

Claudio Punzo, Ph.D.

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Education

- Ph.D., (Cell Biology), University of Basel, Basel, BS, Switzerland 2001
Thesis Title: Functional analysis of *Pax-6* genes during eye development and evolution.
Advisor: Dr. Walter J. Gehring
- B.S., (Cell Biology), Biozentrum University of Basel, Basel, Switzerland 1997
Thesis Title: Regulation of temperature-induced heat shock reaction of *Drosophila ambigua* and *Drosophila melanogaster*.
Advisor: Dr. Walter J. Gehring

Postdoctoral Training

- Postdoctoral Fellow/Scholar 2002-2010
Supervisor: Constance L. Cepko
Department of Genetics, Harvard Medical School, Boston, MA

Academic Appointments

- Associate Professor 6/2021-present
Vice Chair of Research
Department Ophthalmology & Visual Sciences
University of Massachusetts Medical School, Worcester, MA
- Tenure Promotion April 2021
Department Ophthalmology & Visual Sciences
University of Massachusetts Medical School, Worcester, MA
- Affiliation: Horae Gene Therapy Center 2019-present
University of Massachusetts Medical School
- Associate Professor 2017-present
Department Ophthalmology & Visual Sciences
University of Massachusetts Medical School, Worcester, MA
- Affiliation: Department of Neurobiology 2015-present
University of Massachusetts Medical School
- Assistant Professor 2010-2017
Department Ophthalmology & Visual Sciences,
University of Massachusetts Medical School, Worcester, MA

Honors and Awards

- Long-Term Fellowship (EMBO: European Molecular Biology Organization) 2002-2004
- Award for best Poster presentation (USGEB Young Investigator Meeting, Lausanne, Switzerland) 2001

Updated:

Recipient of Research to Prevent Blindness / American Macular Degeneration
Foundation Catalyst Award for Innovative Research Approaches for AMD

2023

Educational Activities

Educational Leadership, Administration and Service

- Course Coordinator, BBS 782: Tutorial: Bases of Brain Diseases. 2018-2020,
2022-present
- Course Coordinator, Ophthalmology: Vision Seminar Series. 2016-present

Teaching Activities in Programs and Courses

- BBS 782 Tutorial: Bases of Brain Diseases. Role: One two hours session with approximately 10-15 students. Session Title: 2011-2019: Retinitis Pigmentosa; 2020-present: AMD 2011, 2012,
2014, 2016,
2018-2020,
2022, 2024
- BBS 760: Introduction into Neuroscience. Role: One hour lecture on visual processing from retina to brain with approximately 10-15 students. Session Title: Visual Processing. (Course is held yearly) 2014-2021
- BBS 820: System and Circuit Neuroscience. Role 3 classes of 90 min. each as part of a full semester course. Approximately 10 students per year. 2022, 2023
- Foundation Class (Gene Therapy Course). Role: Paper discussion ~ 1.5h 2022,
- Summer RAPS: Two hours of paper discussion for new incoming students. Approximately 10-15 students per session. 2012, 2013
- FM 201: Brain Course for 2nd year medical students. Role: One lecture of 1 hour on the visual system for the entire medical student class. 2014
- Summer Course on Gene Therapy held at the Gene Therapy Center. Role: Annual lecture on Ocular Gene Therapy: 15 students/post-docs. 2011-2014
- University of Basel (Basel, Switzerland): Teaching assistant: Lab instructor for upper division undergraduate biology course. Led course discussion section, held review sessions, designed laboratory experiments, corrected exams, held lectures for Dr. W.J. Gehring. Class size: 40 students. 1998-2000

Research Education

Graduate Student Education

- Member, Neuroscience Program 2010-present
- Member, Interdisciplinary Graduate Program 2010-present

-Qualifier Exam Committee

- Michael Whalen (M.S.) 05/2011
- Subhadeep Ray (Ph.D.) 06/2011
- Alisha Gruntman (Ph.D.) 03/2012
- Ozge Yildiz (Ph.D.) 04/2013
- Leticia Fridman (Ph.D.) 06/2013
- Neda Baniyasi (Ph.D.) (UMass Lowell) 09/2016
- Dominic Gessler (Ph.D.) 10/2018
- Huang Wenjia (Ph.D.) 06/2020
- Jillian Gallagher (Ph.D.; Chair) 06/2020

- Mohona Gupta (Ph.D.) 7/2020
- Abigail McElroy (Ph.D.; Chair) 4/2023
- Motahareh Arjomandnejad (Ph.D.; Chair) 1/2024
- Ruixuan Xiao (Ph.D.; Chair) 3/2024
- Anoushka Lotun (Ph.D.; Chair) 4/2025

-Thesis Defense Exam Committee (DEC):

- Michael Whalen (M.S.) 06/2011
- Rory Coffey (Ph.D.) (Brandeis University) 08/2015
- Sourav Chouhury (Ph.D.) 01/2016
- Diane Golebiowski (Ph.D.) 10/2016
- Alisha Gruntman (Ph.D.) 11/2016
- Neda Baniyadi (Ph.D.) (Umass Lowell) 02/2017
- Ozge Yildiz (Ph.D.) 03/2019
- Dominic Gessler (Ph.D.) 10/2020
- Anastasia Schultz (Ph.D.) 08/2023
- Mohona Gupta (Ph.D, Chair) 03/2024

-Thesis TRAC committee:

- Alisha Gruntman (Ph.D.) 2012-2016
- Diane Golebiowski (Ph.D.) 2012-2016
- Ozge Yildiz (Ph.D.) 2013-2019
- Dominic Gessler (Ph.D.) 2018-2020
- Mohona Gupta (Ph.D.; Chair) 2021-2024
- Anastasia Schultz (Ph.D.), (Montana State University) 2021-2023
- Colin King (M.S.), (Montana State University) 2021-2023
- Thomas Leland (Ph.D.; Chair) 2021-present
- Abigail McElroy (Ph.D.; Chair) 2023-present
- Motahareh Arjomandnejad (Ph.D.; Chair) 2024-present
- Anoushka Lotun (Ph.D.; Chair) 2025-presnet

External Educational Activities

- **University of Houston** (Houston, TX, USA): Retinal Vascular Pathologies and potential long-term treatments. Guest lecturer (Drug Delivery and Design course; ~10 students) 03/2022
- **University of Houston** (Houston, TX, USA): Metabolism of the retina and the RPE in health and disease. Guest lecturer (Bioe 4311-01; ~15 students) 09/2020
- **University of Connecticut** (Storrs, CT, USA): The discovery of ectopic eyes: A retrospective view. Role: Invited speaker for lecture within a course. 11/2013
- **Becker College** (Leicester, MA, USA): Use of animals in research. Role: One 45 min lecture on the use of animals in research. 04/2013
- **Harvard Medical School** (Boston, MA, USA): Ocular Gene Therapy. Role: Invited speaker for a 2 hour lecture on the use of gene therapy for eye diseases within a course that focuses on gene therapy. 12/2010

Advising and Mentoring**Students**

-Ph.D. students GSBS Program (Role: Mentor)

Weekly meetings for 1 hour to discuss progress and next steps.

- Shun-Yun Cheng (Michelle), GSBS Ph.D. Student 2016-2021
- Aditya Venkatesh, GSBS Ph.D. Student (Recipient of GSBS student mentoring award in 2013; GSBS Class speaker at graduation ceremony in 2016). Current Position: Stoke Therapeutics 2011-2016
- Shan Ma, Ph.D., Exchange student from China who did Ph.D. work in my lab and graduated in May 2015. Current Position: Optometry School Boston. 2011-2016

-Rotation Students GSBS Program (Role: Mentor)

Weekly meetings for 1 hour to discuss progress and next steps.

- Aditya Venkatesh Spring 2011
- William Monis Spring 2011
- Maeve Tischbein Summer 2012
- Yung Hwang Winter 2013
- Monika Chitre Summer 2015
- Shun-Yun Cheng Summer 2016
- MeenakshiSundaram Kumar Fall 2017
- Jiang Min Fall 2019
- Jonathan Jung Winter 2020
- Jarin Snyder Spring 2021
- YiHan Lee Fall 2021

-Course MDP 740B: Individual Student Mentoring: Eric Schmidt. (Role: Mentor) 03-04/2014

-Course: Medical Student Summer Research Program: Viswanath Ramaswamy. (Role mentor) 06-08/2014

-Course: Medical Student Summer Research Program: Olivia Mallari (Role: Mentor). 06-08/2018

-Visiting M.D. student: Nicholas Belizaire (Role: Mentor) Summer 2021 & 2022

-Visiting student: Samson Jolly (M.D./Ph.D), working 3 months in lab 2021-2022

-Visiting summer high school student: Aditi Dosi (Role: Mentor) 2022

-Research Pathway for UMass Med Students:

- Ilana Silverstein 2023
- Adrienne Conza 2023
- Isreal Adam 2024
- WenTing Zhang 2024

-Harvard Medical School: 2003-2007

- Student Mentoring for BBS graduate Program: Knatokie Ford, Huchung Chung
- Student Mentoring for HHMI Summer student Program: Danielle Andrews-Lovell
- Student Mentoring for Harvard Work Study Program: Soledad Jorge

Postdoctoral Trainees

Weekly meetings for 1 hour to discuss progress and next steps.

- Fernanda Langelotto, Ph.D.; Role: Mentor 2010-2012
Current position: Research assistant at Childrens Hospital, Boston MA
- Marina Zieger, Ph.D.; Role: Mentor 2013-2016
Current position: Research assistant Müller Lab UMASS Medical School
- Lolita Petit, Ph.D.; Role: Mentor; (Sparks Therapeutic) 2014-2017
- Shun-Yun Cheng, Ph.D.; Role: Mentor 2021-present

Investigation**Grants****Current**

- 1R01EY032461-01, NEI 2021-2026
Claudio Punzo (Role: PI)
Title: Identifying the cause for photoreceptor-mediated retinal-pigmented epithelium atrophy.
Total direct costs: US\$ 1,250,000
- Research to Prevent Blindness/American Macular Degeneration Catalyst Award 2023-2025
Claudio Punzo (Role: PI, Anastasia Khvorova, Co-PI)
Title: Development of Stable siRNA therapeutics for the treatment of neovascular AMD
Total direct costs: US\$ 300,000 (Total direct cost for Punzo Lab 156,448)

Completed

- International Retinal Research Foundation (IRRF) 2013-2014
Claudio Punzo (PI)
Title: Modulation of the mTOR pathway: A novel approach to extend vision in dry Age-related macular degeneration.
Total, direct & indirect costs: \$96,450
Role: PI
- International Retinal Research Foundation (IRRF) 2014-2015
Claudio Punzo (PI)
Title: Modulation of the mTOR pathway: A novel approach to extend vision in dry Age-related macular degeneration.
Total, direct & indirect costs: \$99,685
Role: PI
- Massachusetts Lions Eye Research Fund Inc. 2015-2016
Claudio Punzo (Role: PI)
Title: Prolonging vision in Age-related Macular degeneration
Total, direct & indirect costs: \$ 15,333
- 1R01EY023570, NEI 2013-2019
Claudio Punzo (Role: PI)
Title: Delaying cone death in retinitis pigmentosa.
Total direct costs: US\$ 1,250,000
- M2017071, BrightFocus Foundation for Macular Degeneration 2017-2019
Claudio Punzo (Role: PI)
Title: Role of photoreceptors in age-related macular degeneration
Total direct costs: US\$ 160,000

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| Kang Hong Pharmaceutical Claudio Punzo (Role: Co-Pi with Guangping Gao) Title: AAV vector based in vivo delivery of therapeutic genes to retina for the treatment of ophthalmic disorders. Total direct cost: US\$ 2,041,093 (Total direct cost for Punzo Lab: US\$ 435,930) | 2018-2021 |
| 1R21EY031130-01, NEI Frances Lefcort (Role: Co-PI; Total direct cost for Punzo Lab: US\$ ~40,000) Title: Therapeutic strategies for mitigating loss of retinal ganglion cells in familial dysautonomia. Total direct costs: US\$ 275,000 | 2020-2022 |
| 1R21EY030166-01, NEI Claudio Punzo (Role: PI) Title: Testing the role of glucose deprivation during secondary cone death in retinitis pigmentosa. Total direct costs: US\$ 275,000 | 2019-2021 |
| M2020016, BrightFocus Foundation for Macular Degeneration Claudio Punzo (Role: PI) Title: Elucidating how smoking contributes to AMD Total direct costs: US\$ 185,000 | 2020-2022 |
| Kang Hong Pharmaceutical Claudio Punzo (Role: Co-Pi with Guangping Gao) Title: AAV vector based in vivo delivery of therapeutic genes to retina for the treatment of ophthalmic disorders. Total direct cost: US\$ 790,010 (Total direct cost for Punzo Lab: US\$ 113,070) | 2021-2023 |

Scholarship

Peer-reviewed publications

1. Cui M., Su Q., Yip M., McGowan J., **Punzo C.**, Gao G., Tai P.W.L. (2024). The AAV2.7m8 capsid packages a higher degree of heterogeneous genomes than AAV2. **Gene Therapy**: 31, 489-498 <https://doi.org/10.1038/s41434-024-00477-7>.
2. Seddon J.M., De. D., Casazza W., Cheng S.Y., **Punzo C.**, Daly M., Zhou D., Coss S.L., Atkinson J.P., Yu C.Y. (2024). Risk and protection of different protein coding variants of complement component C4A in age-related macular degeneration. **Frontiers in Genetics**: 14: 1274743
3. Cheng S.Y., Caiazzi J., Biscans A., Alterman J.F., Echeverria D., McHugh N., Hassler M., Jolly S., Giguere D., Cipi J., Khvorova A., **Punzo C.** (2023). Single intravitreal administration of a tetravalent siRNA exhibits robust and efficient gene silencing in rodent and swine photoreceptors. **Molecular Therapy Nucleic Acid**: 35: <https://doi.org/10.1016/j.omtn.2023.102088>
4. Schultz A., Cheng S.Y., Kirchner E., Costello S., Miettien H., Chaverra M., King C., George L., Zhao X., Narasimhan J., Weetall M., Slaughter S., Morini E., **Punzo C.**, Lefcort F. (2023). Reduction of retinal ganglion cell death in mouse model of familial dysautonomia using AAV-mediated gene therapy and splicing modulators. **Scientific Reports**: 13: 186000.
5. Huan T., Cheng S.Y., Tian B., **Punzo C.**, Lin H., Daly M., Seddon J.M. (2022). Identifying Novel Genes and Variants in Immune and Coagulation Pathways Associated with Macular Degeneration. **Ophthalmology Science**: 3 (1): 100206
6. Cheng S.Y., **Punzo C.** (2022). Update on Viral gene Therapy Trials for Retinal Diseases. **Human Gene Therapy**: 33 (17-18): 865-878.
7. Wang Y., **Punzo C.**, Ash J.D., Lobanova E.S. (2022). Tsc2 knockout counteracts ubiquitin-proteasome system insufficiency and delays photoreceptor loss in retinitis pigmentosa. **PNAS**: 119 (11): 2118479119.
8. Cheng S.Y., **Punzo C.** (2021). Ocular Inflammation with Anti-Vascular Endothelial Growth Factor Treatments. **Human Gene Therapy**: 32 (13-14): 639-641.

9. Cheng S.Y., Luo Y., Malachi A., Ko J., Su Q., Xie J., Tian B., Lin H., Ke X., Zheng Q., Tai P.L.W., Gao G., **Punzo C.** (2021). Low-Dose Recombinant Adeno-Associated Virus-Mediated Inhibition of Vascular Endothelial Growth Factor Can Treat Neovascular Pathologies Without Inducing Retinal Vasculitis. **Human Gene Therapy**: 32 (13-14): 649-666.
10. Cheng S.Y., Malachi A., Cipi J., Ma S., Brush R.S., Agbaga M.P., **Punzo C.** (2021). HK2 mediated glycolytic metabolism in mouse photoreceptors is not required to cause late stage age-related macular degeneration-like pathologies. **Biomolecules**: 2021, 11, 871.
11. Cheng S.Y., Cipi J., Ma S., Hafler B.P., Kanadia R.N., Brush R.S., Agbaga M.P., **Punzo C.** (2020). Altered photoreceptor metabolism in mouse causes late stage age-related macular degeneration like pathologies. **PNAS**: 117 (23): 13094-13104 (2020).
12. Li H., Li Q., Dang K., Ma S., Cotton J.L., Yang S., Zhu L.J., Deng A.C., Ip Y.T., Johnson R.L. Wu X., **Punzo C.**, Mao J. (2019). YAP/TAZ activation drives uveal melanoma initiation and progression. **Cell Reports**: 29(10): 3200-3211.
13. Petit L., Ma S., Cipi J., Cheng S.Y., Zieger M., Hay N., **Punzo C.** (2018). Aerobic glycolysis is essential for normal rod function and control secondary cone death in retinitis pigmentosa. **Cell Reports**: 23(9): 2629-2642.
14. Venkatesh A., Cheng S.Y., **Punzo C.** Loss of the cone-enriched caspases-7 does not affect secondary cone death in retinitis pigmentosa. **Molecular Vision**: **23**: 944-951 (2017).
15. Petit L., Ma S., Cheng S.Y., Gao G., **Punzo C.** (2017). Rod outer segment development influences AAV-mediated photoreceptor transduction after subretinal injection. **Human Gene Therapy**: 28 (6): 464-481 (2017).
16. Petit L. & **Punzo C.** (2016). Gene Therapy Approaches for the Treatment of Retinal Disorders. **Discovery Medicine**: 22 (121).
17. Camacho E.T., **Punzo C.**, Wirkus S.A. Quantifying the metabolic contribution to photoreceptor death in retinitis pigmentosa via a mathematical model. **Journal of Theoretical Biology**: 408: 75-87 (2016).
18. Venkatesh A., Ma S., **Punzo C.** TSC but not PTEN loss in starving cones of retinitis pigmentosa mice leads to an autophagy defect and mTORC1 dissociation from the lysosome. **Cell Death & Disease**: 7 (6): e2279 (2016)
19. Petit L., Khanna H., **Punzo C.** Advances in gene Therapy for Diseases of the Eye. **Human Gene Therapy**: 27 (8): 563-579 (2016).
20. Choudhury S.R., Fitzpatrick Z., Harris A.F., Maitland S.A., Ferreira J.S., Zhang Y., Ma S., Sharma R.B., Gray-Edwards H.L., Johnson J.A., Johnson A.K., Alonso L.C., **Punzo C.**, Wagner K.R., Maguire C.A., Kotin R.M., Martin D.R., Sena-Esteves M. (2016) *In vivo* selection yields AAV-B1 capsid for CNS and muscle gene therapy. **Molecular Therapy**: 24 (7): 1247-1257 (2016).
21. Zieger M. & **Punzo C.** Improved cell metabolism prolongs photoreceptor survival upon retinal-pigmented epithelium loss in the sodium iodate induced model of geographic atrophy. **Oncotarget**: 7 (9): 9620-9633 (2016).
22. Petit L. & **Punzo C.** mTORC1 sustains vision in retinitis pigmentosa. **Oncotarget**: 6 (19): 16786-16787 (2015).
23. Cepko C.L. & **Punzo C.** Sugar for Sight. **Nature**: 522 (7557): 428-29 (2015).
24. Ma S., Venkatesh A., Langellotto F., Le Y. Z., Hall M. N., Ruegg M. A., **Punzo C.** Loss of mTOR signaling affects cone function, cone structure and expression of cone specific proteins without affecting cone survival. **Exp. Eye Res**: 135: 1-13 (2015).
25. Venkatesh A., Ma S., Le Y. Z., Hall M. N., Ruegg M. A., **Punzo C.** Activated mTORC1 promotes long-term cone survival in retinitis pigmentosa mice. **J of Clin. Inves.**: 125 (4): 1446-58 (2015).
26. Banday A.R., Baumgartner M., Al Seesi S., Karunakaran D.K., Venkatesh A., Congdon S., Lemoine C., Kilcollins A.M., Mandoiu I., **Punzo C.**, Kanadia R.N. Replication-dependent histone genes are actively transcribed in differentiating and aging retinal neurons. **Cell Cycle**: 13 (16): 2526-2541 (2014).
27. Venkatesh A., Ma S., Langellotto F., Gao G., **Punzo C.** Retinal gene delivery by rAAV and DNA electroporation. **Current Protocol**: Chapter 14: Unit 14D.4 (2013).
28. Molnar T., Barabas P., Birnbaumer L., **Punzo C.**, Kefalov V., Krizaj D. Store-operated channels regulate intracellular calcium in mammalian rods. **J. of Physiol.**: 590 (15): 3465-3481 (2012).

29. Hafler B.P., Surzenko N., Beier K.T., **Punzo C.**, Trimarchi J.M., Kong J.H., Cepko C.L. (2012) Transcription factor Olig2 defines subpopulations of retinal progenitor cells biased toward specific cell fates. *Proc. Natl. Acad. Sci.*: 109 (20): 7882-7887 (2012).
30. **Punzo C.**, Xiong W, Cepko C.L. Loss of daylight vision in retinal degeneration: are oxidative stress and metabolic dysregulation to blame? *J. of Bio. Chem.*: 287 (3): 1642-1648 (2012).
31. Huang W, Xing W, Ryskamp DA, **Punzo C.**, Križaj D. Localization and phenotype-specific expression of ryanodine calcium release channels in C57BL6 and DBA/2J mouse strains. *Exp. Eye Res.*: 93 (5): 700-709 (2011).
32. Križaj D., Huang W., Furukawa T., **Punzo C.**, Xing W. Plasticity of TRPM1 expression and localization in the wild type and degenerating mouse retina. *Vision Res.*: 50 (23): 2460-2465 (2010).
33. **Punzo C.**, Kornacker K., Cepko C.L. Stimulation of the insulin/mTOR pathway delays cone death in a mouse model of Retinitis Pigmentosa. *Nature Neuroscience*: 12 (1): 44-52 (2009).
34. Kanadia R.N., Clark V.E., **Punzo C.**, Trimarch J., Cepko C.L. Temporal requirement of the alternative splicing factor Sfrs1 for the survival of retinal neurons. *Development*: 135: 3922-33 (2008).
35. Plaza S., Prince F., Adachi Y., **Punzo C.**, Cribbs D., Gehring W.J. Cross-regulatory Protein-Protein Interactions between Hox and Pax Transcription factors. (2008) *Proc. Natl. Acad. Sci.* 134:39-44 (2008).
36. **Punzo C.**, and Cepko C.L. (2008) Ultrasound-guided *in utero* injections allow studies of ocular development and function. *Developmental Dynamics*: 237 (4): 1034-42 (2008).
37. Liu F., Jenssen T.K., Trimarchi J., **Punzo C.**, Cepko C.L., Ohno-Machado L., Hovig E., Patrick Kuo W. Comparison of hybridization-based and sequencing-based gene expression technologies on biological replicates. *BMC Genomics*: 8: 153 (2007).
38. **Punzo C.**, and Cepko C.L. Cellular responses to photoreceptor death in the rd1 mouse model of retinal degeneration. *Invest Ophthalmol Vis Sci.*: 48 (2): 849-857 (2007).
39. Kuo W.P., Liu F., Trimarchi J., **Punzo C.**, Lombardi M., Sarang J., Whipple M.E., Maysuria M., Serikawa K., Lee S.Y., McCrann D., Kang J., Shearstone J.R., Burke J., Park D.J., Wang X., Rector T.L., Ricciardi-Castagnoli P., Perrin S., Choi S., Bumgarner R., Kim J.H., Short G.F. 3rd, Freeman M.W., Seed B., Jensen R., Church G.M., Hovig E., Cepko C.L., Park P., Ohno-Machado L., Jenssen T.K. A sequence-oriented comparison of gene expression measurements across different hybridization-based technologies. *Nat Biotechnol.*: 24 (7): 832-840 (2006).
40. **Punzo C.**, Plaza S., Seimiya M., Schnupf P., Kurata S., Jaeger J., and Gehring W.J. Functional divergence between *eyeless* and *twin of eyeless* in *Drosophila melanogaster*. *Development*: 131 (16): 3943-53 (2004).
41. **Punzo C.**, Seimiya M., Gehring W.J., and Plaza S. (2002) Differential interaction of *eyeless* and *twin of eyeless* with the *sine oculis* enhancer. *Development*: 129 (3): 625-34 (2002).
42. **Punzo C.**, Kurata S., and Gehring W.J. (2001) The *eyeless* homeodomain is dispensable for eye development in *Drosophila*. *Genes & Development*: 15. 1716-1723 (2001).

Books & Chapters

1. **Punzo C.** Gene Therapy in Animal Models. (Book Chapter; Book title: genetics and Genomic of eye diseases). Publisher: **Elsevier**. ISBN: 978012816222-4. Published: 09/13/2019.
2. Cheng S.Y., **Punzo C.**(2024). New Insights into AMD Pathogenesis. In: Prakash, G., Iwata, T. (eds) *Advances in Vision Research, Volume IV. Essentials in Ophthalmology*. Springer, Singapore. https://doi.org/10.1007/978-981-99-4436-1_12.

Patents

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| Methods for inhibiting starvation of a cell (US2010/031211) | 2010 |
| Adeno-Associated virus for delivery of KH902 and uses thereof (US63/179,700) | 2021 |
| Provisional patent filed: Methods and composition for treatment of AMD (U0120.70134US00) | 2020 |

Invited Presentations**International**

- Role of photoreceptors in AMD pathogenesis. ISER Biennial Meeting, Gold Coast, Queensland, Australia. 02/20/2023
- siRNA-mediated Therapy for the prevention of age-related macular degeneration. ARVO meeting: Revolutionary Eye and Vision Research (on Zoom). Speaker in SIG (Special Interest Group): New Drug Therapeutics for Inherited Retinal Degeneration Diseases (IRDs). 05/04/2021
- Unraveling Age-related macular degeneration. Wet AMD & DME Drug Development Summit (Organizer: Hasonwade; Online). Premeeting workshop on "Developing Preclinical Models for Wet AMD & DME". 04/13/2021
- Role of aerobic glycolysis in photoreceptors. ISER Biennial Meeting, Belfast, Northern Ireland, UK. 09/12/2018
- The duality of mTORC1 in promoting cone survival in Retinitis Pigmentosa. ISER Biennial Meeting, Tokyo, Japan. 09/29/2016
- Prolonging Cone survival in Retinitis Pigmentosa. ISER Biennial Meeting, San Francisco, California, USA. 07/20/2014
- The mechanism of disease pathogenesis in Retinitis Pigmentosa. Meeting on Evolution of Vision, Les Treilles, France. 06/09/2009
- Starvation: A new mechanism for cone death in Retinitis Pigmentosa. ARVO meeting: Reducing disparities in eye disease and treatment. Florida, Fort Lauderdale USA. 05/07/2009
- Understanding rod mediated cone death. Twelfth annual Vision Research conference: Mechanism of macular degeneration. Florida, Fort Lauderdale, USA. 05/01/2009
- Rod toxin and rod trophic factor in Retinitis Pigmentosa. ARVO meeting: Visual Prostheses Research. Florida, Fort Lauderdale USA. 05/14/2008
- The rod toxin and rod trophic factor hypothesis in Retinitis Pigmentosa. University of Basel, Basel, Switzerland. 12/17/2007
- Pax-6 function during development and evolution. IDAC Summer Workshop on Developmental Biology, Sendai, Japan. 07/14/2001

National

- Understanding Disease Progression in AMD. Cole Eye Institute at Cleveland Clinic, Cleveland, OH, USA. In person. 12/12/2024
- Age-related Macular Degeneration: From Humans to mice. University of Texas at San Antonio (UTSA), San Antonio, TX, USA. In person. 11/14/2024
- The role of Photoreceptor in AMD Pathogenesis. Oakland University, Rochester, MI, USA. In person and over Zoom. 12/12/2023
- Photoreceptor Metabolism in AMD Pathogenesis. National Eye Institute, Bethesda, USA. Retinal Disease Interest Group (RDIG) seminar series in person and over Zoom. 12/15/2022
- Role of Photoreceptor Metabolism in Retinal Diseases. Online Zoom; University of Iowa, USA 11/04/2021
- Photoreceptor metabolism in AMD. Online Zoom presentation to Metabolic data club during Covid-19 Pandemic in lieu of meetings. 05/18/2020
- Aerobic Glycolysis in retinal diseases. University of Florida, Gainesville, Florida, USA. 10/16/2019
- Aerobic Glycolysis in retinal diseases. FASEB Meeting, Steamboat Springs, Colorado, USA. 06/25/2019
- Mechanism of cone protection in Retinitis Pigmentosa. FASEB Meeting, Big Sky, Montana, USA. 06/18/2015
- Prolonging cone survival in Retinitis Pigmentosa. Department of Ophthalmology, University of Oklahoma Health and Science Center, Oklahoma City, OK, USA 11/21/2013

- Mechanism of disease pathogenesis in Retinitis Pigmentosa. National Eye Institute, Bethesda, MD, USA. 06/23/2009
- Mechanism of disease pathogenesis in Retinitis Pigmentosa. University of North Carolina, Chapel Hill, NC, USA. 05/11/2009
- Mechanism of disease pathogenesis in Retinitis Pigmentosa. Yale Eye Center, New Haven, CT, USA. 02/25/2009
- Retinitis Pigmentosa: The mechanism of disease pathogenesis. Wright State University, Dayton, OH, USA. 02/20/2009
- Prolonging vision in Retinitis Pigmentosa by saving cones. Washington University School of Medicine, St Louis, MO, USA 06/26/2008
- Prolonging vision in Retinitis Pigmentosa by saving cones. Moran Eye Center, Salt Lake City, UT, USA. 06.03/2008
- From light to genetic inherited eye diseases. 41st SBAO (Swiss Ophthalmology Association) Conference. Bern, Switzerland. 03/19/2001

Regional

- Insulin signaling in rods and cones: Lessons for diabetic retinopathy and age-related macular degeneration. Massachusetts Eye and Ear Infirmary, Harvard Medical School, Boston, MA, USA, 07/26/2016
- Mechanism of disease pathogenesis in Retinitis Pigmentosa. Massachusetts Eye and Ear Infirmary, Harvard Medical School, Boston, MA, USA. 03/27/2009
- The *eyeless* homeodomain is dispensable for eye development in *Drosophila*. Swiss Drosophila Meeting, Fribourg, Switzerland. 03/31/2001
- The *eyeless* homeodomain is dispensable for eye development in *Drosophila*. Regional Meeting, Freiburg, Germany). 03/02/2001

Local

- Understanding Age-related Macular Degeneration. Vision Seminar Series, Department of Ophthalmology, UMass Chan Medical School, Worcester, MA, USA. 01/16/2025
- Unraveling Photoreceptor Metabolism in AMD pathogenesis. Vision Seminar Series, Department of Ophthalmology, UMass Chan Medical School, Worcester, MA, USA. 01/12/2023
- Unraveling the underlying cause for blindness in retinitis pigmentosa. Neuroscience of Disease Forum seminar series, UMass Medical School, Worcester, MA, USA. 10/25/2018
- The Theory of everything: Why, where and what causes Age-related Macular Degeneration to develop. Albert Biology Colloquium (ABC seminar series), UMass Medical School, Worcester, MA USA 02/09/2018
- Retinitis Pigmentosa: from disease to treatment. UMass Neuroscience Seminar Series 09/08/2016
- Insulin signaling in rods and cones: Lessons for diabetic retinopathy and age-related macular degeneration. UMass Vision Seminar Series, Worcester, MA, USA. 08/18/2016
- Delaying Vision loss in Retinitis Pigmentosa. Albert Biology Colloquium (ABC seminar series), UMass Medical School, Worcester, MA USA. 11/14/2014

Other Presentations, Posters & Abstracts

International

- Conza A., Silverstein I., Giguere D., Cheng S.Y., **Punzo C.** Role of Alpha-1 antitrypsin in ocular health. ARVO: 2024, Seattle Washington. Revolutionary (Investigative Ophthalmology & Visual Science 65 (7), 1296-1296). 05/05/2024
- Cheng S.Y., Echeverria D., Giguere D., Alterman A., Agbaga M.P., Seddon J., Khvorova A., **Punzo C.** Genetic and pharmacological reduction of S6K1 alleviates pathologies in the mouse model of age-related macular degeneration (AMD). ARVO: 2024, Seattle Washington. Revolutionary (Investigative Ophthalmology & Visual Science 65 (7), 6519-6519). 05/09/2024

- Silverstein I., Conza A., Giguere D., Alterman A., Echeverria D., Gross K., Cheng S.Y., Khvorova A., **Punzo C.** siRNA mediated gene silencing of Vasculae Endothelial Growth Factor (VEGF) for the treatment of neovascular pathologies of the eye. ARVO: 2024 Seattle, Washington (Investigative Ophthalmology & Visual Science 65 (7), 236-236). 05/05/2024
- Cheng S.Y., Tai P., Malachi A., Zheng Q., Ke X., Tian B., Lin H., Gao G., **Punzo C.** Low-dose rAAV-mediated inhibition of VEGF can treat neovascular pathologies without inducing retinal vasculitis. ARVO: Revolutionary Eye and Vision Research. (Investigative Ophthalmology & Visual Science 62 (8), 1201-1201). 05/05/2021
- Cheng S.Y., Lou Y., Malachi A., Ko J., Su Q., Xie J., Tian B., Lin H., Ke X., Zheng Q., Tai P., Gao G., **Punzo C.** rAAV-mediated inhibition of Vascular Endothelial Growth Factor for the treatment of retinal vascular pathologies without causing vasculitis. ASGCT Meeting. (Molecular Therapy: 29 (4): 161-162). 404/27/2021
- Cheng S.Y., Ma S., Agbaga M.P., **Punzo C.** Elucidating the role of photoreceptors in AMD pathogenesis. ARVO: From Bench to bedside and back. Vancouver, Canada. (Investigative Ophthalmology & Visual Science 60 (15), 1220-1220) 04/28/2019
- Petit L., Ma S., Cheng S.Y., Gao G., **Punzo C.** Rod outer segments influence the efficiency of AAV-mediated rod transduction. ARVO: Imaging in the eye conference. Baltimore, Maryland, USA. (Investigative Ophthalmology & Visual Science 58 (13), 4094-4094). 05/10/2017
- Venkatesh A., Ma S., **Punzo C.** Activation of mTORC1 is sufficient for long-term cone survival in retinitis pigmentosa. ARVO: Leading eye and vision research. Orlando, Florida, USA. (Investigative Ophthalmology & Visual Science 55 (13), 3985-3985). 04/30/2014
- Ma S., Venkatesh A., **Punzo C.** Loss of mTORC1 & mTORC2 but not mTORC1 or mOTRC2 leads to reduction in cone function. ARVO: Leading eye and vision research. Orlando, Florida, USA. (Investigative Ophthalmology & Visual Science 55 (13), 378-378). 04/30/2014
- Molnar T., Barabas P., **Punzo C.**, Krizaj D. Store-operated Calcium Entry Regulates Intracellular Calcium Homeostasis In Mouse Rod Photoreceptors. ARVO: Visionary Genomics. Fort Lauderdale, Florida, USA (Investigative Ophthalmology & Visual Science 52 (14), 6581-6581). 04/22/2011
- Krizaj D., Witkovsky P., Barabas P., **Punzo C.**, Renteria R. C., Liedtke W., Huang W. H. Expression and Function of TRPV4 Channels in the Vertebrate Retina. ARVO: The Future of Eye & Vision Research. Fort Lauderdale, Florida, USA (Investigative Ophthalmology & Visual Science 51 (13), 1860-1860). 04/17/2010
- Krizaj D., Huang W., Zou J., **Punzo C.**, Birnbaumer L., Barabas P. The Canonical Trpc1 Channel Modulates Rod Signals in the Mammalian Retina. ARVO: Reducing disparities in eye disease and treatment. Fort Lauderdale, Florida, USA (Investigative Ophthalmology & Visual Science 50 (13), 5177-5177). 04/28/2009
- Roesch K., Jadhav A., **Punzo C.**, Sun B., Cepko C.L. Muller glia cell response to retinal degeneration. ARVO: The aging eye. Florida, Fort Lauderdale USA (Investigative Ophthalmology & Visual Science 48 (13), 2948-2948). 05/10/2007
- **Punzo C.**, Cepko C. L. Ultrasound guided in utero gene delivery: A tool to manipulate early born retinal cell types. ARVO: The aging eye. Fort Lauderdale, Florida, USA (Investigative Ophthalmology & Visual Science 48 (13), 4603-4603). 05/10/2007
- **Punzo C.**, Cepko C. L. Distinct cellular responses to rod and cone death in the rd1 mouse model of retinal degeneration. ARVO: Building international collaborations. Fort Lauderdale, Florida, USA (Investigative Ophthalmology & Visual Science 47 (13), 5774-5774). 05/01/2006

National

- Venkatesh A., Ma S., **Punzo C.**, Applying m'TORC'1 to prolong vision in Retinitis Pigmentosa. American Society for Cell Biology (ASCB). San Diego, California, USA. 12/2015

- Venkatesh A., Ma S., **Punzo C.** mTOR signaling and autophagy in retinal disease. Autophagy Keystone Symposium. Breckenridge, Colorado, USA. 06/2015

Regional

- **Punzo C.** Preventing blindness in retinitis pigmentosa: One size fits all. inKNOWvation Gene Therapy. Cambridge, Massachusetts, USA. 05/2016

Local

- Cheng S.Y., Ma S., **Punzo C.** A paradigm-shifting hypothesis for the development of age-related macular degeneration. UMass Metabolic Meeting at Pfizer. Cambridge Massachusetts, USA. 05/2017
- Cheng S.Y., Ma S., **Punzo C.** A paradigm-shifting hypothesis for the development of age-related macular degeneration. UMass Basic Sciences Retreat 10/2016
- Venkatesh A., Ma S., **Punzo C.** Applying m'TORC'1 to prolong vision in Retinitis Pigmentosa. UMass Basic Sciences Retreat. 10/2015
- Venkatesh A., Ma S., Le Y. Z., Hall M. N., Rüegg M. A., **Punzo C.** Activation of mTORC1 is sufficient for long-term cone survival in Retinitis Pigmentosa. UMass Basic Sciences Retreat (Venkatesh: Poster Award winner). 07/2014
- Venkatesh A., **Punzo C.** Apoptotic cone cell death in retinitis pigmentosa. UMass Basic Science Retreat. 01/2013

Academic Service

Internal Administration and Service

Department

- Gene Therapy Center: Webpage design, development & maintenance 2014-2018
- Gene Therapy Center: Training & Maintenance of microscope facility of GTC. 2012-2018
- Ophthalmology: Member of Departmental Personnel Action Committee (DPAC) 2019-present
- Ophthalmology: Interviewed Faculty candidates 2017-present
- Ophthalmology: Reviewed application for residency program 2019-present
- Ophthalmology: Interviewed applicants for residency program 2019-present
- Ophthalmology: Training and maintenance of Ophthalmology research equipment 2019-present
- Ophthalmology: Vice Chair of research appointment 2021-present
- Ophthalmology: Chair of Departmental Personnel Action Committee (DPAC) 2023-present

School

- Multiple Mini Interviews (MMI) for prospective Medical School Students (Each session consists of 8 interviews: Sessions conducted 37 to date) 2014-2020
- Interviews of prospective M.D./Ph.D. and Ph.D. Students for GSBS Program: (Interviews conducted to date: 25 M.D./Ph.D. students and 34 Ph.D. students) 2011-present
- Interviews with prospective Faculty Candidates 2010-present

University

- IACUC: Help select new software for online protocol submission portal 2018
- IACUC (Full member), UMASS Medical School (reviewed to date >100 protocols and performed >10 semiannual inspections) 07/2016-present
- IACUC (Alternate member), UMASS Medical School 2012-06/2016
- Committee to redesign IACUC protocol for online portal (Organization of logical flow), UMASS Medical School 2014-2015
- Committee to redesign IACUC web-page (Organization of logical flow), UMASS Medical School 2013

Professional Memberships and Activities

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| ARVO (Association for Research in Vision and Ophthalmology). Member | 2004-present |
| AAAS (American Association for the Advancement of Science). Member | 2010-present |
| ISER (International Society for Eye Research). Member | 2014-present |
| ASGCT (American Society of Gene and Cell Therapy). Member | 2016-present |

Editorial Responsibilities

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| BMC Ophthalmology, Associate Editor | 2012-2023 |
| BMC Ophthalmology, Reviewer (11 Assignments to date) | 2012-present |
| Frontiers Journals, Reviewer (6 assignment to date) | 2009-present |
| Genomics, Reviewer (1 assignment to date) | 2011-present |
| Investigative Ophtha. & Visual Science (IOVS), Reviewer (4 assignments to date) | 2011-present |
| Journal of Neuroscience, Reviewer (1 assignment to date) | 2011-present |
| Proceedings National Academy of Sciences (PNAS), Reviewer (3 assignment to date) | 2011-present |
| Proceedings National Academy of Sciences (PNAS), Guest Editor (1 assignment to date) | 2020-present |
| Cold Spring Harbor Protocols, Reviewer (1 assignment to date) | 2012-present |
| PLOS One, Reviewer (2 assignment to date) | 2012-present |
| Molecular Vision, Reviewer (4 assignment to date) | 2012-present |
| Human Gene Therapy (12 assignments to date) | 2013-present |
| Experimental Eye Research, Reviewer (4 assignment to date) | 2015-present |
| Cell Report, Reviewer (3 assignments to date) | 2015-present |
| Cellular and Molecular life Science, Reviewer (1 assignment to date) | 2016-present |
| FASEB Journal, Reviewer (2 assignments to date) | 2016-present |
| Cell Death & Disease (2 assignments to date) | 2016-present |
| Science Signaling (1 assignment to date) | 2017-present |
| Molecular Therapy (1 assignment to date) | 2018-present |
| NeuroReport (1 assignment to date) | 2018-present |
| American Journal of Physiology (1 assignment to date) | 2018-present |
| Visual Neuroscience (1 assignment to date) | 2019-present |

External Professional Service**International**

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| Swiss National Science Foundation: Ad hoc mail Reviewer (1Grant) | 01/2014 |
| Swiss National Science Foundation: Ad hoc mail Reviewer (2 Grants) | 11/2014 |
| Swiss National Science Foundation: Ad hoc mail Reviewer (1 Grant) | 01/2016 |
| Retina France: Ad hoc mail reviewer (1 Grant) | 10/2017 |
| Israel Science Foundation: Ad hoc mail reviewer (1 Grant) | 02/2018 |
| Swiss National Science Foundation: Ad hoc mail Reviewer (1 Grant) | 06/2018 |
| MRC United Kingdom (1 Grant) | 02/2019 |
| Swiss National Science Foundation: Ad hoc mail Reviewer (1 Grant) | 06/2019 |
| Scientific Advisory Board: LimnoPharma (Spain) | 2020-2023 |
| Deutsche Forschungsgemeinschaft (DFG): Ad hoc mail reviewer (1 Grant) | 09/2021 |
| United States-Israel Binational Science Foundation (BSF; Ad hoc mail reviewer) | 01/2022 |
| Deutsche Forschungsgemeinschaft (DFG): Ad hoc mail reviewer (1 Grant) | 07/2022 |

National

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| NIH: Ad hoc reviewer BVS Study Section (7 R01 Grants) | 10/2017 |
| Fight for Sight: Ad hoc mail reviewer (1 Fellowship) | 10/2017 |
| NIH: Ad hoc reviewer BNVT Study Section (1 R01, 2 R21 Grants) | 02/2018 |

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| NIH: Ad hoc reviewer BVS study section (8 R01 Grants) | 02/2019 |
| NIH: Ad hoc reviewer for special emphasis panel (F05-U) (Grants 8) | 07/2020 |
| FFB: Foundation Fighting Blindness (TRAP LOI: 8) | 08/2020 |
| NIH: Ad hoc reviewer for special emphasis panel (F05-U) (Grants 8) | 10/2020 |
| NIH: Ad hoc reviewer PED1 study section (8 R01 Grants) | 02/2022 |
| NIH: Ad hoc reviewer BIVT study section (5 R01, 2 R21 Grants) | 05/2023 |
| NIH: Ad hoc reviewer ZEY1 VNS (1K08, 1 K23 Grant) | 10/2023 |
| FFB: FFB: Foundation Fighting Blindness (LOI: 1) | 12/2023 |
| FFB: FFB: Foundation Fighting Blindness (LOI + Grant) | 05/2024 |
| NIH: Ad hoc reviewer PED2 study section (6 R01 Grants, 3 R21 Grants) | 03/2025 |

Regional

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| Scientific Advisory Board: Gemini Therapeutics, Cambridge MA | 2017-2020 |
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Professional Development

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| • Basic training ... ABC's for IACUC & IBCs: Frameworks for compliance (UMass) | 2013 |
| • Junior Faculty Development Program (UMass) | 2011-2012 |