Ophthalmology

Division Photo

First Row: M. Yang, F. Hamada, S. Riazuddin, C. West, N. Brown, T. Cook, M. Bodack
Second Row: S. Lopper, D. Saltarelli, W. Motley, Z. Ahmed, R. Lang, D. Bonsall, R. North

Division Data Summary

Research and Training Details

<table>
<thead>
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Clinical Activities and Training

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Significant Publications


Featured on Journal Cover and on cover of 40th SFN Anniversary Supplement of invited monographs distributed at annual Society for Neuroscience Meeting. This paper significantly extends mechanistic understanding of Notch signaling regulation of vertebrate retinal neurogenesis.


HoxA1 mutations are associated with autism, as well as Athabaskan brainstem dysgenesis syndrome and Bosley-Salih-Alorainy syndrome, disorders characterized by horizontal gaze palsy, deafness, central hypoventilation, and developmental delay. Up-regulation of HoxA1 is also linked with several cancers. In our study, we demonstrate that HoxA1expression is up-regulated in the pancreas by the TGFb signaling pathway, a pathway intimately linked with
pancreatic cancer. In addition, we map HoxA1's transcriptional regulatory function to the same region that encompasses the autism-linked codon variant. Thus, this paper should provide an important foundation for future studies related to cancer and autism.


Featured on Journal Cover page and in Editors' Corner of the American Journal of Human Genetics. This paper represents the first report on the identification of mutant allele of Barttin causing nonsyndromic hearing loss in humans and described a unique genotype-phenotype correlation, which will be helpful for therapeutic inventions.


Featured on the Cover page of Cell, in Preview as well as in Faculty 1000 website. This paper significantly extends our knowledge about the development of hair cell stereocilia rootlets and function of TrioBP as a novel actin bundling protein.


In this work, we showed that using the kidney as a model system, the signaling ligand Wnt7b produced by macrophages has an important role in tissue repair.

**Division Highlights**

**Zubair Ahmed, PhD**

Dr. Ahmed's research focuses on the identification and characterization of signaling pathways common between vision and sound perceptions. Last year, Dr. Ahmed's lab has contributed in the identification of three new genes that cause inherited hearing loss in the 60 human families and also characterized the novel actin bundling protein, TRIOBP. Dr. Ahmed's work was presented at the Annual meeting of American Society of Human Genetic, Hawaii and 2010 Annual meeting of Association for Research in Otolaryngology, Anaheim, CA. His work on Usher syndrome was recently recognized by Research Horizons.

**Nadean Brown, PhD**

In the past fiscal year, Nadean Brown was awarded a four year renewal of an NIH R01 grant on retinal neurogenesis. Dr. Brown gave invited research presentations at Washington University, Medical College of Wisconsin and Oxford University (England). She also was the principal organizer of the Midwest Society for Developmental Biology, held at Cincinnati Children's in May 2010. This conference had record attendance with participants from 25 research universities and institutes, located in 10 different states.

**Tiffany Cook, PhD**

Dr. Cook's research examines the processes underlying retina and lens formation. Last year, Dr. Cook's work was presented at the University of Idaho, Indiana, and Dayton, the Great Lakes Vision Research Conference, and the Gordon Research Conference on Visual Systems Development in Barga, Italy. Her retina work has led to several collaborative research projects, and was recognized for its applicability to better understanding genetically-based retinal degenerative disorders by Research Horizons and two families afflicted with this disease.

**Fumika Hamada, PhD**

The long-term goal of our research is to understand the molecular mechanisms of thermo- and pain sensation. In FY2010, we carried out a genetic screen using Drosophila, and identified a G-protein coupled receptor, whose mutants show defective temperature preference behavior. We hypothesize that the GPCR is required for temperature processing and may modulate activity of the warmth- or cold-sensitive neural circuits. The mammalian homologue of this GPCR is known to be involved in pain sensation. Therefore, an assessment of the GPCR function in the fly is likely to provide the basis for understanding pain in humans.

**Richard Lang, PhD**

Dr. Lang's laboratory continued making significant scientific contributions during FY2010. His lab has made important advances in our understanding of epithelial morphogenesis mechanisms and has shown that Cdc42-dependent filopodia are critical during the epithelial invagination that results in eye formation. Dr. Lang has also shown that during tissue repair, macrophages produce Wnt pathway ligands to re-capitulate the developmental programs that can re-build a damaged organ. Dr. Lang's research has wide-ranging implications for tissue repair therapies. In this past year, Dr. Lang has presented his work at the World Conference of Regenerative Medicine in Leipzig, Germany, at the Van Andel Research Institute in Grand Rapids, MI and the National Eye Institute at the National Institutes of Health.

**Saima Riazuddin, PhD**

Dr. Riazuddin's research focuses on the identification of genetic factors contributing to inherited hearing impairment in the human population. Last year, Dr. Riazuddin's lab identified two new genetic loci associated with recessive deafness and identified three new genes essential for normal hearing. Dr. Riazuddin's work was presented at the Annual meeting of American Society of Human Genetics in Hawaii and at the 2010 Annual meeting of Association for Research in Otolaryngology, Anaheim, CA. Her work on inherited hearing loss was recognized by Research Horizons and Deafness Research Foundation.
Collaboration with Developmental Biology

**Collaborating Faculty: Jim Wells**
Wntless in Pancreas Development with Richard Lang

**Collaborating Faculty: Aaron Zorn; Rashmi Hegde; Matt Kofron**
CRIM1 Function with Richard Lang

**Collaborating Faculty: Yutaka Yoshida**
Wntless in Neurogenesis with Richard Lang

**Collaborating Faculty: Geraldine Guasch**
Sox2 and Wnt in Transitional Zone Formation with Richard Lang

**Collaborating Faculty: Yi Zheng**
GTPase Function in Morphogenesis with Richard Lang

**Collaborating Faculty: Noah Shroyer**
Wnts in Gut Regeneration with Richard Lang

**Collaborating Faculty: Xinhua Liu**
Wntless Function with Richard Lang

**Collaborating Faculty: Marsha Wills-Karp**
Microglial Function in Vascular Patterning with Richard Lang

**Collaborating Faculty: Brian Gebelein**
Molecular control of Drosophila nervous system development with Tiffany Cook

**Collaborating Faculty: Rashmi Hegde**
Molecular modeling of USH1 protein to identify the effect on the structure with Zubair Ahmed

**Collaborating Faculty: Saulius Sumanus**
Analysis of functional variants causing USH1 using zebrafish as a model system with Zubair Ahmed

**Collaborating Faculty: Robert Sisk**
Genetic studies of retinal disorders, particularly, Blue Cone Monochromat syndrome with Zubair Ahmed

**Collaborating Faculty: Katherine Yutzey**
Jag1 and Rbpj regulations of lens, heart and liver development with Nadean Brown

**Collaborating Faculty: Xinhua Lin**
Characterization of a novel Drosophila BTB domain gene with Nadean Brown

**Collaborating Faculty: David Brown**
Universal newborn hearing screen with Saima Riazuddin

**Collaborating Faculty: Ravi Samy**
Geentics of hearing loss with Saima Riazuddin

**Collaborating Faculty: Saulius Sumanas**
Analysis of DFNB26 mutation using zebrafish as a model system with Saima Riazuddin

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**Faculty Members**

Constance E. West, MD, Associate Professor; Division Director

James J. Augsburger, MD, FACS, Professor; Chairperson, Department of Ophthalmology

Richard A. Lang, PhD, Professor; Emma & Irving Goldman Scholar; Head, Visual Systems Group

Zubair Ahmed, PhD, Assistant Professor

Marie I. Bodack, OD, FAAO, FCOVD, Instructor Clinical

Dean J. Bonsall, MD, MS, FACS, Associate Professor

Tiffany Cook, PhD, Assistant Professor

Fumika Hamada, PhD, Assistant Professor

Adam H. Kaufman, MD, FACS, Associate Professor

Sarah Lopper, OD, Instructor Clinical
Joint Appointment Faculty Members

Nadean Brown, PhD, Associate Professor
Department of Developmental Biology

Saima Riazuddin, PhD, Assistant Professor
Department of Otolaryngology

Clinical Staff Members

- Laurie Hahn-Parrott, CO, COT, MBA
- Corey Bowman, COT, LDO, ABOC
- Brandy Dearwater, COA
- Adrienne Distler, COA
- Jennifer Duncan, COA
- Lisa Fite, COA
- Ashley Jackson, COA
- Debbie Lipps, COA
- Patty Lucas, COA
- Melody Klayer,
- Judy Masters, COT
- Nicole McLeod, COA
- Debbie Meister, COA
- Jill Simmons, COA
- Kelli Vieson, COT

Trainees

- Hope Brown, , Undergraduate Student, Georgia Institute of Technology, Atlanta, GA
- Manpreet Chhabra, MD, Ophthalmology Resident, University of Cincinnati, Cincinnati, OH
- Ian Conner, MD, Ophthalmology Resident, University of Cincinnati, Cincinnati, OH
- April Carpenter-Elrod, PhD, Research Fellow, Hospital for Special Surgery, New York, NY
- Bhavesh Chauhan, PhD, Research Associate, Oxford University, Oxford England
- Abigail Evans, , Undergraduate Student, The Ohio State University, Columbus, OH
- Jieqing Fan, , Graduate Student, Tsinghua University, Beijing, China
- Raja GoI, MD, Ophthalmology Resident, University of Cincinnati, Cincinnati, OH
- M. Victoria Gomez, , Undergraduate Student, Xavier University, Cincinnati, OH
- Michael Gray, MD, Ophthalmology Resident, University of Cincinnati, Cincinnati, OH
- Faiz Khaja, MD, Ophthalmology Resident, University of Cincinnati, Cincinnati, OH
- Haruna Kaneko, PhD, Research Fellow, Tokyo Medical and Dental University, Japan
- Rachel Kominsky, , Undergraduate Student, Xavier University, Cincinnati, OH
- Manna Li, PhD, Research Fellow, Peking University Health Science Center, Beijing, China
- Tianyi Lu, , Summer Student, Duke University, Durham, NC
- Amina Malik, MD, Ophthalmology Resident, University of Cincinnati, Cincinnati, OH
- Kate Maurer, , Graduate Student, Susquehanna University, Selinsgrove, PA
- Elizabeth McDonald, , Graduate Student, Hartwick College, Oneonta, NY
- Mehta Mitul, MD, Ophthalmology Resident, University of Cincinnati, Cincinnati, OH
- Myung-Soon Moon, PhD, Research Fellow, University of Wisconsin, Madison, WI
- Jamey Osher, MD, Ophthalmology Resident, University of Cincinnati, Cincinnati, OH
- Timothy Plageman, PhD, Research Fellow, University of Cincinnati, Cincinnati, OH
- Virgilio Ponferrada, PhD, Research Associate, Wright State University, Dayton, OH
- Sujata Rao, PhD, Research Associate, Cornell University, Ithaca, New York
The Visual Systems Group, part of the Division of Ophthalmology, welcomed a new faculty member to our team this year. Fumika Hamada, PhD, from Brandeis University, studies the molecular mechanisms of heat and pain sensation in drosophila. Her recruitment continues to fulfill the mission of Constance West, MD, and Richard Lang, PhD, to develop a successful visual systems basic science research initiative at Cincinnati Children’s. As we enter the coming year, our focus remains dedicated to studying the development and disease processes of visual systems.

**Significant Accomplishments**

**Visual Systems Group**

The Visual Systems Group, part of the Division of Ophthalmology, welcomed a new faculty member to our team this year. Fumika Hamada, PhD, from Brandeis University, studies the molecular mechanisms of heat and pain sensation in drosophila. Her recruitment continues to fulfill the mission of Constance West, MD, and Richard Lang, PhD, to develop a successful visual systems basic science research initiative at Cincinnati Children’s. As we enter the coming year, our focus remains dedicated to studying the development and disease processes of visual systems.

**Division Publications**


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### Grants, Contracts, and Industry Agreements

#### Grants and Contract Awards

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<td>Molecular Genetics of Usher Syndrome Type I</td>
<td>National Institutes of Health</td>
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**Note:** The above text includes a list of scientific publications with authors and titles, followed by a table of grants and contracts awarded. The table includes the full name of the grant recipient, the title of the research, the funder, the annual direct costs, and the project period with associated direct costs.
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<td><strong>The Roles of Sox2 in Lens and Retinal Development</strong></td>
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<td>The Research Institute at Nationwide Hosp (Ohio Department of Health)</td>
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West, C