99	Assistant	Ophthalmology	Children's Hospital Boston
04/97–06/99	Associate Surgeon to the Courtesy Staff	Surgery	Brigham and Women's Hospital
10/99–10/11	Active and Courtesy Staff		Presbyterian Medical Center, University of Pennsylvania Health System
01/00–10/11	Active and Courtesy Staff		Hospital of the University of Pennsylvania, University of Pennsylvania Health System
1/00–8/11	Member	Cell and Molecular Biology Graduate Group	University of Pennsylvania School of Medicine
1/00–8/11	Member	Mahoney Institute of Neurological Sciences	University of Pennsylvania School of Medicine
10/00–10/11	University Associate	Ophthalmology	Children's Hospital of Philadelphia
09/11–	Associate Surgeon	Ophthalmology	Massachusetts Eye and Ear Infirmary
10/11–	Clinical Associate	Ophthalmology	Massachusetts General Hospital
09/13–	Associate Member		Broad Institute

Other Professional Positions

2003–	Scientific Advisory Board 2004–2005, 2015 - 2005–2014	Foundation Fighting Blindness Vice Chair Chair
2012-2013	Consultant	Genzyme
2012-2015	Consultant	Novartis
2012-2014	Consultant	Allergan
2014-2015	Member	Beckman-Argyros Review Panel
2014-2015	Consultant	Isis Pharmaceuticals
2014-2016	Consultant	Biogen

2014-	Consultant	Editas Medicine
2015-	Member, Data Safety Monitoring Committee	Applied Genetics Technologies Corp (AGTC)
2015-	Member, Data Safety Monitoring Board	GenSight Biologics
2016-	Member, Advisory Board	UC Davis - Barr Foundation Retinal Disease Program
2017-	Consultant	Sanofi-Genzyme
2017-	Member, Advisory Board	Opsis Therapeutics
2018-	Consultant	Astellas Pharma

Major Administrative Leadership Positions

Local

2010-2011	Core member, Genome Frontiers Institute	University of Pennsylvania School of Medicine
2011–	Director, Ocular Genomics Institute	Massachusetts Eye and Ear Infirmary
2011–2014	Associate Director, Berman-Gund Laboratory for the Study of Retinal Degenerations	Massachusetts Eye and Ear Infirmary
2011–2014	Associate Director, ERG Service	Massachusetts Eye and Ear Infirmary
2012–2018	Director, Retinal Degeneration Fellowship	Massachusetts Eye and Ear Infirmary
2013–	Research Leadership Group, Department of Ophthalmology	Harvard Medical School
2014–	Director, Berman-Gund Laboratory for the Study of Retinal Degenerations	Massachusetts Eye and Ear Infirmary
2014–	Director, Inherited Retinal Disorders Service	Massachusetts Eye and Ear Infirmary

National and International

2007	Section Chair	FASEB Research Conference on the Biology and Chemistry of Vision Aspen, CO
2007	Co-Organizer, Course “Retinal Degeneration and Gene Therapy”	Vision Research Conference
2011–2015	Member	National Advisory Eye Council, National Institutes of Health
2012	Chair, Planning Workshop Program	National Advisory Eye Council, National Institutes of Health

2013	Co-Organizer, Course "Genetics and Genomics of Ocular Disease"	Ocular Genomics Institute Boston, MA
2015 - 2016	Site PI for clinical trial: Safety and Dose Escalation study of AAV2-hCHM in subjects with *CHM* gene mutations	Spark Therapeutics, NCT02341807
2015 -	PI for clinical trial: Safety and Tolerability of hRPC in Retinitis Pigmentosa	ReNeuron, NCT02464436
2017 -	Organizing Committee	XVIIIth International Symposium on Retinal Degeneration (RD2018)
2017 -	PI for clinical study: Natural History Study of *CEP290*-Related Retinal Degeneration	Editas Medicine (NCT03396042)

Committee Service

Local

1991–1992	Residency Steering Committee	Massachusetts Eye and Ear Infirmary
1996–1999	Ambulatory Council	Children's Hospital Boston
2002–2011	DNA Sequencing Facility Advisory Committee	University of Pennsylvania School of Medicine
2004–2011	Biomedical Imaging Core Facility Advisory Committee	University of Pennsylvania School of Medicine
2005–2009	Resident Selection Committee, Department of Ophthalmology	University of Pennsylvania School of Medicine
2007–2011	School of Medicine Animal Research Committee	University of Pennsylvania
2008–2010	Interviewer, Admissions Committee, Cell and Molecular Biology Graduate Group	University of Pennsylvania School of Medicine
2011	Scientific Advisory Committee	Penn Genome Frontiers Institute
2011	Thesis Committee Chair, Cell and Molecular Biology Graduate Group	University of Pennsylvania School of Medicine
2012–2015	Research Committee	Massachusetts Eye and Ear Infirmary
2013	Resident Selection Committee, Department of Ophthalmology	Harvard Medical School
2016-	Research Steering Committee	Massachusetts Eye and Ear Infirmary

Professional Societies

1992–	Association for Research and Vision and Ophthalmology	Member

	2003	Organizer, Course "Conditional Gene Targeting"
	2012	Co-Organizer, Symposium “RNA Editing in Retinal Health and Disease”
	2012	Organizer, Session "Retina Ciliopathies and Animal Models"
1998-2005	American Association for Pediatric Ophthalmology and Strabismus	Member
	1997	Organizer, Course "Retinopathy of Prematurity: New Findings Regarding the Pathogenesis and New Possibilities for Therapy"
	1999	Co-Organizer, Course "Retinopathy of Prematurity: New Research Findings and Implications for Treatment"
2008–	American Society for Human Genetics	Member

Grant Review Activities

2000–2002	Grant Review Panel	Fight for Sight, Prevent Blindness America Member
2001	Special Emphasis Panel ZRG1	Center for Scientific Review National Institutes of Health Member
2005	Annual Grant Review Panel	Foundation Fighting Blindness Canada Member
2006–2014	Annual Grant Review Panel	Foundation Fighting Blindness, Chair
2008	Biology and Diseases of the Posterior Eye Study Section	Center for Scientific Review National Institutes of Health Ad Hoc Member
2018-	Diseases and Pathophysiology of the Visual System (DPVS)	Center for Scientific Review National Institutes of Health Permanent Member

Editorial Activities

Ad hoc reviewer

- *American Journal of Pathology*
- *eLife*
- *Gene Therapy*
- *Genetics in Medicine*
- *Human Molecular Genetics*

- *Human Mutation*
- *Investigative Ophthalmology and Visual Sciences*
- *Journal of Biological Chemistry*
- *Journal of Cell Biology*
- *Journal of Clinical Investigation*
- *Journal of Neuroscience*
- *Molecular and Cellular Proteomics*
- *Nature Genetics*
- *Neuron*
- *Proceedings National Academy of Sciences USA*
- *Science Translational Medicine*

Other Editorial Activities

2011-	Editorial Review Board	Molecular Vision
2012-	Editorial Board	Translational Vision Science and Technology
2015	Guest Editor	Proceedings National Academy of Sciences USA

Honors and Prizes

1981	Inductee	Phi Beta Kappa Alpha Chapter of New Hampshire
1984-1986	Babcock Fellow	University of Wisconsin-Madison
1994-1995	Fellow	Heed Ophthalmic Foundation
1996	Von L. Meyer Fellowship Award	Children's Hospital, Boston
1999-2011	Fellow	Rosanne H. Silbermann Foundation
2001	Everson Lecturer in Biochemistry	University of Wisconsin-Madison
2005	Lloyd Morgan Lecturer in Ophthalmology	Hospital for Sick Children, University of Toronto
2007	Distinguished Visiting Scholar	University of Oklahoma Health Science Center
2008	Robb-Petersen Lecturer in Pediatric Ophthalmology	Children's Hospital Boston, Harvard Medical School
2009	Distinguished Speaker	Gerald Fishman Lecture Series, University of Illinois at Chicago
2009	Board of Directors Award	Foundation Fighting Blindness
2010	Roger Johnson Lecturer in Ophthalmology	Children's Hospital and Regional Medical Center, Seattle, Washington
2010	Best Senior Poster Presentation	XIV International Symposium on Retinal Degeneration
2012	Richard Weleber Lecturer in Ophthalmology	Casey Eye Institute, Oregon Health Sciences University, Portland, Oregon
2012	Foundation for Retina Research Family Focus Award	Foundation for Retina Research

2013	Ephraim Friedman Lecturer in Ophthalmology	Massachusetts Eye and Ear Infirmary, Harvard Medical School
2013	Distinguished Lecturer	Department of Ophthalmology, University at Buffalo, State University of New York
2014	Robert Meyers Lecturer in Ophthalmology	Penn State Hershey Eye Center, Hershey, Pennsylvania
2014	Crystal Ball Award	Foundation for Retina Research
2014	Nelson Trust Award for Retinitis Pigmentosa	Research to Prevent Blindness
2015	Purnell Lecturer in Ophthalmology and Visual Sciences	Department of Ophthalmology and Visual Sciences, Case Western Reserve University, Cleveland, Ohio
2016	Alcon Research Award	The Alcon Research Institute

Report of Funded and Unfunded Projects

Funding Information

Past

1994–1995
The Biochemistry of Retinopathy of Prematurity
Private foundation grant, Knight's Templar Eye Foundation
PI
The goal of this project was to investigate the role of vascular endothelial growth factor (VEGF) in retinal neovascularization.

1994–1996
The Biochemical Basis of Retinal Neovascularization
Private foundation grant, V. Kann Rasmussen Foundation
PI
The goal of this project was to identify genes that undergo changes in their level of expression in the retina in response to hypoxia, and thus may be involved in the pathogenesis of retinal neovascular disease.

1994–1999
The Biochemical Basis of Retinal Neovascularization
NEI K11 EY00343
PI
The goals of this project were to investigate the role of vascular endothelial growth factor (VEGF) in retinal neovascularization, and to identify other genes that undergo changes in their level of expression in the retina in response to hypoxia, and thus may be involved in the pathogenesis of retinal neovascular disease.

1998–1999
Vascular Endothelial Growth Factor in Retinopathy of Prematurity
Private foundation grant, Blind Children's Center
PI
The goal of this pilot clinical project was to study the use of the angiogenic factor VEGF as a marker for the severity of retinopathy of prematurity (ROP).

1998–1999 Vascular Endothelial Growth Factor in Retinopathy of Prematurity
Private foundation grant, The Peabody Foundation
PI
The goal of this project was to test the hypothesis that VEGF levels in blood and urine correlate with the severity of ROP.

1998–1999 Vascular Endothelial Growth Factor in Retinopathy of Prematurity
Private foundation grant, Massachusetts Lions Eye Research Fund
PI
The goal of this pilot clinical project was to study the use of the angiogenic factor VEGF as a marker for the severity of retinopathy of prematurity.

1998–1999 Characterization of a Novel Photoreceptor Gene and Investigation of its Role in Retinitis Pigmentosa
Private foundation grant, Boston Children's Hospital Pilot Study Award
PI
The objective of this project was to investigate the role of a newly discovered oxygen-sensitive, photoreceptor-specific gene (*ORP1*) in retinitis pigmentosa.

1998–1999 Characterization of a Novel Photoreceptor Gene and Investigation of its Role in Retinitis Pigmentosa
Private foundation grant, Fight for Sight
PI
The objective of this project was to investigate the role of a newly discovered oxygen-sensitive, photoreceptor-specific gene (*ORP1*) in retinitis pigmentosa.

1999–2002 Vascular Endothelial Growth Factor In Retinopathy Of Prematurity
Private foundation grant, March of Dimes Birth Defects Foundation 6-FY99-724
PI
The goal of this clinical project was to study the use of the angiogenic factor vascular endothelial growth factor as a marker for the severity of retinopathy of prematurity.

1999–2006 A Novel Photoreceptor Protein & Retinal Degenerations
Private foundation grant, Foundation Fighting Blindness T-GE-0902-0223
PI
The goal of this project was to investigate the mechanism by which mutations in *RP1* lead to retinal degeneration.

1999–2011 Investigations Of Retinal Degenerations
Private foundation grant, Rosanne H. Silbermann Foundation
PI
The objective of this project was to improve our understanding of how age-related and inherited retinal degenerations cause blindness.

2000–2003 Career Development Award
Private foundation grant, Research to Prevent Blindness
PI
The goals of this project were to study to the role of the RP1 protein in vision, and to develop an animal model of the *RP1* form of retinitis pigmentosa.

2001–2003 Animals with Targeted Mutations in Photoreceptor Genes
NIH/NEI RO3 EY013776
PI
The goal of the proposed research was to develop the use of oligonucleotides for creating animals with targeted mutations in photoreceptor genes, to use as models of retinal disease.

2002–2005 RNA Splicing Proteins and Retinal Degenerations
Private foundation grant, E. Matilda Zeigler Foundation For The Blind
PI
The goal of this project was to create mouse models of retinitis pigmentosa 13 (RP13), which is caused by mutations in the Pre-RNA Processing Factor 8 (*PRPF8*) gene, and use the mice to study the pathogenesis of RP13.

2003–2008 Program: Examining Oligonucleotide Effects on Gene Expression (Gewirtz)
Project: Gene Manipulation in Stem Cells by Oligonucleotides (Pierce)
Program project, NIH/NCI 2PO1-CA72765
Project Leader
The goal of the funded research was to develop the use of oligonucleotides for gene targeting in mouse embryonic stem cells.

2004–2006 A Mouse Model Of Inherited Macular Degeneration
Private foundation grant, Research to Prevent Blindness
PI
The goals of this project were to create a mouse model of Doyne Honeycomb Retinal Dystrophy (DHRD)/Malattia Leventinese (ML) by using gene targeting techniques to introduce the R345W mutation into the mouse *Efemp1* gene, and then to use the resulting mice to investigate the pathogenesis of drusen formation.

2004–2005 The Nanomedicine of Supramolecular Cellular Compartments
Program project, NIH PN1 EY016555
Co-PI
Planning grant for the Nanomedicine Roadmap Initiative

2006–2009 Photoreceptor Cell Death and Rescue in *RP1* Disease
Private foundation grant, Foundation Fighting Blindness BR-CMM-0406-0337
PI
The goal of this project was to investigate the mechanism by which mutations in *RP1* cause programmed cell death of photoreceptors, and test the use of non-sense suppression to treat *RP1* disease.

2006–2009 Development of C57BL/6 ES Cell Technology for High Throughput Use
Program project, NIH UO1 DA 021912
Investigator
This goal of this project was to develop C57BL/6 embryonic stem cell lines for high-throughput gene targeting for use in the NIH-sponsored Knockout Mouse Project (KOMP).

2007–2010 Basal Deposit Formation In *Efemp1*-R345W Mice
Private foundation grant, Ruth And Milton Steinbach Fund
PI
The goal of this project was to investigate the pathogenesis of basal deposit formation in the *Efemp1*-R345W mice.

2009 RPB Research Sabbatical
Private foundation grant, Research to Prevent Blindness
PI
This funding helped support a research sabbatical for the PI at the Institut de la Vision in Paris and the Nijmegen Center for Molecular Life Sciences in the Netherlands.

2007–2011 CHOP-Penn Pediatric Center For Retinal Degenerations
Private foundation grant, Foundation Fighting Blindness C-CL-0607-389-UPA01
Project Leader
The goal of this Center is to improve our understanding of and to develop therapies for inherited retinal degenerations that affect children.

2009–2012 RNA Splicing Factors In Retinitis Pigmentosa
Private foundation grant, Foundation Fighting Blindness BR-CMM-0409-0482-UPA
PI
The goal of this project was to investigate the mechanism by which mutations in RNA splicing factors cause retinitis pigmentosa.

2010–2012 Translational and Personalized Genomics of Inherited Retinal Degenerations
Private foundation grant, Penn Genome Frontiers Institute
P1 and Investigator
The goals of the funded research were to use next-generation sequencing approaches to identify new IRD disease genes, and to investigate the pathogenesis of RP caused by mutations in RNA splicing factors.

2012-2013 Disease Gene Discovery for Leber Congenital Amaurosis
Curing Kids Fund, Massachusetts Eye and Ear Infirmary
PI
The goal of this project was to identify novel Leber congenital amaurosis disease genes.

2008–2013 Customized iPS Cell Therapy For Recessive Monogenetic Retinal Degenerative Disease
Private foundation grant, Foundation Fighting Blindness TA-CBT-0607-0444-UWI-WG
Project Leader
The goal of this project was to test the hypothesis that iPS cell and gene repair technologies can be combined to create a customized therapeutic strategy for monogenetic degenerative diseases of the retina.

2010–2014 TreatRush EU Project (Fighting Blindness of Usher Syndrome: Diagnosis, Pathogenesis and Retinal Treatment)
Federal grant, European Union 7th Framework Program
Investigator
The goals of this project were to investigate the molecular bases of and develop therapies for Usher syndrome.

2014-2016 The Role of Complement in an Inherited Macular Degeneration
Sponsored Research Agreement, Novartis
PI
The objective of this project was to further investigate the role of the pathogenesis of inherited macular dengeration

2013–2016 Gene Therapy for *NMNAT1* LCA
Private foundation grant, Foundation for Retinal Research/Gavin R Stevens Foundation
PI
The goal of this project was to develop gene augmentation therapy for Leber congenital amaurosis (LCA) caused by mutations in the *NMNAT1* gene.

2014-2017 Gene Therapy for *PRPF31*-Associated Retinal Degeneration
Private foundation grant, Rosanne Silbermann Foundation
PI
The goal of this project is to test the use of gene augmentation therapy to treat *PRPF31*-associated retinal degeneration.

2013-2017 Finding Elusive RP Genes
Private foundation grant, Foundation Fighting Blindness BR-GE-1213-0633-MEEI
PI
The goal of this project was to utilize whole genome sequencing analysis and human iPS cell (hiPSC)-based approaches to develop technique(s) for the discovery of RP-causing gene mutations that escape standard methods of detection.

Current

1999–2018 Novel Photoreceptor Proteins And Retinal Degenerations
NIH 2R01 EY012910
PI ($1,848,254) (Total direct costs for current funding cycle)
The major goals of this project are to identify novel retinal degeneration disease genes, investigate the mechanisms by which mutations in the *TTC21B* and *NMNAT1* genes cause photoreceptor degeneration, and test the hypothesis that gene augmentation therapy can provide therapeutic benefit for retinal degeneration caused by mutations in the *RP1* gene.

2011–2020 The Pathogenesis of RNA Splicing Factor Retinitis Pigmentosa
NIH 1R01 EY020902
PI ($2,059,862)(total direct costs for current funding cycle)
The goal of this project is to test the hypotheses that mutations in RNA splicing factors cause RP by disrupting mRNA splicing in the RPE and retina, and develop gene therapy for one form of RNA splicing factor RP.

2014-2019 RP Research Center at the Berman-Gund Laboratory
Private foundation grant, Foundation Fighting Blindness
Director, PI Modules I and II ($2,500,000)
The overall goals of the funded research are to continue our translational studies directed toward developing gene-independent and gene-specific therapies for inherited retinal degenerative disorders.

2014-2018 Core Grant for Vision Research
NIH 2P30EY014104 Wiggs(PI)
Module Director ($655,795)(Direct costs, Module 1)
This grant provides support for shared research resources for Harvard vision investigators.

2016-2018	Efficacy, Safety, and Toxicity of AAV-mediated Human *RPGRIP1* Gene Augmentation Therapy Private foundation grant, Foundation Fighting Blindness PI ($1,241,368) The goal of the funded research is to perform IND-enabling studies for AAV-mediated Human *RPGRIP1* Gene Augmentation Therapy.
2016-2020	Joint Center for Mendelian Genomics NIH 1UM1HG008900 MacArthur/Rehm (PIs) Investigator ($50,438)(Pierce) The goal of this Joint Center is to apply genomic techniques to large collections of families to discover the genetic causes of Mendelian diseases.
2017-2022	Genetic Causes and Genetic Modifiers of Inherited Retinal Degenerations NIH 1R01EY026904 Bujakowska/Pierce (PIs) Multi-PI ($1,746,321; total direct costs for current funding cycle) The goals of the funded research are to improve our ability to identify the genetic cause of disease for patients affected by IRDs, and to identify additional genetic features that modify the severity of disease.
2017-2020	CRISPR/Cas Gene Editing for USH2A-Associated Retinal Degeneration Editas Medicine Liu (PI) Co-PI ($1,005,709; total direct costs) The goal of the funded research is to develop a CRISPR/Cas – mediated genome editing strategy for the treatment of USH2A-associated retinal degeneration.

Report of Local Teaching and Training

Teaching of Students in Courses

1994-1999	HST Functional Anatomy 40 HST medical and MEMP students	Harvard Medical School Lecturer, lab instructor for 1 lecture and lab session per year
1999-2001	Ophthalmology Clerkship Medical students	University of Pennsylvania School of Medicine Preceptor, 3 hours per year
09/2003	Pediatric Ophthalmology: Amblyopia and Strabismus Medical students	University of Pennsylvania School of Medicine One 1-hour lecture
02/2004	Neuroscience Curriculum: Bovine Eye Dissection Medical students	University of Pennsylvania School of Medicine Preceptor, 1 hour
2004–2006	Biomedical Graduate Studies: Bioethics Workshop Graduate students	University of Pennsylvania Facilitator, 1 hour/year
11/2005	Clinical Neuroscience Track: Inherited Retinal Degenerations Medical Students	University of Pennsylvania School of Medicine One 1-hour lecture

2005–2008	Genetic Medicine course Medical students	University of Pennsylvania School of Medicine Section leader, one 1- to 2-hour section/week (12 hours/year)
09/2008	Inherited Retinal Degenerations—A Class of Neurodegenerative Disorders Clinical Neuroscience Training Program students	University of Pennsylvania School of Medicine One 1-hour lecture
2008–2009	Inherited Retinal Degenerations and Retinal Gene Therapy Medical students	University of Pennsylvania School of Medicine One 1-hour lecture/month (10 hours/year)
04/2011	Inherited Retinal Degenerations Clinical Neuroscience Training Program students	University of Pennsylvania School of Medicine One 1-hour lecture
03/2012	Genetics of Inherited Retinal Degenerations Molecular Bases of Eye Diseases course students	Harvard Medical School One 1-hour lecture
04/2013	Genetics of Inherited Retinal Degenerations Molecular Bases of Eye Diseases course students	Harvard Medical School One 1-hour lecture
01/2014	Faculty member, Nanocourse on Ciliogenesis	Harvard Medical School One 1-hour lecture
04/2014	Genetics of Inherited Retinal Degenerations Molecular Bases of Eye Diseases course students	Harvard Medical School One 1-hour lecture
03/2015	Genetics of Inherited Retinal Degenerations Molecular Bases of Eye Diseases course students	Harvard Medical School One 1-hour lecture

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

1996-1999	Pediatric Ophthalmology Conference Ophthalmology residents and fellows	Children's Hospital Boston 8 1-hour seminars per year
12/2003	Current Concepts in Pediatric Ophthalmology and Strabismus: Children with Retinal Degenerations Ophthalmology residents and fellows	Children's Hospital of Philadelphia One 1-hour lecture
2004–2005	Pediatric Ophthalmology Course: Retinal Degenerations in Children Ophthalmology residents	University of Pennsylvania School of Medicine One 1-hour lecture/year
2008	Retinal Degenerations and Dystrophies in Children Ophthalmology residents	University of Pennsylvania School of Medicine One 1-hour lecture

2008	Inherited Retinal Degenerations and Retinal Gene Therapy Ophthalmology residents	University of Pennsylvania School of Medicine One 1-hour lecture
2013	Inherited Retinal Degenerations and Gene Therapy Ophthalmology residents	Massachusetts Eye and Ear Infirmary One 1-hour lecture
2013	Genetic Bases of Inherited Retinal Degenerations Ophthalmology residents	Massachusetts Eye and Ear Infirmary One 1-hour lecture

Clinical Supervisory and Training Responsibilities

1996-1999	Attending, Pediatric Ophthalmology Clinic and Surgery Medical students, ophthalmology residents, and pediatric ophthalmology fellows	Children's Hospital Boston 2 days/week
2000–2005	Attending, Pediatric Ophthalmology Clinic and Surgery Medical students, ophthalmology residents, and pediatric ophthalmology fellows	Children's Hospital of Philadelphia 1 day/week
2005–2011	Attending, Retinal Degeneration Clinic Medical students, ophthalmology residents, and pediatric ophthalmology fellows	Children's Hospital of Philadelphia 4–6 half-day clinic sessions/month
2012–	Attending, Inherited Retinal Disorders Service Retinal degeneration fellow, Ophthalmology residents	Massachusetts Eye and Ear Infirmary 4 to 8 full day clinic sessions/month

Laboratory and Other Research Supervisory and Training Responsibilities

1995–1999	Laboratory pre-doctoral research mentorship, Harvard Medical School	Daily supervision of 2 pre-doctoral students
1999–2011	Laboratory postdoctoral research mentorship (residents and research/clinical fellows), University of Pennsylvania School of Medicine	Weekly mentorship of 1-5 postdoctoral trainees/year; 2 hours/trainee/week
2001–2011	Laboratory pre-doctoral research mentorship (undergraduate, medical, and graduate students), University of Pennsylvania School of Medicine	Weekly mentorship of 1-4 pre-doctoral trainees/year; 4 hours/trainee/week
2004, 2005	Examination Committee Member, Cell and Molecular Biology Graduate Group, University of Pennsylvania School of Medicine	3 PhD students; 3 hours/student

2007–2009	Thesis Committee Member, Neuroscience Program, University of Pennsylvania School of Medicine	2 PhD students; 6 hours/student
2011–present	Laboratory postdoctoral research mentorship, Ocular Genomics Institute, Harvard Medical School	Weekly mentorship of 4-5 postdoctoral trainees/year; 1-2 hours/trainee/week

Formally Supervised Trainees and Faculty

1995–1996 Eliot Foley, MD / Private practice, Concord, NH
Pre-doctoral student: The Role of Vascular Endothelial Growth Factor in Retinal Neovascularization. Co-authored five publications

1996–1998 Terrence Meehan, PhD / Scientific Curator, Jackson Laboratories
Pre-doctoral student: Identification of Genes Involved in Retinal Neovascularization. Co-authored one publication in Nature Genetics.

1999–2005 Qin Liu, MD, PhD / Assistant Professor of Ophthalmology, Harvard Medical School
Postdoctoral fellow: The Retinitis Pigmentosa 1 (RP1) Protein and Retinal Degenerations. Co-authored 16 publications to date.

2000–2001 Franco Recchia, MD / Private practice, Nashville, TN
Postdoctoral fellow: Identification of Genes Involved in Retinal Neovascularization

2000–2001 Haiching Ma, PhD/ Chief Technology Officer, Reaction Biology Corporation
Postdoctoral fellow: Lipofection of Mouse Embryonic Stem Cells. Co-authored two publications.

2000–2002 Jie (Jean) Zhou, MD, PhD / Research Associate, Laboratory for Nutrition and Vision Research, Jean Mayer USDA Human Nutrition Research Center on Aging (HNRCA) at Tufts University
Postdoctoral fellow: Regulation of RP1 Expression. Co-authored one publication.

2001–2004 Leonard Feiner, MD, PhD / Private practice, Retinal Consultants of New Jersey
Ophthalmology resident: Photoreceptor-Specific Cre Mice. Co-authored two publications.

2001–2003 Hadas Mechoulam, MD / Senior Physician, Pediatric Ophthalmology, Haddassah Medical Center, Jerusalem, Israel
Postdoctoral fellow: Gene Expression Analysis in Retinal Neovascularization. Co-authored two publications.

2001–2003 Jason Skalet, MD / Private practice, Oakland, CA
Medical student: The Role of the Retinitis Pigmentosa 1 Protein in Photoreceptor Biology. Co-authored one publication.

2002 Gregory Supple, MD / Assistant Professor of Medicine, University of Pennsylvania School of Medicine
Medical student: Identification of RP1 Interacting Proteins

2002 David Garbe, PhD / Senior Research Associate, University of Pennsylvania School of Medicine
Graduate student: Generation of Pre-RNA Processing Factor 3 (Prpf3) T494M Knock-in Mice

2002–2004	Sarah Achenbach, PhD / Senior Field Applications Specialist, Promega Corporation Graduate student: Identification of RP1 Interacting Proteins
2002–2006	Bertrand Deramaudt, PhD / Biotechnology professional, Strasbourg, France Postdoctoral fellow: Pre-RNA Processing Factor 8 and Inherited Retinal Degenerations. Co-authored one publication.
2002–2009	John Graziotto, PhD / Scientist, Lysosomal Therapeutics Inc PhD thesis supervisor: Animal Models of RNA Splicing Factor Retinal Degenerations. Co-authored three publications.
2003	Cathy Cukras, MD, PhD / Staff Physician, Clinical Trials Branch, National Eye Institute Ophthalmology resident: Identification of RP1 Interacting Proteins
2003	Kathryn Claiborn, PhD / Research Associate, Department of Genetics and Complex Diseases, Harvard T.H. Chan School of Public Health Graduate student: Generation of *Rp1* Knockout Mice
2003–2007	Brian Murphy, PhD / Research Engineer, Celgene PhD thesis supervisor: Optimization and Analysis of Single-Stranded Oligonucleotide-Directed Gene Targeting. Co-authored one publication.
2004	David Diaz, MD / Private practice, Burlington, VT Medical student: Animal Models of RNA Splicing Factor Retinal Degenerations
2004	Stacie Dilks, PhD / Senior Medical Writer at Health Interactions, San Diego, California Graduate student: Characterization of *Efemp1*-R345W Knock-in Mice
2004–2005	Hamideh Moayedpardazi, MD/ Glaucoma Fellow, UC San Diego Shiley Eye Institute Undergraduate student: Oligonucleotide-Directed Gene Targeting
2004–2005	Nicole Benitah, MD / Private practice, Los Angeles, CA Howard Hughes Medical Institutes Medical Student Fellow: The Microtubule-Associated Protein Functions of RP1
2006–2016	Qi Zhang MD, PhD / Senior Research Investigator Co-authored seven publications
2007–2008	Lauren Danielle, PhD / Postdoctoral Fellow, University of California, Davis PhD thesis committee member: Insights Into Cone Function From the "All-Cone" Neural Retina Leucine Zipper Null Mouse. Co-authored one publication.
2007–2011	Barbara Zangerl, DVM, PhD / Research Assistant Professor of Medical Genetics in Ophthalmology, University of Pennsylvania School of Veterinary Medicine Junior faculty member mentoring committee
2007–2016	Donna Garland, PhD / Senior Research Investigator Co-authored four publications
2006	Gagan Sawhney, MD / Private practice, Atlanta, GA Medical student: RP1 Interacting Proteins
2010–2011	Aaron Black, P.D candidate, University of Pennsylvania School of Medicine PhD thesis committee chair: Pathophysiology of and Gene Therapy for Choroideremia
2009–2015	Mike Farkas, PhD / Assistant Professor of Ophthalmology, University at Buffalo Postdoctoral Fellow: The pathogenesis of RNA splicing factor retinitis pigmentosa Co-authored seven publications

2010–2011	Yolanda Cui, PhD candidate, University of Pennsylvania School of Medicine PhD thesis supervisor; Novel Cilia Proteins in Photoreceptor Biology
2011	Yafeng Li, MD, PhD candidate, University of Pennsylvania School of Medicine PhD thesis supervisor; The Genetic Mechanism of RP1 Disease
2011-2012	Zoe Fonseca-Kelly, PhD / Assistant Dean, Harvard University Co-authored two publications
2011-2016	Rosario Fernandez Godino, PhD / Instructor in Ophthalmology, Harvard Medical School Postdoctoral Fellow: The pathogenesis of early macular degeneration Co-authored four publications
2011-2012	Inderjeet Kaur, PhD / Investigator, LV Prasad Eye Institute Hyderabad, India Supervisor, visiting scientist
2011-	Jason Commander, MD, PhD / K12 Mentor
2012-2013	Miriam Ehrenberg, MD/Clinical Fellow
2012	Christine Binder, MD, PhD / Resident Radiation Medicine, Oregon Health & Sciences University, Portland, Oregon PhD thesis committee member; Toward the Development of Ocular Gene Therapy
2012-2016	Kinga Bujakowska, PhD / Instructor in Ophthalmology, Harvard Medical School Postdoctoral Fellow: The genetics of inherited retinal degenerations Co-authored five publications
2012-2013	Daniel G. Taub/Pre-doctoral researcher Co-authored one publication
2013-2014	Xiang Werdich, MD/Clinical Fellow Co-authored one publication
2013-	Scott Greenwald, PhD/ Postdoctoral researcher Co-authored one publication to date
2014-2015	Brian Hafler, MD, PhD / Postdoctoral Fellow, Department of Genetics, Harvard Medical School Clinical-Research Fellow Co-authored one publication
2014-	Clara Jiayun Men/Medical Student Recipient of Medical Student Fellowship, Research to Prevent Blindness
2015-2016	Rachel Huckfeldt, MD, PhD / Instructor in Ophthalmology, Harvard Medical School Clinical-Research Fellow
2016-2017	Pingjuan Li, PhD / Postdoctoral researcher Co-authored one publication
2016-2017	Mauricio Vargas, MD, PhD / Clinical-Research Fellow
2016-	Nachiket Pendse, PhD / Postdoctoral researcher
2017-2018	Farzad Jamshidi, MD, PhD / Postdoctoral researcher Co-authored one publication
2017-	Caitlin Collin, PhD / Postdoctoral researcher

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

No presentations below were sponsored by outside entities.

03/1998	Ocular Trauma Injuries to Children Course, Harvard Medical School	Single presentation, Boston, MA
12/2003	Children with Retinal Degenerations Current Concepts in Pediatric Ophthalmology and Strabismus, Children's Hospital of Philadelphia	Single presentation, Philadelphia, PA
02/2007	Retinal Physiology and Hereditary Retinal Degenerations Ophthalmology Review Course, Scheie Eye Institute	Single lecture, Philadelphia, PA
10/2009	Retinal Degenerations and Dystrophies in Children Sixth Annual Update on Pediatric Ophthalmology: The Pediatric Retina, Children's Hospital of Philadelphia	Single lecture, Philadelphia, PA
10/2013	"Genetics of Inherited Retinal Degenerations"/ Course "Genetics and Genomics of Ocular Disease" Ocular Genomics Institute, American Society of Human Genetics Annual Meeting	Single lecture, Boston, MA
4/2018	"The Promise of Genome Editing"/Course "Inherited Retinal Diseases: Divergent Viewpoints of Pathogenesis and Treatment" ARVO Annual Meeting	Single presentation, Honolulu, HI

Local Invited Presentations

12/1994	"Vascular Endothelial Growth Factor in Retinopathy of Prematurity" / Plenary Talk Longwood Medical Area Ophthalmology Conference, Harvard Medical School, Boston, Massachusetts
04/1998	"ROP and RP: A Tale of Two Diseases" / Plenary Talk Schepens Eye Research Institute Boston, Massachusetts
05/1998	"ROP and RP: A Tale of Two Diseases" / Plenary Talk Massachusetts Eye and Ear Infirmary, Boston, Massachusetts
12/1998	"The Role of VEGF in Retinopathy of Prematurity and a Novel Oxygen-Regulated Protein in Retinitis Pigmentosa" / Plenary Talk Seminars in Vascular Biology, Harvard Medical School, Boston, Massachusetts
10/1999	"Mutations in *ORP1* cause retinitis Pigmentosa 1" / Plenary Talk Scheie Eye Institute, University of Pennsylvania
01/2002	"Inherited Retinal Degenerations" / Grand Rounds Children's Hospital of Philadelphia, Pennsylvania
01/2003	"Mutations in the photoreceptor protein RP1 cause inherited blindness" / Plenary Talk Biochemistry Department, University of Pennsylvania School of Dental Medicine Philadelphia, Pennsylvania
03/2006	"The Proteome of a Mammalian Sensory Cilium: The Photoreceptor Outer Segment" / Plenary Talk Renal Research Conference, Department of Medicine, University of Pennsylvania School of Medicine, Philadelphia, Pennsylvania

06/2010 "Next Generation Sequencing Approaches to Study the Genetics and Pathogenesis of Retinal Degenerations"/ Plenary Talk
Children's Hospital of Philadelphia Next Generation Sequencing Symposium

05/2011 "Next Generation Sequencing for Retinal Degeneration Genetics" / Plenary Talk
Scheie Eye Institute, Philadelphia, Pennsylvania

06/2011 "Next Generation Sequencing for Inherited Retinal Degeneration Genetics" / Plenary Talk
Children's Hospital of Philadelphia Next Generation Sequencing Symposium

10/2011 "Mechanism of Disease and Potential Therapy for Retinitis Pigmentosa 1" / Plenary Talk
Bertarelli Symposium, Harvard Medical School, Boston, Massachusetts

12/2011 "Next Generation Sequencing for Retinal Degeneration Genetics" / Plenary Talk
Genome Sequencing Seminar Series, Department of Genetics, Harvard Medical School, Boston, Massachusetts

03/2012 "NGS for Inherited Retinal Degenerations Genetics" / Plenary Talk
Massachusetts Eye and Ear Infirmary Research Retreat, Portsmouth New Hampshire

06/2012 "*NMNAT1* Mutations Cause Leber Congenital Amaurosis in the LCA9 Locus"/
Plenary Talk, Massachusetts Eye and Ear Infirmary Annual Meeting, Boston, Massachusetts

06/2012 "Inherited Retinal Degenerations and Potential Interventions" / Plenary Talk
Cell Therapy in the Retina, Harvard Stem Cell Institute, Boston, Massachusetts

09/2012 "Neurodegenerative Disease" / Panel Discussion
Symposium on Age-Related Macular Degeneration 2012, Boston, MA

02/2013 "Deciphering the role of complement and the extracellular matrix in early stage macular degeneration using mouse genetics and proteomics"
Ephraim Friedman Lecture, Massachusetts Eye and Ear Infirmary, Boston, Massachusetts

03/2016 "Ocular Genomics Institute" / Plenary Talk, Faculty Retreat, Department of Ophthalmology, Harvard Medical School, Boston, MA

09/2016 "Phenotype vs. Genotype – Updating Our Definitions of Disease"/ Grand Rounds, Department of Ophthalmology, Harvard Medical School, Boston, MA

Report of Regional, National and International Invited Teaching and Presentations

Invited Presentations and Courses

Those presentations below sponsored by outside entities are so noted and the sponsors are identified.

Regional

12/1996 "Pediatric Cataract To Implant or Not to Implant" / Plenary Talk
New England Ophthalmological Society, Boston, Massachusetts

10/1999 "Mutations in the Novel Oxygen-Regulated Photoreceptor Protein ORP1 Cause Retinitis Pigmentosa 1 (RP1)" / Plenary Talk
Coriell Institute for Medical Research, Camden, New Jersey

04/2001 "Mutations in RP1 cause Inherited Blindness by Disrupting Photoreceptor Protein Transport" / Plenary Talk
Thomas Jefferson Institute of Molecular Medicine, Philadelphia, Pennsylvania

01/2005	"RP1 is a Photoreceptor MAP" / Plenary Talk Visual Neuroscience Training Program, Johns Hopkins University, Baltimore
03/2005	"Investigations of Dominant Retinitis Pigmentosa" / Grand Rounds Wills Eye Hospital, Philadelphia, PA
03/2007	"The Proteome of the Photoreceptor Sensory Cilium" / Plenary Talk Edward S. Harkness Eye Institute, Columbia University College of Physicians and Surgeons, New York, New York
01/2008	"Gene Targeted Mouse Models of Inherited Retinal Degenerations" / Plenary Talk National Eye Institute, National Institutes of Health, Bethesda, Maryland
11/2010	"Investigations of Inherited Retinal Degeneration Genetics Using Exon Capture and Illumina Sequencing" / Plenary Talk Illumina Users Group Meeting, Philadelphia, Pennsylvania (Illumina, San Diego, CA)
01/2011	"Exon Capture and Next Generation Sequencing for Retinal Degeneration Genetics"/ Plenary Talk Agilent Users Group Meeting, Philadelphia, Pennsylvania (Agilent, Wilmington, DE)
04/2011	"Next Generation Sequencing for Retinal Degeneration Genetics" / Plenary Talk Illumina Diagnostics Meeting, Philadelphia, Pennsylvania
12/2011	"Genetics of and Genetic Therapies for Inherited Retinal Degenerations" / Plenary Talk Nurses and Allied Health Personnel in Ophthalmology Meeting, New England Ophthalmological Society, Boston, Massachusetts
04/2012	"Genetics of Inherited Retinal Degenerations" / Plenary Talk Genetics Clinic Conference, Department of Medicine, Massachusetts General Hospital, Boston, Massachusetts
06/2012	"Efficacy Study in Retinal Diseases" / Plenary Talk Foundation Fighting Blindness Optogenetics Workshop, Boston, Massachusetts
06/2012	"Genetics of Inherited Retinal Dystrophies" / Plenary Talk Neuroscience Grand Rounds, Massachusetts General Hospital, Boston, Massachusetts
09/2013	"Genetics Diagnostic Testing for Patients with Inherited Retinal Degenerations" / Keynote Address Illumina Users Group Meeting, Boston, Massachusetts
09/2013	"Genetics and Genomics of Inherited Retinal Degenerations" / Plenary Talk Frontiers in Vision Lecture Series, Tufts University School of Medicine, Boston, Massachusetts
10/2013	"Inherited Retinal Degenerations: Genetics and Gene Therapy"/ Plenary Talk, Joubert Syndrome Scientific Meeting, Boston, MA
04/2014	"Genetics of and Gene Therapies for Inherited Retinal Degenerations"/ Plenary Talk, Medical Population Genetics, Broad Institute, Cambridge, MA
04/2015	"Gene Editing, Gene Therapy, and the Eye as a Gateway"/ Invited Panel Member, Partners World Medical Innovation Forum, Boston, MA
06/2015	"Genetics Diagnosis of and Gene Therapies for Inherited Retinal Degenerations" / Plenary Talk, New England Ophthalmological Society, Boston, Massachusetts

06/2015 "Ocular Genomics Institute"/ Session Organizer, Medical Population Genetics, Broad Institute, Cambridge, MA

07/2015 "Genome Editing and Gene Therapy"/ Invited Panel Member, Leerink Healthcare Insights Conference, Boston, Massachusetts

03/2016 "Ocular Genomics Institute"/ Session Organizer, Medical Population Genetics, Broad Institute, Cambridge, MA

03/2016 "Genetics of and Gene Therapies for Inherited Retinal Degenerations"/ Plenary Talk, Medical Population Genetics, Broad Institute, Cambridge, MA

04/2018 "Gene Therapies for Mendelian Diseases"/ Session Organizer, Medical Population Genetics, Broad Institute, Cambridge, MA

National

05/1995 "Vascular Endothelial Growth Factor in Retinopathy of Prematurity" / Plenary Talk
Department of Biochemistry, University of Wisconsin-Madison

11/1995 "Vascular Endothelial Growth Factor in Retinopathy of Prematurity" / Plenary Talk
Department of Biology, Bowdoin College

08/1998 "ROP and RP: A Tale of Two Diseases" / Plenary Talk
Scheie Eye Institute, University of Pennsylvania School of Medicine, Philadelphia, Pennsylvania

10/1998 "ROP and RP: A Tale of Two Diseases" / Plenary Talk
Department of Ophthalmology, University of Washington, Seattle, Washington

04/2001 "Mutations in the Novel Photoreceptor Protein RP1 Cause Inherited Blindness" / Plenary Talk
Everson Lecture, Department of Biochemistry, University of Wisconsin-Madison, Madison, Wisconsin

04/2001 "The Use of Oligonucleotides to Produce Mice with Targeted Mutations in Photoreceptor Genes" / Plenary Talk
Vision Research Conference, Ft Lauderdale, Florida

05/2001 "Investigations of the Pathogenesis of Retinopathy of Prematurity" / Plenary Talk
Division of Newborn Medicine, Harvard Medical School, Boston, Massachusetts

06/2001 "The Use of Oligonucleotides to Produce Mice with Targeted Mutations in Photoreceptor Genes" / Plenary Talk
FASEB Conference on the Biology and Chemistry of Vision, Tucson, Arizona

04/2002 "How do Mutations in RP1 Cause Photoreceptor Cell Death." / Plenary Talk
Schepens Eye Research Institute, Boston, Massachusetts

05/2002 "Gene -Targeted and Transgenic Models of Retinitis Pigmentosa 1" / Plenary Talk
ARVO Symposium "Recent Advances in Transgenic Animal Models of Retinal Disease."
Fort Lauderdale, Florida

05/2002 "Use of Oligonucleotides and Single-Stranded DNA to Introduce Mutations into Mouse Embryonic Stem Cells." / Plenary Talk
ARVO Mini symposium "Oligonucleotide-mediated Gene Manipulation in the Eye." Fort Lauderdale, Florida

11/2003 “RP1: A MAP for Outer Segment Organization.” / Plenary Talk
Department of Ophthalmology, University of Michigan

12/2003 “RP1: A MAP for Outer Segment Organization.” / Plenary Talk
Department of Ophthalmology, University of Tennessee

01/2004 “RP1: A MAP for Outer Segment Organization.” / Plenary Talk
Department of Ophthalmology, University of Illinois, Chicago.

04/2005 “The Retinitis Pigmentosa 1 (RP1) Protein and the Cell Biology of Photoreceptor Outer Segments” / Plenary Talk
Department of Biology, Dartmouth College, Hanover, NH

05/2005 “The Retinitis Pigmentosa 1 (RP1) Protein is a Photoreceptor MAP Involved in Outer Segment Organization” / Plenary Talk
ARVO Symposium on Cellular Pathways in Photoreceptor Neurodegeneration, Fort Lauderdale, Florida

06/2005 “Animal Models for Retinitis Pigmentosa 1 (RP1)” / Plenary Talk
FASEB Research Conference on the Biology and Chemistry of Vision

11/2005 “Cilia and Vision: Insights from Investigation of Retinitis Pigmentosa 1” / Plenary Talk
Vanderbilt Eye Institute, Vanderbilt University Medical Center, Nashville, TN

05/2006 "Molecular Dissection of Pathogenic Mechanisms" / Plenary Talk
Association for Research in Vision and Ophthalmology meeting Ft. Lauderdale, FL

08/2006 "Quantitative Aspects of Therapies for Retinal Degenerations: Insights from Proteomic Analyses" / Plenary Talk
Moran Eye Center Symposium on Retinal Degenerations, University of Utah, Salt Lake City, Utah

09/2006 "Targeted Mouse Models of Retinal Degenerations" / Plenary Talk
"Experimental Tools in Model Systems of Translational Vision Research" Conference, Jackson Laboratories, Bar Harbor, Maine

05/2007 "The Proteome of the Photoreceptor Sensory Cilium Complex" / Plenary Talk
11th Annual Vision Research Conference, Ft. Lauderdale, Florida

10/2007 "Proteomic Analysis of a Mammalian Sensory Cilium, the Mouse Photoreceptor Outer Segment" / Distinguished Visiting Scholar
University of Oklahoma Health Sciences Center, Oklahoma City, Oklahoma

04/2008 "Novel Cilia Proteins from the Photoreceptor Sensory Cilium Proteome" / Plenary Talk
ARVO Symposium "Ciliary Proteins and Retinal Degeneration: New Perspectives and Future Directions," Ft. Lauderdale, Florida

09/2008 "Novel Cilia Proteins and Inherited Retinal Degenerations" / Robb-Petersen Lecturer in Pediatric Ophthalmology
Children's Hospital, Harvard Medical School, Boston, Massachusetts

01/2009 "Novel Cilia Proteins in Photoreceptor Biology and Disease" / Distinguished Speaker, Gerald Fishman Lecture Series in Vision Science
Department of Ophthalmology, University of Illinois at Chicago, Chicago, Illinois

03/2009 "Novel Photoreceptor Sensory Cilia Proteins and Inherited Retinal Degenerations" / Plenary Talk
Kellogg Eye Institute, University of Michigan, Ann Arbor, Michigan

05/2009 "Retinal Diseases and Animal Models" Plenary Talk
Association for Research in Vision and Ophthalmology meeting Ft. Lauderdale, FL

05/2009 "Novel Photoreceptor Sensory Cilia Proteins and Inherited Retinal Degenerations" / Plenary Talk
ARVO Symposium "Photoreceptor Cilium and Associated Retinal Diseases," Ft. Lauderdale, Florida

03/2010 "Biology and Diseases of Photoreceptor Sensory Cilia: Nothing Silly About Cilia" / Grand Rounds
Cole Eye Institute, Cleveland Clinic, Cleveland, Ohio

05/2010 "The Severity of Retinal Degeneration in *Rp1* Gene Targeted Mice is Dependent Upon Genetic Background" / Plenary Talk
ARVO Workshop "Genetic Modifiers in Inherited Ocular Disease," Ft. Lauderdale, Florida

05/2010 "Genetics of Retinal Dystrophies" Association for Research in Vision and Ophthalmology meeting Ft. Lauderdale, FL

05/2010 "History of Gene Therapy" / Plenary Talk
ARVO History Special Interest Group, Ft. Lauderdale, Florida

05/2010 "Age-Dependent Effects of *RPE65* Gene Therapy for Leber Congenital Amaurosis: A Phase 1 Dose-Escalation Trial" / Plenary Talk
ARVO Symposium "Therapy for Sight," Ft. Lauderdale, Florida

09/2010 "Gene Therapy for Inherited Retinal Degenerations" / Roger Johnson Lecturer in Ophthalmology
Children's Hospital and Regional Medical Center, Seattle Washington

09/2010 "Presentations and Molecular Bases of Inherited Retinal Degenerations" / Roger Johnson Lecturer in Ophthalmology
Children's Hospital and Regional Medical Center, Seattle, Washington

10/2010 "Photoreceptor Sensory Cilia and Inherited Retinal Degenerations" / Plenary Talk
Massachusetts Eye and Ear Infirmary,
Harvard Medical School, Boston, Massachusetts

10/2010 "The Pathogenesis of RNA Splicing Factor Retinitis Pigmentosa" / Plenary Talk
Kellogg Eye Institute, University of Michigan, Ann Arbor, Michigan

02/2011 "Novel Photoreceptor Sensory Cilia Proteins and Insights into Inherited Retinal Degenerations" / Plenary Talk
Genetics, Cell Biology and Anatomy Seminar Series, University of Nebraska Omaha

05/2011 "Identification and Characterization of Novel Photoreceptor Sensory Cilia Proteins" / Plenary Talk
ARVO Symposium "Intraflagellar Transport," Ft. Lauderdale, Florida

01/2012 "Photoreceptor Sensory Cilia and Retinal Degenerations" / Keynote Address
Vision Research Symposium, Center for Visual Sciences, University of California, Davis, California

05/2012 "RPI Interacting Proteins and the Mechanism if RPI Disease" / Plenary Talk
14th Annual Vision Research Conference, Ft. Lauderdale, Florida

05/2012 "*NMNAT1* Mutations Cause Leber Congenital Amaurosis at the LCA9 Locus" / Plenary Talk (abstract)
ARVO Late-Breaking Papers Session, Ft. Lauderdale, Florida

05/2012 "RNA Splicing Factor Retinitis Pigmentosa: / Plenary Talk
ARVO Symposium "RNA Editing in Retinal Health and Disease," Ft. Lauderdale, Florida

05/2012 "Genetics of Inherited Retinal Degenerations" / Plenary Talk
Richard Weleber Lecturer in Ophthalmology, Casey Eye Institute, Oregon Health Sciences University, Portland, Oregon

05/2012 "Gene Therapies for Inherited Retinal Degenerations" / Plenary Talk
Richard Weleber Lecturer in Ophthalmology, Casey Eye Institute, Oregon Health Sciences University, Portland, Oregon

06/2012 "NEI Challenge to Identify Audacious Goals: Establishing a National Vision Research Agenda" Plenary / Talk
NAEC Meeting, Bethesda, Maryland

11/2012 "Genetic Testing for Patients with Retinal Degenerations" / Plenary Talk
American Academy of Ophthalmology Annual Meeting Chicago, IL

12/2012 "Genetics of Inherited Retinal Degenerations: Genetic Diagnostic Testing and Novel Disease Gene Discovery" / Plenary Talk
University of Utah, Salt Lake City, Utah

01/2013 "Genetics of Inherited Retinal Degenerations: Challenges and Opportunities"/ Plenary Talk
John Hopkins University, Bethesda, MD

02/2013 "Molecular Therapy at the Gene Level" / Plenary Talk
NEI Audacious Goals Development Meeting , Bethesda, MD

09/2013 "Deciphering the Role of Complement and the Extracellular Matrix in Early Stage Macular Degeneration Using Mouse Genetics and Proteomics" / Distinguished Lecturer
Vision Science Seminar Series, State University of New York, Buffalo, New York

11/2013 "Genetics and Genomics of Inherited Retinal Degenerations"/ Plenary Talk, Department of Molecular and Human Genetics Seminar, Baylor College of Medicine, Houston, TX

02/2014 "Genetics and Genomics of Inherited Retinal Degenerations: Challenges and Opportunities"/ Plenary Talk, Storm Eye Institute, Medical University of South Carolina, Charleston, SC

03/2014 "Genetics and Genomics of Inherited Retinal Degenerations"/ Plenary Talk, Department of Ophthalmology, University of California San Francisco School of Medicine, San Francisco, CA

05/2014 "Next Generation Sequencing in Inherited Retinal Disease" /Plenary Talk, ARVO Symposium 2014, Orlando, FL

05/2014 "Novel Elements in the Human Retinal Transcriptome"/ Plenary Talk, ARVO Symposium 2014, Orlando, FL

06/2014 "Genetics and Genomics of Inherited Retinal Degenerations: Challenges and Opportunities"/ Robert Meyers Lecture, Penn State Hershey Eye Center, Hershey, PA

10/2014 "The Genetics of Inherited Retinal Degenerations: Challenges and Opportunities" / Plenary Talk, Illumina, San Diego, CA

02/2015 "Complement and the Extracellular Matrix in Early Macular Degeneration"/ Plenary Talk, Angiogenesis, Exudation, and Degeneration Meeting, Miami, FL

04/2015 "Gene Therapy in the Eye"/ Plenary Talk, Hurdles to Gene Therapy session, American Otological Society Meeting, Boston, MA

06/2015 "Complement and the Extracellular Matrix in Early Macular Degeneration"/Purnell Lecture, Department of Ophthalmology and Visual Sciences, Case Western Reserve University, Cleveland, OH

06/2015 "Genetics of Inherited Retinal Degenerations"/Purnell Lecture, Department of Ophthalmology and Visual Sciences, Case Western Reserve University, Cleveland, OH

11/2015 "Inherited Retinal Degenerations"/Invited Panel Member, Retina Sub-Specialty Day, American Academy of Ophthalmology Meeting, Las Vegas, Nevada

05/2016 "Alternative Delivery and *In Vivo* Genome Editing"/ Plenary Talk, ARVO Symposium 2016, Seattle, WA

12/2016 "Studies of the Genetic Causality of Inherited Retinal Degenerations" / Plenary Talk, Emory Eye Center, Emory University, Atlanta, GA

01/2017 "Retrograde IFT and Inherited Retinal Degenerations" / Plenary Talk, Biology and Therapy of the Ciliated Senses Symposium, Center for Vision Research, University of Florida, Gainesville, FL

05/2017 "Early Experience with the ReNeuron-Sponsored Clinical Trial of Subretinally Transplanted Human Retinal Progenitor Cells in Patients with Advanced Retinitis Pigmentosa"/ Plenary Talk, Retinal Cell and Gene Therapy Innovation Summitt, Baltimore, MD

05/2017 "RNA Sequencing for Novel Genetic Discovery"/ Plenary Talk, ARVO Symposium 2017, Baltimore, MD

08/2017 "Allele-Specific Genome Editing for Dominant Inherited Retinal Degenerations"/Plenary Talk, 5th Annual Re-writing Genomes: A New Era in Genome Biology, California Institute for Quantitative Biosciences at Berkeley and the Innovative Genomics Institute, Berkeley, CA

05/2018 "Novel Genetic Causality of Inherited Retinal Degenerations"/Keynote Address, Sequencing Finishing and Analysis in the Future Meeting, Santa Fe, NM

International

05/1998 "Vascular Endothelial Growth Factor in Retinal Neovascularization" / Keynote Address Council on Complications, German Diabetes Association Meeting, Lübeck, Germany

01/2000 "New Methods for Producing Animals Models of Inherited Retinal Degenerations" / Plenary Talk
International Congress of Eye Research, Santa Fe, New Mexico

10/2005 "The RP1 Protein and the Cell Biology of Photoreceptor Outer Segments" / Grand Rounds Clinical Genetics, Hospital for Sick Children, University of Toronto, Ontario, Canada

10/2005 “Animal Models for Inherited Retinal and Macular Degenerations” / Plenary Talk
Lloyd Morgan Lecture, Department of Ophthalmology, Hospital for Sick Children, University of Toronto

06/2006 "Proteomic Analysis of the Mouse Photoreceptor Sensory Cilium Shows that it is a Highly Complex Organelle" / Plenary Talk
Laboratory of Cellular and Molecular Pathophysiology of the Retina, Université Pierre et Marie Curie, Paris, France

10/2006 "Proteomic Analysis of the Photoreceptor Outer Segment, a Conserved and Complex Mammalian Sensory Cilium" / Plenary Talk
Symposium "Proteomics of the Eye: Successes and Challenges," International Congress of Eye Research, Buenos Aires, Argentina

03/2007 "Insights into Photoreceptor Cilia Biology from Quantitative Proteomic Analyses" / Plenary Talk
Leeds Institute of Molecular Medicine, St. James's University Hospital, Leeds, England

07/2009 "Novel Photoreceptor Sensory Cilia Proteins and Retinal Degenerations" / Plenary Talk
Institut für Zoologie, Johannes Gutenberg-Universität, Mainz, Germany

07/2009 "Novel Photoreceptor Cilia Proteins and Inherited Retinal Degenerations" / Plenary Talk
Institut de la Vision, Université Pierre et Marie Curie, Paris, France

08/2009 "Novel Photoreceptor Sensory Cilia Proteins and Inherited Retinal Degenerations" / Plenary Talk
Department of Human Genetics, Nijmegen Center for Molecular Life Sciences, Radboud University Medical Center, Nijmegen, The Netherlands

02/2010 "Physiopathology and Molecular Mechanisms of Retinitis Pigmentosa in Usher Syndrome" / Plenary Talk
TreatRush Meeting, Institut de la Vision, Université Pierre et Marie Curie, Paris, France

02/2012 “Understanding of Usher Retinal Pathophysiology by Multidisciplinary Approaches in *In Vivo* and *In Vitro* Models” / Plenary Talk
TreatRush Consortium Meeting, Rome, Italy

08/2013 "Genetics of and Gene Therapies for Inherited Retinal Degenerations” / Plenary Talk
School of Biomedical Sciences, Monash University, Melbourne, Australia

11/2013 “Genetic Analysis of an Usher 1 Cohort” / Plenary Talk
TreatRush Consortium Meeting, Paris, France

07/2014 “Genetic Diagnostic Testing for Usher Syndrome” / Plenary Talk
International Symposium on Usher Syndrome, Boston, Massachusetts

09/2014 “Ophthalmology and Visual Sciences at Harvard Medical School” / Plenary Talk
Department of Ophthalmology and Visual Sciences, Chinese University of Hong Kong, Hong Kong, China

09/2014 “Genetics of Inherited Retinal Degenerations” / Plenary Talk
Department of Ophthalmology and Visual Sciences, Chinese University of Hong Kong, Hong Kong, China

09/2014 “Harvard Medical School Department of Ophthalmology: Ocular Genomics Institute” / Plenary Talk. Department of Ophthalmology, Hong Kong University, Hong Kong, China

09/2014	"Genetics of Inherited Retinal Degenerations / Plenary Talk Zhongshan Ophthalmic Center, Sun Yat-Sen University, Guangzhou, China
09/2014	"Genetics and Genomics of Inherited Retinal Degenerations" / Plenary Talk Shanghai Eye and ENT Hospital, Shanghai Medical College of Fudan University, Shanghai, China
11/2015	"Studies of the Genetic Causality of Inherited Retinal Degeneration"/ Plenary Talk, Retina 2015 Conference, Fighting Blindness Ireland, Dublin, Ireland
09/2016	"Copy Number Variation Contributes Significantly to the Genetic Causality of Inherited Retinal Degenerations"/Plenary Talk, XVIIth International Symposium on Retinal Degeneration, Kyoto, Japan
10/2016	"From Gene Discovery to Clinical Trials for Inherited Retinal Diseases"/Invited Presentation, Clinical Spotlight Session on Gene Discovery, Genetic Counseling, and Clinical Care of Patients with Inherited Retinal Diseases, American Society of Human Genetics Meeting, Vancouver, Canada
01/2018	"Precision Medicine for Inherited Retinal Degenerations"/Plenary Talk. Krembil Research Institute, University of Toronto, Toronto, Canada
02/2018	"Progress Towards Therapies for Inherited Retinal Degenerations"/Keynote Talk, Retina International Meeting, Auckland, New Zealand
02/2018	"Experience with the ReNeuron-Sponsored Clinical Trial of Subretinally Transplanted Human Retinal Progenitor Cells in Patients with Advanced Retinitis Pigmentosa"/Plenary Talk, Retina International Meeting, Auckland, New Zealand
02/2018	"Novel/Non-coding Genetic Causes of Inherited Retinal Degenerations"/Plenary Talk, Retina International Meeting, Auckland, New Zealand

Report of Clinical Activities and Innovations

Current Licensure and Certification

1997	American Board of Ophthalmology (renewed 2017)
1999–2011	Pennsylvania Medical License
2011–	Massachusetts Medical License

Practice Activities

1996–1999	Ophthalmology	Department of Ophthalmology, Children's Hospital Boston	Ambulatory and surgical care, 2 days per week
1997–1999	Surgery	Department of Surgery Brigham and Women's Hospital	Neonatal screening, ½ day per week, NICU
2000–2011	Ophthalmology	Division of Ophthalmology Children's Hospital of Philadelphia	Ambulatory and surgical care, 1 day per week
2012–	Ophthalmology	Department of Ophthalmology Massachusetts Eye and Ear Infirmary	Ambulatory care, 1-2 days per week

Clinical Innovations

Identification the Retinitis Pigmentosa 1 (*RP1*) gene and its role in inherited retinal degenerations	We identified RP1 protein as a photoreceptor-specific microtubule-associated protein that is required for the correct formation of photoreceptor outer segments. More recently, we determined that mutations in *RP1* cause dominant disease via a dominant-negative mechanism, suggesting that gene augmentation therapy has potential benefit for *RP1* patients.
Generation of a photoreceptor sensory cilia (PSC) proteome database to facilitate identification of inherited retinal degeneration genes	We were among the first investigators to recognize the relevance of photoreceptor sensory cilia (PSC), which compose the photoreceptor outer segments, in inherited retinal degenerations (IRDs). Our laboratory performed the first comprehensive analysis of a mammalian PSC proteome, and we have successfully used this highly complex murine PSC proteome (which contains almost 2000 proteins) to identify novel IRD disease genes and additional candidate genes.
Clinical evaluation of gene augmentation therapy for Leber congenital amaurosis caused by mutations in the *RPE65* gene	Through my clinical practice at Children's Hospital of Philadelphia (CHOP), I was involved in clinical trials of gene augmentation therapy for Leber congenital amaurosis (LCA) caused by mutations in the *RPE65* gene. Results obtained from these trials indicate that gene augmentation therapy is both safe and effective for LCA, and may be applied to other inherited retinal degenerations.
Identification of *NMNAT1* mutations at the LCA9 locus as a novel LCA disease gene	Whole exome sequencing was used to investigate the genetic basis of LCA in patients whose disease was not caused by mutations in known LCA genes. This sequencing identified mutations in the *NMNAT1* gene as a novel cause of LCA.
Development of next-generation sequencing based genetic diagnostic testing for inherited eye disorders	We developed a CLIA-certified genetic diagnostic testing panel for patients with inherited eye disorders, including inherited retinal degenerations, optic atrophy and glaucoma. The test is based on selective exon capture and next-generation sequencing techniques. Validation studies show the test to be highly accurate and reproducible.

Report of Technological and Other Scientific Innovations

03/28/1989	United States Patent 4,816,417 DeLuca HF, Dame MC, Pierce EA. Assay for 1,25-dihydroxy vitamin D in sample containing vitamin D transport protein. An assay for 1,25-dihydroxy vitamin D is disclosed. One aspect of the invention involves adding pig receptor protein, radiolabeled, 1,25-dihydroxy vitamin D and biotinylated antibody capable of binding to the receptor to untreated blood serum. In performing a competitive binding assay, vitamin D transport protein, DBP, acts as a screen to minimize interference from related metabolites. A kit and an assay are disclosed.
06/28/2008	Invention Disclosure "*Efemp1*-R345W Knockin Mice"

Eric A. Pierce

Several lines of evidence indicate that alterations in the integrity of the extracellular matrix (ECM) are important contributors to the pathogenesis of macular degeneration. The mechanisms by which alterations in ECM components lead to macular degeneration are not understood. We generated *Efemp1*-R345W mice to investigate this question. The disorder Doyne Honeycomb Retinal Dystrophy/Malattia Leventinese (DHRD/ML) is caused by the R345W mutation in the *EFEMP1* gene (EGF-containing fibrillin-like extracellular matrix protein 1; also called Fibulin-3). The EFEMP1 protein is a member of the fibulin family of ECM proteins. The *Efemp1*-R345W mice develop deposits between Bruch's membrane and the RPE which resemble basal deposits. In addition, we detected evidence of complement activation in the RPE and Bruch's membrane of the mutant mice when basal deposits are present. The *Efemp1*-R345W mice thus present a useful model system for studying the pathogenesis of basal deposit formation, and the role of complement in this process.

05/08/2013 Invention Disclosure "Primary RPE cell culture system that recapitulates pathologic features of mice carrying the R345W mutation in Efemp1 and the requirement for complement C3."
Chari Fernandez-Godino, Donia L. Garland, Eric A. Pierce

The invention is an RPE primary cell culture system using RPE cells from an animal model of inherited macular dystrophy, Doyne Honeycomb Retinal Dystrophy/Malattia Leventinese (DHRD/ML). This disorder is caused by the R345W mutation in EFEMP1 and the hallmark of DHRD/ML is the presence of extensive drusen. The animals carry the R345W mutation in Efemp1 and develop basal deposits which are considered precursors to drusen. Basal deposits are also histopathological features of age-related macular degeneration. In the animal model we have shown a critical role for complement C3 in the formation of the basal deposits. The primary RPE cell cultures from the animal model (invention) develop deposits and the formation of deposits is dependent on the presence of a functional complement C3. These RPE cells thus provide a model to investigate the role of complement in basal deposit formation, and to test drugs designed to prevent or reduce deposit formation.

11/2014 Invention Disclosure and Patent Application "Panel-based Genetic Diagnostic Testing for Inherited Eye Diseases."
Eric Pierce, Mark Consugar, Xiaowu Gai, Daniel Navarro
Patent Application filed 11-2014

We developed a targeted enrichment and NGS approach for genetic diagnostic testing of patients with inherited eye disorders, including inherited retinal degenerations (IRDs), optic atrophy and glaucoma, with improved accuracy, sensitivity and specificity, reproducibility and repeatability and/or clinical sensitivity as compared to previously reported uses of NGS techniques for genetic diagnostic testing of patients with IRDs.

01/2015 Invention Disclosure "Gene augmentation therapy for patients with *PRPF31*-associated retinitis pigmentosa."
Eric Pierce, Michael Farkas, Maria Sousa

Mutations in genes that encode pre-RNA processing factors (PRPFs) are a common cause of dominant inherited retinal degeneration (IRD). Our work has shown that the primary retinal cell type affected in the PRPF forms of IRD is the retinal pigment epithelium (RPE). Further, genetic data indicate that mutations in *PRPF31* gene cause disease via haploinsufficiency. Gene augmentation therapy directed to RPE cells should thus be beneficial for decreasing vision loss from *PRPF31*-associated disease.

Report of Education of Patients and Service to the Community

Activities

02/2006	"Meet the Docs" information session on Retinitis Pigmentosa / Participant 2006 Day of Science Meeting, Foundation Fighting Blindness, Los Angeles, CA
01/2007	"Meet the Docs" information session on Retinitis Pigmentosa / Participant 2007 Day of Science Meeting, Foundation Fighting Blindness, Orlando, FL
02/2008	2008 Day of Science Meeting / Speaker Foundation Fighting Blindness, San Diego, CA
09/2008	Philadelphia Vision Seminar / Speaker Foundation Fighting Blindness, Philadelphia, PA
02/2009	"Meet the Docs" information session on Retinitis Pigmentosa / Participant 2009 Day of Science Meeting, Foundation Fighting Blindness, Orlando, FL
02/2010	"Meet the Docs" information session on Retinitis Pigmentosa / Participant 2010 Day of Science Meeting, Foundation Fighting Blindness, Los Angeles, CA
07/2010	6th Biannual LCA Family Conference / Co-host Foundation for Retina Research, Philadelphia, PA
04/2011	"Genetics of and Gene Therapy for Inherited Retinal Degenerations" / Presentation Philadelphia Chapter, Foundation Fighting Blindness, Devon, PA
09/2011	"Genetics of and Genetic Therapies for Inherited Retinal Degenerations" / Plenary Talk Massachusetts Chapter, Foundation Fighting Blindness, Boston, MA
10//2011	"The Genetics of Inherited Retinal Degenerations" / Presentation Massachusetts Chapter, Foundation Fighting Blindness, Boston, MA
12//2011	"Research in Retinal Degenerations: Genomics and Gene Therapy" / Presentation, Annual Trustee Meeting, Massachusetts Eye and Ear Infirmary, Boston, MA
04/2012	"Research in Retinal Degeneration: Genomics and Gene Therapy" / Plenary Talk Tsoi/Kobus-Massachusetts Eye and Ear Development
04/2012	"The Study of Inherited Retinal Diseases" / Presentation Massachusetts Lions Eye Research Fund, Inc. Board of Directors meeting
06/2012	"Gene Therapies for Inherited Retinal Degenerations" / Plenary Talk Bardet Biedl Syndrome Conference, Duke University, Durham, North Carolina
06/2012	"Rare Disease" / Panel Discussion Foundation Fighting Blindness Visions Conference, Minneapolis, Minnesota
06/2012	"Participating in a Clinical Trial" / Panel Discussion Foundation Fighting Blindness Visions Conference, Minneapolis, Minnesota
07/2012	"Genetics of LCA: Diagnostic Testing and Disease Gene Discovery" / Plenary Talk Foundation for Retina Research 2012 Family Conference, Philadelphia, PA

09/2012	"Ocular Genomics Institute"/ Plenary Talk Foundation Fighting Blindness, Boston, MA
10/2012	"Get the scoop on NEI's Audacious Goals Initiative"/ ARVO Webinar
02/2013	"Inherited Retinal Degenerations" / Plenary Talk Resident Lecture Series, Harvard University, Boston, MA
02/2013	"A Fireside Chat: Catching Up on Research Advances on Retinal Degenerative Diseases" / Plenary Talk Foundation Fighting Blindness Chapter Meeting, Belmont, MA
04/2013	"Foundation Fighting Blindness Clinical Research Institute" / Plenary Talk Foundation Fighting Blindness Symposium Boston, MA
04/2013	"Gene Discovery, Diagnostics and Therapy for Eye Disease in the Genomics Era" / Plenary Talk Massachusetts Eye and Ear Infirmary, Boston, Massachusetts
06/2013	"Update and Overview: Usher Syndrome"/ Panel Discussion Foundation Fighting Blindness Visions Conference, Baltimore, MD
06/2013	"Genetic Testing and Counseling"/ Panel Discussion Foundation Fighting Blindness Visions Conference, Baltimore, MD
06/2013	"The Doctor is In: Usher Syndrome"/ Panel Discussion Foundation Fighting Blindness Visions Conference, Baltimore, MD
07/2014	"Genetic Eye Disease (GEDi) Diagnostic Testing in Usher Syndrome"/Plenary Talk, International Symposium on Usher Syndrome, Boston, MA
07/2014	"Genetics: Gene Mutations and LCA" / Plenary Talk Foundation for Retina Research 2014 Family Conference, Boston, MA
07/2014	"*NMNAT1*, *RDH12* and *CRB1* Gene Therapy" / Plenary Talk Foundation for Retina Research 2014 Family Conference, Boston, MA
06/2015	"Update and Overview: Retinitis Pigmentosa"/ Panel Discussion Foundation Fighting Blindness Visions Conference, Baltimore, MD
06/2015	"The Doctor is In: Retinitis Pigmentosa"/ Panel Discussion Foundation Fighting Blindness Visions Conference, Baltimore, MD
06/2016	"Progress Towards Precision Medicine for Inherited Eye Disorders" / Plenary Talk, Annual Meeting, Massachusetts Eye and Ear Infirmary, Boston, Massachusetts
09/2017	"Genes and Gene Therapies: Delivering Hope for Restored Sight"/ Panel Discussion, Foundation Fighting Blindness Investing in Cures Summit, Chicago, IL

Report of Scholarship

Peer reviewed publications in print or other media

Research investigations

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10. **Pierce EA**, Avery RL, Foley ED, Aiello LP, Smith LE. Vascular endothelial growth factor/vascular permeability factor expression in a mouse model of retinal neovascularization. *Proceedings National Academy Sciences USA* 92: 905-909, 1995. PMID:7846076 PMCID:42729

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12. Robinson GS*, **Pierce EA***, Rook SL, Foley E, Webb R, Smith LE. Oligodeoxynucleotides inhibit retinal neovascularization in a murine model of proliferative retinopathy. *Proceedings National Academy Sciences USA*.93:4851-4856, 1996. PMID:8643492 PMCID:39368 (*Co-first authors)

13. **Pierce EA**, Foley ED, Smith LE. Regulation of vascular endothelial growth factor by oxygen in a model of retinopathy of prematurity. *Archives Ophthalmology* 114:1219-1928, 1996. PMID:8859081

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Narrative Report: As a clinician-scientist with expertise in pediatric ophthalmology and retinal disorders, I am dedicated to understanding the molecular mechanisms of inherited retinal degenerations (IRDs), and improving therapies for these conditions. I pursue these objectives using a combination of laboratory research (~60% effort), clinical research and practice (~15% effort), administrative leadership

(~15%), and teaching (~10%). These activities take place primarily at Massachusetts Eye and Ear (MEE), the primary teaching affiliate for the HMS Department of Ophthalmology, and extend globally through international organizations, partnerships, and collaborations.

Laboratory Research
IRDs collectively are a leading cause of blindness worldwide. These conditions are characterized by progressive dysfunction and death of retinal photoreceptor and pigment epithelial cells, and affect people of all ages. IRDs occur in non-syndromic and syndromic forms; the majority of syndromic IRDs relate to cilia dysfunction. Over 260 genetic subtypes of IRD have been identified to date. However, the identified genes account for only ~60% of cases, and the mechanisms by which the identified mutations cause photoreceptor cell death remain to be determined. Through my research program, I aim to define the genetic causality of IRDs, to elucidate the underlying disease pathogenesis, and to develop therapeutic interventions for these disorders.

My research has led to the discovery of several novel IRD disease genes, including *RP1*, *TTC21B*, *NMNAT1* and *IFT172*. The *RP1* gene encodes a photoreceptor-specific microtubule-associated protein that is required for the correct formation of photoreceptor outer segments. We determined that mutations in *RP1*, which are a common cause of dominant RP, cause disease via a dominant-negative mechanism, suggesting that gene augmentation therapy has the potential to be beneficial for *RP1* patients. Through our studies of *RP1*, our laboratory was among the first to recognize that outer segments of photoreceptor cells are specialized primary cilia. This is an important concept, and underlies the connection between retinal degeneration and other cilia disorders. To study photoreceptor sensory cilia (PSC) biology on a broader scale, we performed the first comprehensive proteomic analyses of a mammalian sensory cilium, mouse PSCs. The complete murine PSC proteome, which contains almost 2000 proteins, is one of the most complex reported organelle proteomes. By screening patients with cilia disorders for mutations in novel PSC genes, we identified *TTC21B* and *IFT172* as ciliopathy disease genes.

More recently, we identified *NMNAT1* as a novel LCA disease gene via exome sequencing of families without mutations in known IRD disease genes. *NMNAT1* mutations represent a novel pathophysiologic cause of LCA; *NMNAT1* encodes the nuclear isoform of nicotinamide mononucleotide adenylyltransferase, an enzyme required for nicotinamide adenine dinucleotide (NAD^+) biosynthesis. We are now investigating the mechanism by which mutations in *NMNAT1* cause retinal degeneration as a first step toward developing a therapeutic strategy for *NMNAT1*-related LCA.

In addition to searching for novel IRD disease genes, we are also investigating the roles of non-coding and structural genomic variation in IRDs. For example, we recently reported that copy number variants (CNVs) and non-coding mutations contribute significantly to the genetic causality of IRDs. In our analyses, we identified non-allelic homologous recombination (NAHR) as a potentially common cause of CNVs in IRDs. In support of this hypothesis, we constructed a genomic map of NAHR-prone regions and superimposed known IRD genes. This map shows that 35 IRD genes are likely to be affected by NAHR, 13 of which have not been associated with a CNV to date. We believe that identifying such recombination hotspots will help find missing genetic causality in IRD patients, and perhaps in patients with other Mendelian disorders.

Clinical Expertise and Innovation
I devote 15% of my time to clinical practice as a retinal degeneration specialist. This involves seeing patients on the Inherited Retinal Disorders (IRD) Service at MEE, which I direct. In addition to traditional electrophysiologic testing, we offer genetic diagnostic testing and genetic counseling to patients and families. To facilitate comprehensive genetic testing for patients with inherited eye disorders, we developed the Genetic Eye Disease (GEDi) test, which involves sequencing the 260+ genes

associated with IRDs and optic neuropathies. The GEDi test is now provided for patients at MEE and outside institutions through the CLIA-certified testing lab in the OGI Genomics Core. Mass. Eye and Ear is the first and only location in the northeast to offer this service.

My clinical activities also include clinical research, in the context of the Berman-Gund Laboratory and IRD service. The Berman-Gund lab has a collection of DNA samples and longitudinal clinical data for over 6,000 families with inherited retinal degenerations who have been cared for at MEE. We are now working to investigate the different types of genetic causality of disease in this cohort. My clinical research at MEE builds on my experience at Children's Hospital of Philadelphia (CHOP) and the University of Pennsylvania. At CHOP, I helped establish a Pediatric Center for Retinal Degenerations to provide clinical care for patients and families with inherited retinal disorders. This center conducted one of the first successful trials of gene therapy, in which adeno-associated virus-mediated gene augmentation was used to treat children and adults with severe early onset retinal degeneration due to mutations in the *RPE65* gene. Results from this and other trials established the safety and efficacy of gene therapy for this form of IRD. I am optimistic that gene therapy can be applied to many other forms of IRD, and am working via the Ocular Genomics Institute (OGI) and the Berman-Gund Laboratory to help realize this goal. We are now participating in several clinical trials of gene therapy and one clinical trial of stem cell therapy for IRDs; I am the principal investigator for two of these studies.

Teaching

I have been an active teacher throughout my career. This includes teaching medical students, residents, and fellows when I see patients. My teaching activities also include mentoring medical students, graduate students and postdoctoral fellows through daily supervision of their research projects in my lab. A number of my former trainees have developed their own research programs in academia and industry. For example, Dr. Qin Liu is an Assistant Professor of Ophthalmology at Harvard, Dr. Catherine Cukras is a staff physician in the Clinical Trials Branch at the National Eye Institute, and Dr. Michael Farkas is now an Assistant Professor of Ophthalmology at the University at Buffalo. I am also an active participant in the HMS Department of Ophthalmology Faculty Mentoring Program, and formally mentor 5 junior faculty members. Outside of MEE, I served the international vision research community as Chair of the Scientific Advisory Board (SAB) of the Foundation Fighting Blindness (FFB), the leading non-governmental supporter of research on retinal degenerative disorders, between 2005 and 2014. As Chair of the SAB, I provided mentorship on a national level though a multi-level career development program for clinician-scientists interested in IRDs.

Administration and Institutional Service

In addition to the IRD service, I also direct the Ocular Genomics Institute (OGI) and serve as part of the Research Leadership group for Ophthalmology and the Research Steering Committee for MEE. The goal of the OGI is to translate the promise of precision medicine into clinical care for patients with inherited eye disorders. In the past 7 years, I have built the OGI into a successful, collaborative effort that is working to achieve this goal via a combination of laboratory based translational research, application of modern genomic technologies to improve clinical genetic diagnosis and identification of novel genetic causality, clinical research directed towards clinical trials of novel genetic therapies, and provision of state-of-the-art clinical care for patients with ophthalmic genetic disorders. The OGI also provides access to advanced genomics and gene therapy techniques to vision researchers across Harvard, and includes training programs in ophthalmic genetics for medical students, graduate students and clinical fellows.