

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:

2010

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

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., Krizaj D. (2012). Store-operated channels regulate intracellular calcium in mammalian rods. *J. of Physiol.*: **590** (15): 3465-3481.
16. Venkatesh A., Ma S., Langellotto 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., Langellotto 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