Harvard Medical School Curriculum Vitae

Name: Qin Liu

Education

09/83-06/88	M.D.	Medicine	Xuzhou Medical College, China
09/93-05/98	Ph.D.	Molecular Ophthalmology	Peking Union Medical College, China

Postdoctoral Training

09/88-08/93	Resident	Ophthalmology	Xuzhou Medical College Hospital, China
06/98-08/99	Postdoctoral Fellow	Ocular Pharmacology (Dr. George Chiou)	Institute of Ocular Pharmacology, Texas A & M University
09/99-06/02	Postdoctoral Fellow	Molecular Ophthalmology (Dr. Eric Pierce)	Scheie Eye Institute, University of Pennsylvania

Faculty Academic Appointments

07/02-06/06	Research Associate	Ophthalmology	School of Medicine, University of Pennsylvania
07/06-08/11	Research Assistant Professor	Ophthalmology	School of Medicine, University of Pennsylvania
09/11-08/13	Lecturer	Ophthalmology	Harvard Medical School
09/13-	Assistant Professor	Ophthalmology	Harvard Medical School

Appointments at Hospitals/Affiliated Institutions

09/93-08/95	Assistant Physician	Ophthalmology	Peking Union Medical College Hospital, Beijing, China
09/95-05/98	Attending Physician	Ophthalmology	Peking Union Medical College Hospital, Beijing, China
09/11-08/13	Investigator	Berman-Gund Laboratory for the Study of Retinal Degenerations	Massachusetts Eye and Ear Infirmary, Boston, MA
09/11-08/13	Investigator	Ocular Genomics Institute	Massachusetts Eye and Ear Infirmary, Boston, MA
09/13-	Assistant Scientist	Berman-Gund Laboratory for the Study of Retinal Degenerations	Massachusetts Eye and Ear Infirmary, Boston, MA
09/13-	Assistant Scientist	Ocular Genomics Institute	Massachusetts Eye and Ear Infirmary, Boston, MA

Professional Societies

1998-	Association for Research in Vision and Ophthalmology	Member
1998-1999	Association of Ocular Pharmacology and Therapeutics	Member

2001-	Society for Neuroscience	Member
2017-	The American Society of Human Genetics	Member

Grant Review Activities

2007	*Ad hoc* member	Wellcome Trust
2015	*Ad hoc member*	Early Career Investigator Award Fight for Sight

Editorial Activities

Ad hoc Reviewer:

Progress in Retinal and Eye Research; Investigative Ophthalmology and Visual Science; Journal of Medical Genetics; BMC Research Notes; Molecular Vision; Cell Death and Differentiation; GENE; Journal of Cellular and Molecular Medicine; BMJ Open; Molecular Therapy; Scientific Report; Human Molecular Genetics; Experimental and Therapeutic Medicine

Honors and Prizes

1994	Zhou Zizhuan Foundation Award	Chinese Academy of Medical Sciences	Research
1995	Excellent Graduate Fellowship Award	Peking Union Medical College	Research
1996	Zhou Zizhuan Foundation Award	Chinese Academy of Medical Sciences	Research
1997	Excellent Graduate Fellowship Award	Peking Union Medical College	Research
2000	Retina Research Foundation/Lawrence Travel Fellowship Grant	Association for Research in Vision and Ophthalmology	Research
2006	The Jackson Laboratory Travel Award	The Jackson Laboratory	Research
2008	Young Investigator Award	The XIIIth International Symposium on Retinal Degenerations	
2008	ISER Young Investigator Fellowship Award	International Society for Eye Research	Research
2018	Young Investigator Award	The XVIIIth International Symposium on Retinal Degenerations	Research

Report of Funded and Unfunded Projects (past 3 years)

Funding Information

Past

09/14-8/15 Genetic diagnosis and identification of novel inherited retinal degeneration disease
Lions Grant Award
Co-PI ($78,000)
The goal of this project is to develop the procedures necessary to create an exome variant database for inherited eye diseases using WES and DNA samples from selected patients without any clinical evidence of eye disease.

01/15-12/15 CRISPR/Cas9-Mediated Gene Correction for Inherited Retinal Degeneration
Research to Prevent Blindness
PI ($38,156)
The goal of the proposed research is to test the use of CRISPR/Cas9-Mediated genome editing based strategies for treatment of inherited retinal degeneration.
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04/14-03/17 The Roles of TTC28 in Leber Congenital Amaurosis
Massachusetts Eye and Ear Infirmary
PI ($25,000/annual)
The goals of this project are to investigate the roles of the TTC28 genes in normal function of photoreceptor sensory cilia and inherited retinal degeneration.

04/15-03/17 Delivery of therapeutic proteins into mammalian cells
Bn10 Catalytic Steps Award
Co-Investigator ($81,000/annual)
The goal of the proposed to validate the potential therapeutic relevance of a newly developed protein delivery approach to deliver genome editing proteins into genetic deafness and blindness models in mice. If successful, this effort would provide a strong foundation for the *in vivo* delivery of therapeutic proteins, including ones with genome-editing abilities.

Current

07/99-07/23 Precision Medicine for Inherited Retinal Degenerations
NIH 5RO1 EY12910
Co-Investigator ($663,274 /annual)
The longterm goals of the proposed research are to help define the genetic causality of inherited retinal degenerations (IRDs), improve our understanding of disease mechanisms, and develop effective gene and genetically directed therapies for these disorders.

06/17-05/20 Development of CRISPR/Cas9-based genome editing approaches for RP1 associated autosomal dominant retinitis pigmentosa
Foundation Fighting Blindness
PI ($100,000/annual)
The goals of this project are to develop a set of CRISPR/Cas9-based gene editing tools for the treatment of RP1 disease *via* allele-specific disruption of the dominant alleles in the RP1 gene.

11/17-10/20 CRISPR/Cas gene editing for USH2A associated inherited retinal degeneration
Editas Medicine
PI ($569,398/annual)
The ultimate goal of the proposed research is to develop a CRISPR/Cas genome editing based therapeutic approach for retinal degeneration due to mutations in the *USH2A* gene, which is currently an unmet clinical need for 0.5-1.8 million of people worldwide. We will test the NHEJ-based exon-skipping and homology-independent targeted integration (HITI)-based mutation correction using CRISPR/Cas genome editing strategies to treat the most common *USH2A* mutations in cell-based and animal-based model systems.

02/18-02/20 Gene editing for RHO-P23H associated autosomal dominant retinitis pigmentosa

Casebia Therapeutics
PI ($209,128 /annual)
The goal of the proposed research is to develop a CRISPR/Cas genome editing based therapeutic approach for retinal degeneration due to mutations in the rhodopsin gene.

05/18-04/23 Using functional homology of RP1 isoforms to guide alternative therapeutic strategies
NIH 1R01EY028553
Sub-PI ($129,919/annual)
The goal of the proposed research is to test the hypothesis that lower expressing, novel nRP1 isoform shares functional similarity with the canonical RP1, and can be served as an alternative gene therapy strategy for RP1 associated inherited retinal disease.

Report of Local Teaching and Training

Laboratory and Other Research Supervisory and Training Responsibilities

2002-2005	Supervision of ophthalmology resident research fellow, University of Pennsylvania School of medicine	Daily mentorship of 3 research fellow
2002-2010	Supervision of pre-doctoral research fellow (undergraduate, medical and graduate students, University of Pennsylvania School of medicine	Weekly mentorship of 1-3 pre-doctoral trainees/year
2011-	Supervision of postdoctoral research fellows/ MEEI	Daily mentorship
2011-	Supervision of pre-doctoral research fellow (graduate students, technician pursued PhD), Harvard Medical School	Daily mentorship

Formally Supervised Trainees

2002-2003 Jason Skalet, MD / Private practice, Kaiser Oakland Medical Center, CA
Medical student: RP1 protein in photoreceptor bilogy. Co-authored one publication.

2004-2005 Cathy Cukras, MD, PhD/ Ophthalmologist, National Eye Institute
Ophthalmology resident: Identification of RP1 interacting proteins. Co-authored one presentation in 2005 annual ARVO meeting.

2004-2005 Nicole Benitah, MD / Private practice, Los Angeles, CA
Howard Hughes Medical Institutes Medical Student Fellow: Protein function of RP1. Co-authored one presentation in 2005 annual ARVO meeting.

2002-2004 Sara Achenbach, PhD / Field Appliction Sepcialist. Promega Corporation
Graduate Student: Identification of RP1 interacting proteins. Co-authored three abstracts in annual ARVO meeting.

2006 Gagan Sawhney, MD / Ophthalmology resident, Emory University
Medical Student: RP1 interacting protein.

2007-2008 Jonathan Weiner, MD / Ophthalmology resident, John Hopkins University
Undergraduate Student: Retinal degeneration in RP1 knockin mice. Applied for a fellowship grant.

2009 Ferry Kersten, PhD / Radboud University Nijmegen Medical Centre, The Netherlands
Graduate student: In vivo expression of cilia-related genes in photoreceptor cells.

2009-2011 Yue Cui / PhD candidate, University of Pennsylvania School of Medicine
Graduate Student: Ttc21b conditional knockout mice.

2011 Yafeng Li / MD, PhD candidate, University of Pennsylvania School of Medicine
Medical Student: The genetic mechanism of RP1 disease.

2011 Alan Moore / PhD candidate, University of Pennsylvania School of Medicine
Graduate Student: The novel photoreceptor sensory cilia proteins.

2011-2013 Jingfa Zhang, PhD, Associate Professor, Tongji University School of Medicine, China
Postdoctoral researcher: Wasf3 and Nim1 protein in photoreceptor biology. Co-authored one presentations in 2012 ARVO meeting and two manuscripts.

2012-2013 Conghui Zhang/ PhD candidate , Tongji University School of Medicine
Graduate student: Zebrafish model of retinal degenerative diseases. Co-authored one publication.

2012-2013 Chan Wu, MD / Ophthalmologist, Peking Union Medical College Hospital.
Research Fellow: Light dependent transport of proteins in photoreceptors.

2012-2013 Daniel Taub, Graduate Student, Boston University.
Technologist: Animal models of TTC21b form of ciliopathy. Co-authored one abstract and one publication.

2013-2014 Magdalena Staniszewska, Assistant Professor, Institute of Immunology and Experimental Therapy, Polish Academy of Sciences, Wroclaw, Poland.
Research Scientist: Disease Mechanisms underlying TTC21b form of ciliopathy and Gene therapy for RP1 disease. Co-authored two abstracts and one manuscript.

2014-2017 Mihoko Leon, Research Technician, Mass General Hospital
Research Technologist: Disease Mechanisms underlying TTC21b form of ciliopathy and Gene therapy for RP1 disease. Co-authored three abstracts and one manuscript.

2015-2017 Pingjuan Li, Scientist, Gemini Therapeutics, Cambridge, MA
Postdoc: Development of CRISPR/Cas9-based genome editing approaches for RHO associated autosomal dominant retinitis pigmentosa. Co-authored one paper.

Report of Regional, National and International Invited Teaching and Presentations

Local Invited Presentations

2001-2010 127th-136th Anniversary Research Meeting, Plenary Talk, Scheie Eye Institue, University of Pennsylvania, Philadelphia, Pennsylvania.

2015.5.28 Harvard Medical School Department of Ophthalmology International Mini-Symposium, Boston, Massachusetts.

Invited Presentations and Courses

Regional

2010.10 Invited speaker. "Disease mechanisms in RP1 from retinitis pigmentosa: Implications for therapy". Annual Lions Eye Research Seminar, Philadelphia, PA

National

2004.05 Invited speaker. "IMPDH1 - a candidate interacting protein of RP1". IMPDH workshop, University of Texas, Houston, TX

2015.05 Invited speaker. "The role of intraflagellar transport in the photoreceptor sensory cilium and retinal degeneration" ARVO Education Course, Denver, CO

2017.04 Invited speaker. "Gene editing for rhodopsin associated ADRP" 4th Annual Retinal Cell and Gene Therapy Innovation Summit, Baltimore, MD

International

2004.05	Invited speaker. "PMPDH1 – a novel interacting protein of RP1". PMPDH1 workshop, University of Texas, Houston, TX
2004.05	Invited speaker. "Proteomic analysis of photoreceptor cilium". Tongji University School of Medicine, Shanghai, China
2011.06	Invited speaker. "Genetic studies of inherited retinal degenerative diseases". Peking union medical college hospital international forum of ophthalmology, Beijing, China
2012.12	Invited speaker. "Genetic mechanisms of inherited retinal diseases". Tongji University School of Medicine, Shanghai, China
2015.07	Invited speaker. "Retinal ciliopathy". Wenzhou Medical University, Wenzhou, China
2015.12	Invited speaker. "Applications of next generation sequencing". Xuzhou Medical School, Xuzhou, China
2017.06	Invited speaker. "CRISPR/Cas gene editing for rhodopsin associated retinitis pigmentosa". National Taiwan University, Taipei, Taiwan
2017.09	Invited speaker. "Allele-specific CRISPR/Cas9 Genome Editing for Treating Dominant Retinitis Pigmentosa". The 12th Annual Meeting of the International Conference of Genomics. OGI, Shenzhen, China
2018.06	Invited speaker. "Potential applications of CRISPR/Cas9-based genome editing in the treatment of inherited eye disorders". Peking Union Medical College Hospital, Beijing, China
2018.06	Invited speaker. "CRISPR/Cas9-based gene therapy of inherited retinal degenerations: perspectives and challenges". The 1st International Conference of Genomics on Eyes (ICGEYE). Shenyang, China

Report of Clinical Activities and Innovations

Current Licensure and Certification

1993	Chinese Board of Ophthalmology

Practice Activities

1988-1993	Ophthalmology	Xuzhou Medical School Hospital	Clinic and surgical care, every day
1993-1998	Ophthalmology	Peking Union Medical College Hospital	Clinic and surgical care, 2 days per week

Clinical Innovations

Identification the role of Retinitis Pigmentosa 1 (RP1) in inherited retinal degenerations	I 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* gene cause dominant disease via a dominant-negative mechanism, suggesting that gene augmentation therapy has potential benefit for *RP1* patients.
Identification of NMNAT1 mutation at the LCA9 locus as a likely cause of LCA	Whole exome sequencing was used to investigate the genetic basis of LCA in two pediatric patients whose disease was not caused by known LCA genes. Exome sequencing identified that mutation in *NMNAT1* at the LCA9 locus is the cause of LCA.
Identification of	Whole exome sequencing identified that mutation in IFT172 is the cause of

IFT172 mutation as a cause of RP	isolated isolated retinal degeneration and Bardet-Biedl syndrome.
Development of CRISPR/Cas9 gene editing for RHO associated RP	We performed a proof-of-concept study showing that allele-specific CRISPR-Cas9 genome editing approach can reduce the progression of retinal degeneration in Rhodopsin-P23H Associated Dominant Retinitis Pigmentosa.

Report of Technological and Other Scientific Innovations

Generation of a photoreceptor sensory cilia (PSC) proteome database to facilitate identification of IRD 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). I 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.
Development of lipid-mediated delivery approach of deliver negatively charged protein into retinal cells	In collaboration with Dr. David Liu's group in Harvard University, we were the first investigators to use cationic lipids to effectively deliver negatively charged functional protein into a variety of retinal cells, including photoreceptor cells. This should serve as a basis for designing novel systems for a controlled and scalable delivery of the therapeutic proteins, including genome-editing proteins, into the retinal cells, with promising implications for the treatment of retinal diseases affecting all kinds of cells.

Report of Scholarship

Peer reviewed publications in print or other media

Research Investigations

1. Mou L, Wu J, **Liu Q**: Leber's hereditary optic neuropathy: Analgsis of 6 genealogie. *Acta Academiae Medicinae Xuzhou*. 13(4): 280-282, 1993.

2. Mou L, **Liu Q**, Zhao YM: Exudative retinal detachment after cryosurgery in rhagmatogenous retinal detachment. *Jiangsu Medical Journal* 19: 77-78, 1993.

3. Mou L, **Liu Q**: Modification of external dacryocystorhinostomy in recurrent chronic dacryocystitis. *Xuzhou Medical Journal* 3: 36, 1993.

4. **Liu Q**, Mou L, Li MX: Management of ocular hypotension of contusion. *Acta Academiae Medicinae Xuzhou* 15(1): 58-59, 1995.

5. **Liu Q**, Zhang MF, Ai FR, Hu TS: Treatment of acute optic neuritis with high dose of prednisone. *Acta Academiae Medicinae Sinicae* 20(3): 230-234, 1998.

6. **Liu Q**, Zhang MF, Qiu CC, Hu TS: Association of HLA-DQA1 and -DQB1 alleles with Vogt-Koyanagi-Harada syndrome in Han Chinese population. *Chin J Ophthal* 35: 210-215, 1999.

7. **Liu Q**, Zhou Y H, Xuan B, Chiou G C, Okawara T: Effects of interleukin-1 blockers on corneal fibroblast proliferation in vitro and ocular inflammation in vivo. *J Ocul Pharmacol Ther* 16: 81-96, 2000.

8. Chiou G C, Xuan B, **Liu Q**, Yamasaki T, Okawara T: Inhibition of interleukin-1-induced uveitis and corneal fibroblast proliferation by interleukin-1 blockers. *J Ocul Pharmacol Ther* 16: 407-18, 2000.

9. Zhou W, **Liu Q**, Zhang MF, Du H, Ye JJ, Jiang RX, Ai FR, Hu TS: Treatment of acute optic neuritis with glucocorticoid. *Chin Ophthalmic Res* 18: 51-53, 2000.

10. Bearuregard C, **Liu Q**, Chiou G C: Effects of nitric oxide donors and nitric oxide synthase substrates on ciliary muscle contracted by carbachol and endothelin for possible use in myopia prevention. *J Ocul Pharmacol Ther* 17(1): 1-9, 2001.

11. **Liu Q**, Zhou J, Daiger SP, Farber DB, Heckenlively JR, Smith JE, Sullivan LS, Zuo J, Milam AH, Pierce EA: Identification and subcellular localization of the RP1 protein in human and mouse photoreceptors. *Invest Ophthalmol Vis Sci* 43(1): 22-32, 2002.

12. Gao J, Cheon K, Nusinowitz S, **Liu Q**, Bei D, Atkins K, Azimi A, Daiger SP, Farber DB, Heckenlively JR, Pierce EA, Sullivan LS, Zuo J: Progressive photoreceptor degeneration, outer segment dysplasia, and rhodopsin mislocalization in mice with targeted disruption of the retinitis pigmentosa-1 (Rp1) gene. *Proc Natl Acad Sci U S A* 99(8): 5698-703, 2002.

13. Zhang M, **Liu Q**, Min H, Du H, Hu TS: Vogt-Koyanagi-Harada syndrome: glucocorticoid therapy and visual prognosis. *Chin J Ophthalmol* 38(4): 200-203, 2002.

14. Pierce EA, **Liu Q**, Igoucheva O, Omarrudin R, Ma H, Diamond SL, Yoon K: Oligonucleotide-directed single-base DNA alterations in mouse embryonic stem cells. *Gene Ther* 10(1): 24-33, 2003.

15. Bowne SJ, Daiger SP, Malone KA, Heckenlively JR, Kennan A, Humphries P, Hughbanks-Wheaton D, Birch DG, **Liu Q**, Pierce EA, Zuo J, Huang Q, Donovan DD, Sullivan LS: Characterization of RP1L1, a highly polymorphic paralog of the retinitis pigmentosa 1 (RP1) gene. *Mol Vis* 9: 129-37, 2003.

16. **Liu Q**, Lyubarsky A, Skalet JH, Pugh EN, Pierce EA: RP1 is required for the correct stacking of outer segment discs. *Invest Ophthalmol Vis Sci* 44(10): 4171-83, 2003.

17. Ma H, **Liu Q**, Diamond SL, Pierce EA: Mouse embryonic stem cells efficiently lipofected with nuclear localization peptide result in a high yield of chimeric mice and retain germline transmission potency. *Methods* 33(2): 113-20, 2004.

18. **Liu Q**, Zuo J, Pierce EA: The retinitis pigmentosa 1 protein is a photoreceptor microtubule-associated protein. *J of Neurosci* 24(29): 6427-36, 2004.

19. Bowne SJ, **Liu Q***, Sullivan LS, Zhu J, Spellicy CJ, Rickman CB, Pierce EA, Daiger SP: Why do mutations in the ubiquitously expressed housekeeping gene IMPDH1 cause retina-specific photoreceptor degeneration? *Invest Ophthalmol Vis Sci* 47(9): 3754-65, 2006.

(* co-first authors)

20. **Liu Q**, Tan G, Levenkova N, Li T, Pugh EN, RJ, Speicher DW, Pierce EA: The proteome of the mouse photoreceptor sensory cilium complex. *Mol Cell Proteomics* 6(8): 1299-317, 2007.

21. Spellicy CJ, Daiger SP, Sullivan LS, Zhu J, **Liu Q**, Pierce EA, Bowne SJ: Characterization of retinal inosine monophosphate dehydrogenase 1 in several mammalian species. *Mol Vis* 3(13): 1866-72, 2007.

22. **Liu Q**, Saveliev A, Pierce EA: The severity of retinal degeneration in Rp1h gene-targeted mice is dependent on genetic background. *Invest Ophthalmol Vis Sci* 50(4): 1566-74, 2009.

23. Westfall JE, Hoyt C, **Liu Q**, Hsiao Y, Pierce EA, Page-McCaw PS, Ferland RJ: Retinal degeneration and failure of photoreceptor outer segment formation in mice with targeted deletion of the Joubert syndrome gene, Ahi1. *J Neurosci* 30(26): 8759-68, 2010.

24. Otto EA, Hurd T, Airik R, Chaki M, Zhou W, Stoetzel C, Patel S, Levy S, Ghosh A, Murgas-Zamalloa C, van Reeuwijk J, Letteboer SJF, Sang L, Geles RH, **Liu Q**, Coene KLM, Estrada-Cuzcano A, Collin RWJ, McLaughlin HM, Held S, Kasanuki JM, Ramaswami G, Conte J, lopez I, Washburn J, MacDonald J, Hu J, Yamashita Y, Maher E, Guay-Woodford L, Neumann H, Obermuller N, Koenekoop RK, Bergmann C, Bei X, Lewis RA, Katsanis N, Lopes V, Williams DS, Cyons RH, Dang CV, Brito DA, Dias MB, Zhang X, Cavalcoli JD, Nurnberg G, Nurnberg P, Pierce EA, Jackson P, Antignac C, Saunier S, Roepman R, Dollfus H, Khanna H, Hildebrandt F: Candidate exome capture reveals mutation of SDCCAG8 as causing a retinal- renal ciliopathy. *Nat Genet* 42:840-850, 2010.

25. **Liu Q**, Zhang Q, Pierce EA: Photoreceptor sensory cilia and inherited retinal degeneration. *Adv Exp Med Biol* 664: 223-32, 2010.

26. Bowne SJ, Sullivan LS, Koboldt DC, Ding L, Fulton R, Abbott RM, Sodergren EJ, Birch DG, Wheaton DH, Heckenlively JR, **Liu Q**, Pierce EA, Weinstock GM, Daiger SP: Identification of disease-causing mutations in autosomal dominant retinitis pigmentosa (adRP) using next-generation DNA sequencing. *Invest Ophthalmol Vis Sci* 52(1): 494-503, 2011.

27. Davis EE, Zhang Q, **Liu Q**, Diplas B, Davey LM, Hartley J, Stoetzel C, Muzny DM, Young AC, Wheeler DA, Cruz P, Morgan M, Lewis LA, Cherukuri P, Maskeri B, Mullikin JC, Blakesley RW, Lewis RA, Bergmann C, Otto EA, Antignac C, Saunier S, Scambler PJ, Beales PL, Gleeson JG, Hildebrandt F, Maher ER, Attié- Bitach T, Dollfus H, Johnson CA, Green ED, Gibbs RA, Pierce EA, Katsanis N: Mutations in *IFT139 (TTC21B)* contribute both causal and modifying alleles across the ciliopathy spectrum. *Nat Genet* 189-96, 2011.

28. Falk MJ, Zhang Q, Nakamaru-Ogiso E, Kannabiran C, Fonseca-Kelly Z, Chakarova C, Audo I, Mackay DS, Zeitz C, Borman AD, Staniszewska M, Shukla R, Palavalli L, Mohand-Said S, Waseem NH, Jalali S, Perin JC, Place E, Ostrovsky J, Xiao R, Bhattacharya SS, Consugar M, Webster AR, Sahel JA, Moore AT, Berson EL, **Liu Q,** Gai X, Pierce EA: NMNAT1 mutations cause Leber congenital amaurosis. *Nat Genet* 44(9):1040-5, 2012.

29. Zhang Q, **Liu Q**, Austin C, Drummond I, Pierce EA: Knockdown of ttc26 disrupts ciliogenesis of the photoreceptor cells and the pronephros in zebrafish. *Mol Biol Cell* 23(16):3069-78, 2012.

30. **Liu Q,** Collin RW, Cremers FP, den Hollander AI, van den Born LI, Pierce EA: Expression of Wild-Type Rp1 Protein in Rp1 Knock-in Mice Rescues the Retinal Degeneration Phenotype. *PLoS One* 7(8): 43251, 2012.
31. Song D, Grieco S, Li Y, Hunter A, Chu S, Zhao L, Song Y, DeAngelis RA, Shi LY**, Liu Q**, Pierce EA, Nishina PM, Lambris JD, Dunaief JL: A murine rp1 missense mutation causes protein mislocalization and slowly progressive photoreceptor degeneration. *Am J Pathol* 184 (10):2721-9, 2014.
32. Bujakowska KM, Zhang Q, Siemiatkowska AM, **Liu Q**, Place E, Falk MJ, Consugar M, Lancelot ME, Antonio A, Lonjou C, Carpentier W, Mohand-Saïd S, den Hollander AI, Cremers FP, Leroy BP, Gai X, Sahel JA, van den Born LI, Collin RW, Zeitz C, Audo I, Pierce EA: Mutations in IFT172 cause isolated retinal degeneration and Bardet-Biedl syndrome. *Hum Mol Genet.* 2014 PMID:25168386
33. Consugar MB, Navarro-Gomez D, Place EM, Bujakowska KM, Sousa ME, Fonseca-Kelly ZD, Taub DG, Janessian M, Wang DY, Au ED, Sims KB, Sweetser DA, Fulton AB, **Liu Q**, Wiggs JL, Gai X, Pierce EA: Panel-based Genetic Diagnostic Testing for Inherited Eye Diseases is Highly Accurate and Reproducible and More Sensitive for Variant Detection Than Exome Sequencing. *Genetics In Medicine.* 17(4):253-61, 2015 PMID: 25412400, PMCID: PMC4572572
34. Zhang C, Zhang Q, Wang F, **Liu Q.** Knockdown of poc1b causes abnormal photoreceptor sensory cilium and vision impairment in zebrafish. *Biochemical and biophysical research communications*. 465(4):651-7, 2015 PMID: 26188096, PMCID: PMC4601574
35. Taub DG, **Liu Q**. The Role of Intraflagellar Transport in the Photoreceptor Sensory Cilium. *Adv Exp Med Biol.* 2016; 854:627-33. PMID: 26427468
36. Greenwald SH, Charette JR, Staniszewska M, Shi LY, Brown SD, Stone L, **Liu Q**, Hicks WL, Collin GB, Bowl MR, Krebs MP, Nishina PM, Pierce EA. Mouse Models of NMNAT1-Leber Congenital Amaurosis (LCA9) Recapitulate Key Features of the Human Disease. The American journal of pathology. 2016; 186(7):1925-38. PMID: 27207593, PMCID: PMC4929402
37. Li P, Kleinstiver B, Leon MY, Prew MS, Navarro-Gomez D, Greenwald SH, Pierce EA, Joung JK, and **Liu Q**. Allele-Specific CRISPR-Cas9 Genome Editing of the Single-Base P23H Mutation for Rhodopsin-Associated Dominant Retinitis Pigmentosa. *The CRISPR Journal*. 2018; 1(1): 55-64.
38. Gupta PR, Pendse N, Greenwald SH, Leon M, **Liu Q,** Pierce EA, Bujakowska KM. Ift172 conditional knock-out mice exhibit rapid retinal degeneration and protein trafficking defects. Human molecular genetics. 2018; 27(11): 2012-2024. PMID: 29659833 PMCID: PMC5961092

Non-peer reviewed scientific or medical publications/materials in print or other media

Chapters

Achenbach S, **Liu Q** and Pierce EA. : The RP1 Gene and Protein in Photoreceptor Biology. In Williams D (ed): Cell Biology and Disease of the Outer Retina, World Scientific Press, River Edge NJ Page: 223-267, 2004.

Bujakowska K, **Liu Q** and Pierce EA. Photoreceptor Cilia and Retinal Ciliopathies. Cilia. Cold Spring Harb Perspect Biol. 2017 Oct 3; 9(10). PMID: 28289063

Thesis

Liu, Q. Association of HLA-DQA1 and -DQB1 alleles with Vogt-Koyanagi-Harada syndrome in Han Chinese population. Ph.D. Thesis, Peking Union Medical College, Beijng, 1998.

Abstracts, Poster Presentations and Exhibits Presented at Professional Meetings (last 3 years)

1. **Liu Q,** Rees H, Sousa M, Leon M, Zuris J, Liu D, Pierce EA. Effective delivery of recombinant protein into retinal cells by cationic lipid. ARVO 2016.

2. **Liu Q**, Li P, Kleinstiver BP, Joung KJ, and Pierce EA. CRISPR/Cas-mediated genome editing for RHO-associated dominant RP. RD2016. XVIIth International Symposium on Retinal Degeneration. Kyoto, Japan.

3. Li P, Kleinstiver B, Leon MY, Pierce EA, J Joung K, **Liu Q**. In vivo allele-specific CRISPR/Cas gene editing in the rhodopsin P23H knockin mouse model. ARVO 2017

4. Pendse N, Sebastian Gloskowski S, Maeder M, Pierce EA, **Liu Q.** Evaluation of therapeutic potential of human USH2A gene lacking exon 13 (USH2A-ΔEx13) for restoring ciliogenesis. ASGCT 2018.

5. Pendse N, Veronica Lamas V, Gloskowski S, Maeder M, Chen Z, Pierce EA, **Liu Q.** In vitro functional assessment of potential therapeutic approaches for USH2A associated diseases. RD 2018. XVIIIth International Symposium on Retinal Degeneration. Killarney, Ireland.