

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 2017-present
Department Ophthalmology, Department of Neurobiology & Gene Therapy Center,
University of Massachusetts Medical School, Worcester, MA
- Assistant Professor 2015-2017
Department Ophthalmology, Department of Neurobiology & Gene Therapy Center,
University of Massachusetts Medical School, Worcester, MA
- Assistant Professor 2010-2015
Department Ophthalmology & Gene Therapy Center, 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

Professional Memberships and Activities

- 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) 2016-present

Updated:

Editorial Responsibilities

- Reviewer for several Journals 2010-present
- Associate Editor
BMC Ophthalmology 2012-present

Educational Activities

Teaching Activities

- BBS 782: Tutorial: Bases of Brain Diseases. Role: Course Director 2018-present
- BBS 782 Tutorial: Bases of Brain Diseases. Role: Lecture 2011-present
- BBS 760: Introduction into Neuroscience. Role: Lecture 2014-present
- Summer RAPS: Paper discussions for new incoming students 2012-2013
- FM 201: Brain Course for 2nd year medical students. Role: Lecture 2014
- Summer Course on Gene Therapy held at the GTC. Role: Lecture 2011-2014

Graduate Student Education

Graduate Student Program:

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

Advising, Mentoring & Supervision:

University of Massachusetts Medical School

- Ph.D. students, GSBS Program (3 Students) 2010-present
- Rotation students, GSBS Program (7 Students)
- Postdoctoral Fellows: (3 Post-docs)
- Ph.D. students committee member, GSBS Program (11 Students)

Harvard Medical School

- Rotation students, BBS Program: (2 Students) 2002-2010
- HHMI summer student program: (1 Student)
- Harvard work study student: (1 Student)

External Educational Activities (Lectures)

- **University of Connecticut:** The discovery of ectopic eyes: A retrospective view. (Storrs, CT, USA) 11/2013
- **Backer College:** Use of animals in research. (Leicester, MA, USA). 04/2013
- **Harvard Medical School:** Ocular gene Therapy. (Boston, MA, USA). 12/2010

Grants**Current**

National Institute of Health/ NEI Claudio Punzo (PI) Title: Delaying cone death in retinitis pigmentosa. Description: Identify the downstream targets of mTORC1 that promote cone survival in retinitis pigmentosa Total, direct & indirect costs: \$ 2,084,600 Role: PI (40% effort)	1R01EY023570	2013-2018
BrightFocus Foundation for Macular Degeneration Title: Role of photoreceptors in age-related macular degeneration Description: Studying the role of photoreceptor metabolism in the development of AMD Total, direct & indirect costs: \$ 160,000 Role: PI (5% effort)	Claudio Punzo (PI)	2017-2019

Completed

International Retinal Research Foundation (IRRF) Title: Modulation of the mTOR pathway: A novel approach to extend vision in dry Age-related macular degeneration. Description: The project aims at testing is increasing cell metabolism in a cell autonomous manner in photoreceptors prologs photoreceptor survival in the sodium iodate model of dry Age-related macular degeneration. Total, direct & indirect costs: \$96,450 Role: PI (8% effort)	Claudio Punzo (PI)	2013-2014
International Retinal Research Foundation (IRRF) Title: Modulation of the mTOR pathway: A novel approach to extend vision in dry Age-related macular degeneration. Description: The project aims at testing is increasing cell metabolism in a cell autonomous manner in photoreceptors prologs photoreceptor survival in the sodium iodate model of dry Age-related macular degeneration. Total, direct & indirect costs: \$99,685 Role: PI (8% effort)	Claudio Punzo (PI)	2014-2015
Massachusetts Lions Eye Research Fund Inc. Title: Prolonging vision in Age-related Macular degeneration Description: Identifying why in females mice photoreceptors are more resistant to sodium iodate induced retinal-pigmented epithelium loss. Total, direct & indirect costs: \$ 15,333 Role: PI (5% effort)	Claudio Punzo (PI)	2015-2016

Technology Development**Patents**

Methods for inhibiting starvation of a cell (US2010/031211)	2010
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Publications

Peer-reviewed publications

1. Petit L., Ma S., Cipi J., Cheng S.Y., Zieger M., Hay N., **Punzo C.** Aerobic glycolysis is essential for normal rod function and control secondary cone death in retinitis pigmentosa. *Cell Reports* (In press: 2018).
2. 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)
3. Petit L. Ma S., Cheng S.Y., Gao Guangping., Punzo C. Rod Outer Segment Development Influences AAV-Mediated Photoreceptor Transduction After Subretinal Injection. *Human Gene Therapy*: **28** (6): (2017).
4. Petit L. & **Punzo C.** (2016). Gene Therapy Approaches for the Treatment of Retinal Disorders. *Discovery Medicine*: **22** (121).
5. 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).
6. 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)
7. Petit L., Khanna H., **Punzo C.** Advances in gene Therapy for Diseases of the Eye. *Human Gene Therapy*: **27** (8): 563-579 (2016).
8. 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-Estevés M. (2016) *In vivo* selection yields AAV-B1 capsid for CNS and muscle gene therapy. *Molecular Therapy*: **24** (7): 1247-1257 (2016).
9. 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).
10. Petit L. & **Punzo C.** mTORC1 sustains vision in retinitis pigmentosa. *Oncotarget*: **6** (19): 16786-16787 (2015).
11. Cepko C.L. & **Punzo C.** Sugar for Sight. *Nature*: **522** (7557): 428-29 (2015).
12. 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. *Experimental Eye Research*: **135**: 1-13 (2015).
13. 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).
14. 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).
15. Venkatesh A., Ma S., Langellotto F., Gao G., **Punzo C.** Retinal gene delivery by rAAV and DNA electroporation. *Current Protocol*: Chapter 14:Unit14D.4 (2013).
16. 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).
17. 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).
18. **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).
19. Huang W, Xing W, Ryskamp DA, **Punzo C**, Krizaj 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).
20. 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).
 21. **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).
 22. 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).
 23. 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) *PNAS*: **105**: 13439-44 (2008).
 24. **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).
 25. 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).
 26. **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).
 27. 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).
 28. **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).
 29. **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).
 30. **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).

Invited Presentations (Talks)

International

- 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
- Mechanisms of cone protection in Retinitis Pigmentosa. FASEB Meeting, Blue Sky, Montana, USA. 06/18/2016
- 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

- 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

- 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

- Unravelling 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. UMass Medical School, Worcester, MA USA. 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
- 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

Other Presentations (Posters & Abstracts)

International

- 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

Professional Development

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

Committee Assignments and Administrative Service

Department

- **Gene Therapy Center:** Webpage, designed and developed GTC webpage. 2014-2015
- **Gene Therapy Center:** Training & Maintenance of microscope facility of GTC. 2012-2018
- **Ophthalmology:** Course Director of Vision Seminar Series. 2016-present
- **Ophthalmology:** Training & Maintenance of Ophthalmology microscope facility.

School

- Multiple Mini Interviews (MMI) for prospective Medical School Students 2014-present
- Interviews of prospective M.D./Ph.D. and Ph.D. Students for GSBS Program 2011-present

University

- IACUC (Full member), UMASS Medical School 2016-present
- IACUC (Alternate member), UMASS Medical School 2012-2016
- Committee to redesign IACUC protocol for online portal 2014-2015
- Committee to redesign IACUC web-page 2013

External Professional Service

- Swiss National Science Foundation: Ad hoc mail Reviewer 2014-present
- NIH: Ad hoc reviewer BVS Study Section October 2017
- Fight for Sight: Ad hoc mail reviewer for Fellowship 2017-present
- Retina France: Ad hoc mail reviewer for grant 2017-present
- NIH: Ad hoc reviewer BNVT Study Section February 2018
- NIH: Ad hoc reviewer BVS Study Section February 2019