Stanford

Douglas Vollrath

Professor of Genetics, Emeritus

CONTACT INFORMATION

- **Administrative Contact**

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Bio

ACADEMIC APPOINTMENTS

- Professor Emeritus, Genetics
- Member, Bio-X
- Member, Wu Tsai Neurosciences Institute

ADMINISTRATIVE APPOINTMENTS

- Regular Member, NIH Biology of the Visual System Study Section, (2017-2021)
- Board of Scientific Counselors, National Eye Institute-NIH, (2008-2013)

BOARDS, ADVISORY COMMITTEES, PROFESSIONAL ORGANIZATIONS

- Scientific Advisory Board, Foundation Fighting Blindness (2009 - present)

PROFESSIONAL EDUCATION

- BS, University of Wisconsin-Madison , Biochemistry (1981)
- PhD, Stanford University , Biochemistry (1988)
- MD, Stanford University , Medicine (1989)

LINKS

- Vollrath Lab: https://vollrathlab.su.domains/

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

Work in the Vollrath laboratory is focused on understanding processes in the eye that are relevant to human health and disease. The eye is an organ particularly amenable to genetic analysis because its accessibility facilitates detection and characterization of a variety of disease states, yet such diseases rarely impair life span or fertility. We frequently start with genes known to be important in the eye because of their association with

ocular disease phenotypes, and then investigate molecular mechanisms by uncovering pathways and processes relevant to normal eye function and pathogenesis.

RPE mitochondrial dysfunction is thought to play a causative role in retinal degenerative diseases such as mitochondrial retinopathy and age-related macular degeneration. As a test of this hypothesis, we generated mice with an RPE-selective postnatal loss of mitochondrial oxidative phosphorylation (OXPHOS). OXPHOS-deficient RPE cells are surprisingly long-lived, but lose critical epithelial characteristics through cellular dedifferentiation and, later, an epithelial to mesenchymal-like transition. OXPHOS-deficient RPE cells initiate a stress response that includes dependence upon the HGF/c-Met pathway, upregulation of aerobic glycolysis, activation of the mTOR signaling pathway, and cellular hypertrophy. Activation of mTOR and subsequent dedifferentiation can also be triggered by acute chemical oxidative damage to the RPE in vivo. For both chronic metabolic and acute oxidative RPE stress, the consequences for adjacent photoreceptors are profoundly negative, resulting in a gradual or rapid (respectively) retinal degeneration. Strikingly, treatment of animals with the mTOR inhibitor, rapamycin, blunts RPE dedifferentiation and hypertrophy and preserves photoreceptor numbers and function for both stressors. We would like to understand the mechanism of mTOR-mediated RPE dedifferentiation and determine whether this new in vivo RPE stress response is activated in human retinal disease.

Phagocytosis is an example of a basic process that we study. Every morning in mammalian eyes, the distal portion of the light sensing outer segments of photoreceptors are phagocytized by adjacent cells of the retinal pigment epithelium (RPE). Phagocytosis is balanced by new synthesis at the proximal end of the outer segment. Together, these two processes lead to constant turnover of outer segments and serve to repair light- and oxygen-induced damage. The daily ‘big breakfast’ of outer segment material, summed over the life of an animal, distinguishes the post-mitotic RPE cell as the most phagocytic cell in the body. Photoreceptor degeneration in mutant rats and mice with defective RPE phagocytosis demonstrates that this process is essential for the normal functioning of the mammalian retina. By genetic analysis of these mutant rodents, we identified the receptor tyrosine kinase, MERTK, as a critical part of the phagocytic mechanism. We also identified mutations in the human MERTK gene in individuals with a retinal degenerative disease known as retinitis pigmentosa. We have elaborated our understanding of the mechanism of phagocytosis by demonstrating that MERTK acts locally, at the site of phagocytosis, to promote ingestion of bound outer segment tips. MERTK does so by triggering a striking redistribution of myosin II from the cell periphery to sites of ingestion. We are continuing to investigate the mechanism of RPE phagocytosis with an emphasis on identifying new protein components and understanding its circadian regulation.

Teaching

COURSES

2025-26

- Advanced Genetics: GENE 205 (Win)
- Genetics and Developmental Biology Training Camp: DBIO 200, GENE 200 (Aut)

2024-25

- Advanced Genetics: GENE 205 (Win)
- Genetics and Developmental Biology Training Camp: DBIO 200, GENE 200 (Aut)

2023-24

- Advanced Genetics: GENE 205 (Win)

2022-23

- Advanced Genetics: GENE 205 (Win)

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Genetics (Phd Program)

Publications

PUBLICATIONS

- **Alternative oxidase blunts pseudohypoxia and photoreceptor degeneration due to RPE mitochondrial dysfunction.** *Proceedings of the National Academy of Sciences of the United States of America*
  Chen, M., Wang, Y., Dalal, R., Du, J., Vollrath, D.
  2024; 121 (25): e2402384121
- **In the Eyes of the Beholder-New Mertk Knockout Mouse and Re-Evaluation of Phagocytosis versus Anti-Inflammatory Functions of MERTK.** *International journal of molecular sciences*
  Ghosh, S., Finnemann, S. C., Vollrath, D., Rothlin, C. V.
  2024; 25 (10)
- **Therapeutic blood-brain barrier modulation and stroke treatment by a bioengineered FZD4-selective WNT surrogate in mice.** *Nature communications*
  Ding, J., Lee, S., Vlahos, L., Yuki, K., Rada, C. C., van Unen, V., Vuppalapaty, M., Chen, H., Sura, A., McCormick, A. K., Tomaske, M., Alwahabi, S., Nguyen, et al
  2023; 14 (1): 2947
- **Cilia-associated wound repair mediated by IFT88 in retinal pigment epithelium.** *Scientific reports*
  Ning, K., Bhuckory, M. B., Lo, C. H., Sendayen, B. E., Kowal, T. J., Chen, M., Bansal, R., Chang, K. C., Vollrath, D., Berbari, N. F., Mahajan, V. B., Hu, Y., Sun, et al
  2023; 13 (1): 8205
- **An efficient inducible RPE-Selective cre transgenic mouse line.** *Experimental eye research*
  Chen, M., Kim, L., Lu, C. W., Zeng, H., Vollrath, D.
  2020: 108370
- **AMP-independent activator of AMPK for treatment of mitochondrial disorders.** *PloS one*
  Moore, T., Yanes, R. E., Calton, M. A., Vollrath, D., Enns, G. M., Cowan, T. M.
  2020; 15 (10): e0240517
- **Multi-trait genome-wide association study identifies new loci associated with optic disc parameters** *COMMUNICATIONS BIOLOGY*
  Bonnemaijer, P. W. M., van Leeuwen, E. M., Iglesias, A. I., Gharahkhani, P., Vitart, V., Khawaja, A. P., Simcoe, M., Hoehn, R., Cree, A. J., Igo, R. P., Burdon, K. P., Craig, J. E., Hewitt, et al
  2019; 2: 435
- **Depletion of Mitochondrial DNA in Differentiated Retinal Pigment Epithelial Cells.** *Scientific reports*
  Hu, X., Calton, M. A., Tang, S., Vollrath, D.
  2019; 9 (1): 15355
- **Association of a Primary Open-Angle Glaucoma Genetic Risk Score With Earlier Age at Diagnosis.** *JAMA ophthalmology*
  Fan, B. J., Bailey, J. C., Igo, R. P., Kang, J. H., Boumenna, T., Brilliant, M. H., Budenz, D. L., Fingert, J. H., Gaasterland, T., Gaasterland, D., Hauser, M. A., Kraft, P., Lee, et al
  2019
- **Highly Differentiated Human Fetal RPE Cultures Are Resistant to the Accumulation and Toxicity of Lipofuscin-Like Material.** *Investigative ophthalmology & visual science*
  Zhang, Q., Presswalla, F., Calton, M., Charniga, C., Stern, J., Temple, S., Vollrath, D., Zacks, D. N., Ali, R. R., Thompson, D. A., Miller, J. M.
  2019; 60 (10): 3468–79
- **Primary fetal RPE cultures resist accumulation and toxicity of lipofuscin-like material, and accumulated material can be further reduced by autophagy induction.**
  Zhang, Q., Presswalla, F., McCusker, A., Charniga, C., Calton, M. A., Vollrath, D., Temple, S., Stern, J., Zacks, D. N., Thompson, D. A., Miller, J.
  ASSOC RESEARCH VISION OPHTHALMOLOGY INC.2019

- **Genetic analyses of human fetal retinal pigment epithelium gene expression suggest ocular disease mechanisms.** *Communications biology*
  Liu, B., Calton, M. A., Abell, N. S., Benchorin, G., Gloudemans, M. J., Chen, M., Hu, J., Li, X., Balliu, B., Bok, D., Montgomery, S. B., Vollrath, D.
  2019; 2 (1): 186
- **Myocilin Mutations in Patients With Normal-Tension Glaucoma** *JAMA OPHTHALMOLOGY*
  Alward, W. L. M., van der Heide, C., Khanna, C. L., Roos, B. R., Sivaprasad, S., Kam, J., Ritch, R., Lotery, A., Igo, R. P., Bailey, J., Stone, E. M., Scheetz, T. E., Kwon, et al
  2019; 137 (5): 559–63
- **Abnormal mTORC1 signaling leads to retinal pigment epithelium degeneration.** *Theranostics*
  Huang, J., Gu, S., Chen, M., Zhang, S. J., Jiang, Z., Chen, X., Jiang, C., Liu, G., Radu, R. A., Sun, X., Vollrath, D., Du, J., Yan, et al
  2019; 9 (4): 1170-1180
- **Cross-ancestry genome-wide association analysis of corneal thickness strengthens link between complex and Mendelian eye diseases (vol 9, 1864, 2018)** *NATURE COMMUNICATIONS*
  Iglesias, A. I., Mishra, A., Vitart, V., Bykhovskaya, Y., Hoehn, R., Springelkamp, H., Cuellar-Partida, G., Gharahkhani, P., Bailey, J., Willoughby, C. E., Li, X., Yazar, S., Nag, et al
  2019; 10: 155
- **Abnormal mTORC1 signaling leads to retinal pigment epithelium degeneration** *THERANOSTICS*
  Huang, J., Gu, S., Chen, M., Zhang, S., Jiang, Z., Chen, X., Jiang, C., Liu, G., Radu, R. A., Sun, X., Vollrath, D., Du, J., Yan, et al
  2019; 9 (4): 1170–80
- **Method for measuring extracellular flux from intact polarized epithelial monolayers.** *Molecular vision*
  Calton, M. A., Beaulieu, M. O., Benchorin, G., Vollrath, D.
  2018; 24: 425-433
- **Testosterone Pathway Genetic Polymorphisms in Relation to Primary Open-Angle Glaucoma: analysis in Two Large Datasets** *INVESTIGATIVE OPHTHALMOLOGY & VISUAL SCIENCE*
  Bailey, J., Gharahkhani, P., Kang, J. H., Butkiewicz, M., Sullivan, D. A., Weinreb, R. N., Aschard, H., Allingham, R., Ashley-Koch, A., Lee, R. K., Moroi, S. E., Brilliant, M. H., Wollstein, et al
  2018; 59 (2): 629–36
- **Genetic correlations between intraocular pressure, blood pressure and primary open-angle glaucoma: a multi-cohort analysis** *EUROPEAN JOURNAL OF HUMAN GENETICS*
  Aschard, H., Kang, J. H., Iglesias, A. I., Hysi, P., Bailey, J., Khawaja, A. P., Allingham, R., Ashley-Koch, A., Lee, R. K., Moroi, S. E., Brilliant, M. H., Wollstein, G., Schuman, et al
  2017; 25 (11): 1261–67
- **Assessment of Murine Retinal Function by Electroretinography** *BIO-PROTOCOL*
  Benchorin, G., Calton, M. A., Beaulieu, M. O., Vollrath, D.
  2017; 7 (7)
- **Age at natural menopause genetic risk score in relation to age at natural menopause and primary open-angle glaucoma in a US-based sample** *MENOPAUSE-THE JOURNAL OF THE NORTH AMERICAN MENOPAUSE SOCIETY*
  Pasquale, L. R., Aschard, H., Kang, J. H., Bailey, J. N., Lindstrom, S., Chasman, D. I., Christen, W. G., Allingham, R. R., Ashley-Koch, A., Lee, R. K., Moroi, S. E., Brilliant, M. H., Wollstein, et al
  2017; 24 (2): 150-156
- **Assessing the Association of Mitochondrial Genetic Variation With Primary Open-Angle Glaucoma Using Gene-Set Analyses.** *Investigative ophthalmology & visual science*
  Khawaja, A. P., Cooke Bailey, J. N., Kang, J. H., Allingham, R. R., Hauser, M. A., Brilliant, M., Budenz, D. L., Christen, W. G., Fingert, J., Gaasterland, D., Gaasterland, T., Kraft, P., Lee, et al
  2016; 57 (11): 5046-5052
- **A Common Variant in MIR182 Is Associated With Primary Open-Angle Glaucoma in the NEIGHBORHOOD Consortium** *INVESTIGATIVE OPHTHALMOLOGY & VISUAL SCIENCE*
  Liu, Y., Bailey, J. C., Helwa, I., Dismuke, W. M., Cai, J., Drewry, M., Brilliant, M. H., Budenz, D. L., Christen, W. G., Chasman, D. I., Fingert, J. H., Gaasterland, D., Gaasterland, et al
  2016; 57 (10): 4528-4535

- **Treatment of retinitis pigmentosa due to MERTK mutations by ocular subretinal injection of adeno-associated virus gene vector: results of a phase I trial.** *Human genetics*
  Ghazi, N. G., Abboud, E. B., Nowilaty, S. R., Alkuraya, H., Alhommadi, A., Cai, H., Hou, R., Deng, W., Boye, S. L., Almaghamsi, A., Al Saikhan, F., Al-Dhibi, H., Birch, et al
  2016; 135 (3): 327-343
- **Genome-wide association analysis identifies TXNRD2, ATXN2 and FOXC1 as susceptibility loci for primary open-angle glaucoma.** *Nature genetics*
  Bailey, J. N., Loomis, S. J., Kang, J. H., Allingham, R. R., Gharahkhani, P., Khor, C. C., Burdon, K. P., Aschard, H., Chasman, D. I., Igo, R. P., Hysi, P. G., Glastonbury, C. A., Ashley-Koch, et al
  2016; 48 (2): 189-194
- **Gene Therapy for MERTK-Associated Retinal Degenerations.** *Advances in experimental medicine and biology*
  LaVail, M. M., Yasumura, D., Matthes, M. T., Yang, H., Hauswirth, W. W., Deng, W., Vollrath, D.
  2016; 854: 487-493
- **The mTOR Kinase Inhibitor INK128 Blunts Migration of Cultured Retinal Pigment Epithelial Cells.** *Advances in experimental medicine and biology*
  Calton, M. A., Vollrath, D.
  2016; 854: 709-715
- **Tyro3 Modulates Mertk-Associated Retinal Degeneration.** *PLoS genetics*
  Vollrath, D., Yasumura, D., Benchorin, G., Matthes, M. T., Feng, W., Nguyen, N. M., Sedano, C. D., Calton, M. A., LaVail, M. M.
  2015; 11 (12): e1005723
- **SPP2 Mutations Cause Autosomal Dominant Retinitis Pigmentosa** *SCIENTIFIC REPORTS*
  Liu, Y., Chen, X., Xu, Q., Gao, X., Tam, P. O., Zhao, K., Zhang, X., Chen, L. J., Jia, W., Zhao, Q., Vollrath, D., Pang, C. P., Zhao, et al
  2015; 5
- **DNA copy number variants of known glaucoma genes in relation to primary open-angle glaucoma.** *Investigative ophthalmology & visual science*
  Liu, Y., Garrett, M. E., Yaspan, B. L., Bailey, J. C., Loomis, S. J., Brilliant, M., Budenz, D. L., Christen, W. G., Fingert, J. H., Gaasterland, D., Gaasterland, T., Kang, J. H., Lee, et al
  2014; 55 (12): 8251-8258
- **Hypothesis-independent pathway analysis implicates GABA and Acetyl-CoA metabolism in primary open-angle glaucoma and normal-pressure glaucoma** *HUMAN GENETICS*
  Bailey, J. N., Yaspan, B. L., Pasquale, L. R., Hauser, M. A., Kang, J. H., Loomis, S. J., Brilliant, M., Budenz, D. L., Christen, W. G., Fingert, J., Gaasterland, D., Gaasterland, T., Kraft, et al
  2014; 133 (10): 1319-1330
- **Intrastriatal Transplantation of Retinal Pigment Epithelial Cells for the Treatment of Parkinson Disease: In Vivo Longitudinal Molecular Imaging with (18)F-P3BZA PET/CT.** *Radiology*
  Bu, L., Li, R., Liu, H., Feng, W., Xiong, X., Zhao, H., Vollrath, D., Shen, B., Cheng, Z.
  2014; 272 (1): 174-183
- **Vascular tone pathway polymorphisms in relation to primary open-angle glaucoma** *EYE*
  Kang, J. H., Loomis, S. J., Yaspan, B. L., BAILEY, J. C., Weinreb, R. N., Lee, R. K., Lichter, P. R., Budenz, D. L., Liu, Y., Realini, T., Gaasterland, D., Gaasterland, T., Friedman, et al
  2014; 28 (6): 662-671
- **PRPF4 mutations cause autosomal dominant retinitis pigmentosa** *HUMAN MOLECULAR GENETICS*
  Chen, X., Liu, Y., Sheng, X., Tam, P. O., Zhao, K., Chen, X., Rong, W., Liu, Y., Liu, X., Pan, X., Chen, L. J., Zhao, Q., Vollrath, et al
  2014; 23 (11): 2926-2939
- **Association of CAV1/CAV2 genomic variants with primary open-angle glaucoma overall and by gender and pattern of visual field loss.** *Ophthalmology*
  Loomis, S. J., Kang, J. H., Weinreb, R. N., Yaspan, B. L., Cooke Bailey, J. N., Gaasterland, D., Gaasterland, T., Lee, R. K., Lichter, P. R., Budenz, D. L., Liu, Y., Realini, T., Friedman, et al
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- **Estrogen pathway polymorphisms in relation to primary open angle glaucoma: An analysis accounting for gender from the United States** *MOLECULAR VISION*
  Pasquale, L. R., Loomis, S. J., Weinreb, R. N., Kang, J. H., Yaspan, B. L., Bailey, J. C., Gaasterland, D., Gaasterland, T., Lee, R. K., Scott, W. K., Lichter, P. R., Budenz, D. L., Liu, et al
  2013; 19: 1471-1481
- **A Novel Homozygous BEST1 Mutation Correlates with Complex Ocular Phenotypes** *OPHTHALMOLOGY*
  Sheng, X., Chen, X., Zhao, K., Liu, Y., Vollrath, D., Zhao, C.
  2013; 120 (7): 1511-+
- **Targeted Sequencing of 179 Genes Associated with Hereditary Retinal Dystrophies and 10 Candidate Genes Identifies Novel and Known Mutations in Patients with Various Retinal Diseases** *INVESTIGATIVE OPHTHALMOLOGY & VISUAL SCIENCE*
  Chen, X., Zhao, K., Sheng, X., Li, Y., Gao, X., Zhang, X., Kang, X., Pan, X., Liu, Y., Jiang, C., Shi, H., Chen, X., Rong, et al
  2013; 54 (3): 2186-2197
- **CDKN2B-AS1 Genotype-Glaucoma Feature Correlations in Primary Open-Angle Glaucoma Patients From the United States** *AMERICAN JOURNAL OF OPHTHALMOLOGY*
  Pasquale, L. R., Loomis, S. J., Kang, J. H., Yaspan, B. L., Abdrabou, W., Budenz, D. L., Chen, T. C., DelBono, E., Friedman, D. S., Gaasterland, D., Gaasterland, T., Grosskreutz, C. L., Lee, et al
  2013; 155 (2): 342-353
- **Amyloid Fibril Formation by the Glaucoma-Associated Olfactomedin Domain of Myocilin** *JOURNAL OF MOLECULAR BIOLOGY*
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- **Genome-Wide Analysis of Central Corneal Thickness in Primary Open-Angle Glaucoma Cases in the NEIGHBOR and GLAUGEN Consortia** *INVESTIGATIVE OPHTHALMOLOGY & VISUAL SCIENCE*
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- **Tyrosine-Mutant AAV8 Delivery of Human MERTK Provides Long-Term Retinal Preservation in RCS Rats** *INVESTIGATIVE OPHTHALMOLOGY & VISUAL SCIENCE*
  Deng, W., Dinculescu, A., Li, Q., Boye, S. L., Li, J., Gorbatyuk, M. S., Pang, J., Chiodo, V. A., Matthes, M. T., Yasumura, D., Liu, L., Alkuraya, F. S., Zhang, et al
  2012; 53 (4): 1895-1904
- **Common Variants at 9p21 and 8q22 Are Associated with Increased Susceptibility to Optic Nerve Degeneration in Glaucoma** *PLOS GENETICS*
  Wiggs, J. L., Yapan, B. L., Hauser, M. A., Kane, J. H., Allingham, R. R., Olson, L. M., Abdrabou, W., Fan, B. J., Wang, D. Y., Brodeur, W., Budenz, D. L., Caprioli, J., Crenshaw, et al
  2012; 8 (4): 413-424
- **An ENU-Induced Mutation in the Mertk Gene (Mertk(nmf12)) Leads to a Slow Form of Retinal Degeneration** *INVESTIGATIVE OPHTHALMOLOGY & VISUAL SCIENCE*
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- **mTOR pathway activation in age-related retinal disease** *AGING-US*
  Zhao, C., Vollrath, D.
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  2011; 52 (3): 1378-1383
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  Zhao, C., Yasumura, D., Li, X., Matthes, M., Lloyd, M., Nielsen, G., Ahern, K., Snyder, M., Bok, D., Dunaief, J. L., LaVail, M. M., Vollrath, D.
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- **Focus on Molecules: MERTK** *EXPERIMENTAL EYE RESEARCH*
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  Ogando, D. G., Dahlquist, K. D., Alizadeh, M., Kunchithapautham, K., Li, J., Yu, N., LaVail, M. M., Rohrer, B., Vollrath, D., Danciger, M.
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- **Rescue of Glaucoma-Causing Mutant Myocilin Thermal Stability by Chemical Chaperones** *ACS CHEMICAL BIOLOGY*
  Burns, J. N., Orwig, S. D., Harris, J. L., Watkins, J. D., Vollrath, D., Lieberman, R. L.
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  Strick, D. J., Feng, W., Vollrath, D.
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- **Sustained Delivery of NT-3 from Lens Fiber Cells in Transgenic Mice Reveals Specificity of Neuroprotection in Retinal Degenerations** *JOURNAL OF COMPARATIVE NEUROLOGY*
  LaVail, M. M., Nishikawa, S., Duncan, J. L., Yang, H., Matthes, M. T., Yasumura, D., Vollrath, D., Overbeek, P. A., Ash, J. D., Robinson, M. L.
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- **Rapid and stable knockdown of an endogenous gene in retinal pigment epithelium** *HUMAN GENE THERAPY*
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- **Temperature sensitive secretion of mutant myocilins** *EXPERIMENTAL EYE RESEARCH*
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- **Mutations in TCF8 cause posterior polymorphous corneal dystrophy and ectopic expression of COL4A3 by corneal endothelial cells** *AMERICAN JOURNAL OF HUMAN GENETICS*
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- **Reversal of mutant myocilin non-secretion and cell killing: implications for glaucoma** *HUMAN MOLECULAR GENETICS*
  Liu, Y. H., Vollrath, D.
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