

Claudio Punzo, Ph.D.

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Education

Ph.D., Cell Biology, Biozentrum University of Basel, Basel, Switzerland Thesis Title: Functional analysis of <i>Pax-6</i> genes during eye development and evolution. Advisor: Dr. Walter J. Gehring	1997–2001
B.S. in Cell Biology, Biozentrum University of Basel, Basel, Switzerland Thesis Title: Regulation of temperature-induced heat shock reaction of <i>Drosophila ambigua</i> and <i>Drosophila melanogaster</i> . Advisor: Dr. Walter J. Gehring	1995–1997

Appointments

Assistant Professor Department of Ophthalmology, & Gene Therapy Center; University of Massachusetts Medical School, Worcester, MA	2010–present
Department of Neurobiology, University of Massachusetts Medical School, Worcester, MA	2015-present
Postdoctoral Fellow/Scholar Supervisor: Dr. Constance L. Cepko Department of Genetics, Harvard Medical School, Boston, State	2002–2010

Honors and Awards

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

Professional Development

Junior Faculty Development Program (UMMS)
Basic training... ABC's for IACUC & IBCs: Frameworks for compliance (UMMS)

Professional Memberships and Activities

ARVO (Association for Research in Vision and Ophthalmology).	2004–present
AAAS (American Association for the Advancement of Science)	2010-present
ISER (International Society for Eye Research)	2014-present

Editorial Responsibilities

- Reviewer for several Journals
- Associate Editor
BMC Ophthalmology
- Reviewer for Grants
-Swiss National Science Foundation (SNSF)

Educational Activities

Teaching Activities

University of Massachusetts Medical School

Lecturer:

- Course BBS 782: Tutorial: Bases of brain diseases.
- Course BBS 760: Introduction into neuroscience
- Course FM 201: Brain Course for 2nd year MD students
- Course MDP 740B: Individual student mentoring
- Course: Medical Student Summer Research Program

University of Connecticut (Storrs, CT)

Lecturer:

2014

- Course: The discovery of ectopic eyes; a retrospective view.

Backer College (Leicester MA)

Lecturer:

2013

- Course: Use of animals in research.

Harvard Medical School

Lecturer:

- Participated in Course on Gene Therapy Organized by Tannous, Bakhos A.

2010

University of Basel

Teaching Assistant:

1998-2000

Graduate Program Membership

Neuroscience (UMMS)
Cell Biology (UMMS)
Interdisciplinary Graduate Program (UMMS)

Advising, Mentoring & Supervision:

University of Massachusetts Medical School

- Ph.D. students, GSBS Program (2 Students)
- Rotation students, GSBS Program (6 Students)
- Postdoctoral Fellows: (3 Post-docs)
- Ph.D. students committee member, GSBS Program (5 Students)

Harvard Medical School

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

Funding**Current**

1R01EY023570-01, Claudio Punzo, Ph.D. 8/2013-7/2018
National Institute of Health/NEI
Title: Delaying Cone Death in Retinitis Pigmentosa
Total cost requested (2013–2018): \$ 2,084,600 (direct & indirect)

Massachusetts Lions Eye Research Fund Inc., Claudio Punzo, Ph.D. 9/2015-8/2016
Title: Prolonging vision in Age-related Macular degeneration
Total cost requested (2015-2016): \$ 15,333 (direct)

Completed

International Retinal Research Foundation (IRRF), Claudio Punzo PhD. 10/13-9/2014
Title: Modulation of the mTOR pathway: A novel approach to extend vision in dry Age-related macular degeneration.
Total cost requested (2013–2014): \$ \$96,450 (direct & indirect)

International Retinal Research Foundation (IRRF), Claudio Punzo PhD. 10/14-9/2015
Title: Modulation of the mTOR pathway: A novel approach to extend vision in dry Age-related macular degeneration.
Total cost requested (2013–2014): \$ \$99,685 (direct & indirect)

Patents

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

Publications

Peer-reviewed publications

1. **Punzo C.**, Kurata S., and Gehring W.J. (2001) The *eyeless* homeodomain is dispensable for eye development in *Drosophila*. *Genes & Development*: **15**: 1716-1723.
2. **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.
3. **Punzo C.**, Plaza S., Seimiya M., Schnupf P., Kurata S., Jaeger J., and Gehring W.J. (2004) Functional divergence between *eyeless* and *twin of eyeless* in *Drosophila melanogaster*. *Development*: **131** (16): 3943-53.
4. Kuo W.P., Liu F., Trimarchi J., **Punzo C.**, et al.. (2006) A sequence-oriented comparison of gene expression measurements across different hybridization-based technologies. *Nat Biotechnol.*: **24** (7): 832-840.
5. **Punzo C.**, and Cepko C.L. (2007) Cellular responses to photoreceptor death in the rd1 mouse model of retinal degeneration. *Invest Ophthalmol Vis Sci.*: **48** (2): 849-857.
6. Liu F., Jenssen T.K., Trimarchi J., **Punzo C.**, Cepko C.L., Ohno-Machado L., Hovig E., Patrick Kuo W. (2007) Comparison of hybridization-based and sequencing-based gene expression technologies on biological replicates. *BMC Genomics*: **8**: 153.
7. **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.
8. 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.
9. Kanadia R.N., Clark V.E., **Punzo C.**, Trimarchi J., Cepko C.L. Temporal requirement of the alternative splicing factor Sfrs1 for the survival of retinal neurons. (2008) *Development*: **135**: 3922-33.
10. **Punzo C.**, Kornacker K., Cepko C.L. Stimulation of the insulin/mTOR pathway delays cone death in a mouse model of Retinitis Pigmentosa. (2009) *Nature Neuroscience*: **12** (1): 44-52.
11. Križaj D., Huang W., Furukawa T., **Punzo C.**, Xing W. Plasticity of TRPM1 expression and localization in the wild type and degenerating mouse retina. (2010) *Vision Res.*: **50** (23): 2460-2465.
12. 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. (2011) *Exp. Eye Res.*: **93** (5): 700-709.
13. **Punzo C.**, Xiong W, Cepko C.L. (2012) Loss of daylight vision in retinal degeneration: are oxidative stress and metabolic dysregulation to blame? *J. of Bio. Chem.*: **287** (3): 1642-1648.
14. 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.
15. Molnar T., Barabas P., Birnbaumer L., **Punzo C.**, Kefalov V., Križaj D. (2012). Store-operated channels regulate intracellular calcium in mammalian rods. *J. of Physiol.*: 590 (**15**): 3465-3481.
16. Venkatesh A., Ma S., Langelotto F., Gao G., **Punzo C.** (2013) Retinal gene delivery by rAAV and DNA electroporation. *Current Protocol*: Chapter 14:Unit14D.4.
17. 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. (2014) Replication-dependent histone genes are actively transcribed in differentiating and aging retinal neurons. *Cell Cycle.*: **13** (16): 2526-2541.
18. Venkatesh A., Ma S., Le Y. Z., Hall M. N., Ruegg M. A., **Punzo C.** (2015). Activated mTORC1 promotes long-term cone survival in retinitis pigmentosa mice. *J of Clin. Inves.*: 125 (4): 1446-58
19. Ma S., Venkatesh A., Langelotto F., Le Y. Z., Hall M. N., Ruegg M. A., **Punzo C.** (2015). 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

20. Cepko C.L. & **Punzo C.** (2015) Sugar for Sight. *Nature*: **522** (7557): 428-29.
21. Petit L. & **Punzo C.** (2015) mTORC1 sustains vision in retinitis pigmentosa. *Oncotarget*: 6 (19): 16786-16787
22. Zieger M. & **Punzo C.** (2016) 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.
23. Choudhury S.R., Fitzpatrick Z., Harris A.F., Maitland S.A., Ferreira J.F., 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 central nervous system and muscle gene therapy. *Molecular Therapy*: [Epub ahead of print].
24. Petit L., Khanna H., **Punzo C.** (2016). Advances in gene Therapy for Diseases of the Eye. *Human Gene Therapy*: [Epub ahead of print].
25. Venkatesh A., Ma S., **Punzo C.** (2016). 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.

Presentations & Abstracts

Invited Presentations

- Insulin/mTOR signaling in rods and cones: Lessons for Diabetic Retinopathy and Age-related Macular Degeneration. Massachusetts Eye and Ear Infirmary (MEEI), Harvard Medical School, Boston, MA USA. 07/26/2016
- 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 06/09/2009
- Mechanism of disease pathogenesis in Retinitis Pigmentosa; Harvard University, Boston, MA USA 03/27/2009
- 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
- The rod toxin and rod trophic factor hypothesis in Retinitis Pigmentosa; University of Basel, Basel, Switzerland 12/17/2007
- Punzo C., Kurata S., Gehring W.J. "Pax-6 function during development and evolution." IDAC Summer Workshop on Developmental Biology, Sendai, Japan. 07/14/2001
- Punzo C. & Gehring W.J. "From light to genetic inherited eye diseases." 41st SBAO (Swiss Ophthalmology Association) Conference, Bern, Switzerland. 03/19/2001

National/International Meetings

- Punzo C. "Mechanisms of cone protection in Retinitis Pigmentosa." FASEB Meeting (Montana, USA) Talk. 6/18/2015
- Punzo C., Venkatesh A., Ma S. "Prolonging Cone survival in Retinitis Pigmentosa". ISER Biennial Meeting (California, USA). Talk 07/20/2014
- Venkatesh A., Ma S., Punzo C. "Activation of mTORC1 is sufficient for long-term 05/06/2014

cone survival in retinitis pigmentosa." ARVO: Leading eye and vision research (Florida, USA). Poster

- 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 (Florida, USA). Poster 05/04/2014
- The mechanism of disease pathogenesis in Retinitis Pigmentosa. Evolution of Vision (Les Treilles, France). 05/11/2009
- Punzo C., Kornacker K., Cepko C.L "Starvation: A new mechanism for cone death in Retinitis Pigmentosa." ARVO: Reducing disparities in eye disease and treatment (Florida, USA). Talk 05/07/2009
- Punzo C., Kornacker K., Cepko C.L "Understanding rod mediated cone death." Twelfth annual Vision Research conference: Mechanism of macular degeneration (Florida, USA). Talk 05/01/2009
- Punzo C. & Cepko C.L. "Rod toxin and rod trophic factor in Retinitis Pigmentosa." ARVO: Eyes on innovation (Florida, USA). Poster 04/28/2008

Local/Regional Meetings

- Punzo C., Kurata S., Gehring W.J. "The *eyeless* homeodomain is dispensable for eye development in *Drosophila*." Swiss Drosophila (Fribourg, Switzerland). 03/31/2001
- Punzo C., Kurata S., Gehring W.J. "The *eyeless* homeodomain is dispensable for eye development in *Drosophila*." Regional meeting (Freiburg, Germany). 03/02/2001

Committee Assignments and Administrative Services

Committee member:

- IACUC (UMMS)
- Interview of prospective Ph.D. and M.D./Ph.D. students at UMMS
- Multiple Mini Interview (MMI) for M.D. students at UMMS

Administrative Services:

-Services for Gene Therapy Center

- Developed entire GTC website