Claudio Punzo, Ph.D.

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Education

Ph.D., (Cell Biology), University of Basel, Basel, BS, Switzerland Thesis Title: Functional analysis of *Pax-6* genes during eye development and evolution. Advisor: Dr. Walter J. Gehring	2001
B.S. Major *(magna cum laude)*, University of Basel, Basel, BS, Switzerland Thesis Title: Functional analysis of *Pax-6* genes during eye development and evolution. Advisor: Dr. Walter J. Gehring	1997

Postdoctoral Training

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

Academic Appointments

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

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

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 2017-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 2010-present

-Ph.D. students, GSBS Program (3 Students)

-Rotation students, GSBS Program (7 Students)

-Postdoctoral Fellows: (3 Post-docs)

-Ph.D. students committee member, GSBS Program (5 Students)

Harvard Medical School 2002-2010

-Rotation students, BBS Program: (2 Students)

-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 1R01EY023570 2013-2018
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)

BrightFocus Foundation for Macular Degeneration Claudio Punzo (PI) 2017-2019
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)

Completed

International Retinal Research Foundation (IRRF) Claudio Punzo (PI) 2013-2014
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)

International Retinal Research Foundation (IRRF) Claudio Punzo (PI) 2014-2015
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)

Massachusetts Lions Eye Research Fund Inc. Claudio Punzo (PI) 2015-2016
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)

Technology Development

Patents

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

Publications

Peer-reviewed publications

1. 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).
2. Petit L. & **Punzo C**. (2016). Gene Therapy Approaches for the Treatment of Retinal Disorders. *Discovery Medicine*: **22** (121).
3. 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).
4. 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)
5. Petit L., Khanna H., **Punzo C**. Advances in gene Therapy for Diseases of the Eye. *Human Gene Therapy*: **27** (8): 563-579 (2016).
6. 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).
7. 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).
8. Petit L. & **Punzo C**. mTORC1 sustains vision in retinitis pigmentosa. *Oncotarget*: 6 (19): 16786-16787 (2015).
9. Cepko C.L. & **Punzo C**. Sugar for Sight. *Nature*: **522** (7557): 428-29 (2015).
10. 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).
11. 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 Clinc. Inves.:* 125 (4): 1446-58 (2015).
12. 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).
13. 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).
14. 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).
15. 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).
16. **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).
17. 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).
18. 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).
19. **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).

20. 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).
21. 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).
22. **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).
23. 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).
24. **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).
25. 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).
26. **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).
27. **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).
28. **Punzo C.,** Kurata S., and Gehring W.J. (2001) The e*yeless* homeodomain is dispensable for eye development in *Drosophila*. *Genes & Development*: **15**. 1716-1723 (2001).

Invited Presentations (Talks)

International

- 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

- 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
- 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-present
- **Ophthalmology:** Course Director of Vision Seminar Series. 2016-present

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

Ad hoc mail Reviewer for Swiss National Science Foundation (4 Grants to date) 2014-present