Our Team

The Young Lab partners with pediatric ophthalmologists and glaucoma experts at the University of Wisconsin School of Medicine and Public Health Department of Ophthalmology and Visual Sciences. Our collaborators are:

Yasmin S. Bradfield, MD, FAAP, Professor
Pediatric Ophthalmology and Strabismus
Member, Childhood Glaucoma Research Network
Vice Chair of Education

Gregg A. Heatley, MD, MMM, Associate Professor
Glaucoma Service, Service Chief

Yao Liu, MD, Assistant Professor
Glaucoma Service
Member, Childhood Glaucoma Research Network

Anna C. Momont, MD, Assistant Professor
Glaucoma Service

Melanie Schmitt, MD, Assistant Professor
Pediatric Ophthalmology and Strabismus
Director of Ophthalmic Genetics, Department of Ophthalmology and Visual Sciences

Terri L. Young, MD, MBA, Professor and Chair
Pediatric Ophthalmology and Strabismus
Member, Childhood Glaucoma Research Network

Dr. Young is the Peter A. Duehr Chair of the Department of Ophthalmology and Visual Sciences at the University of Wisconsin School of Medicine and Public Health. She is a professor of ophthalmology, pediatrics and medical genetics. Dr. Young's primary clinical and basic science research focus has been ophthalmic genetics, specifically the disorders of ophthalmic development, such as primary congenital glaucoma, microphthalmia/anophthalmia, corneal dystrophy and myopia.

Bibliography

Mutations in the Angiopoietin receptor TEK cause primary congenital glaucoma with variable expressivity. *Journal of Clinical Investigation.* 2016. (In press)

CYP1B1, MYOC, and LTBP2 mutations in primary congenital glaucoma patients in the United States. *American Journal of Ophthalmology.* 2013 Mar; 155(3):508-517. PMID: 23218701

Meta-analysis of genome-wide association studies identifies novel loci associated with optic disc morphology. *Genetic Epidemiology.* 2015 Mar;39(3):207-16. PMID: 25634615

Meta-analysis of genome-wide association studies identifies novel loci that influence cupping and the glaucomatous process. *Nature Communications.* 2014 Sep 22;5:4883. PMID: 25241763

Genome-wide analysis of multi-ancestry cohorts identifies new loci influencing intraocular pressure and susceptibility to glaucoma. *Nature Genetics.* 2014 Oct;46(10)1126-30. PMID: 25173106

A genome-wide association study of intra-ocular pressure suggests a novel association in the gene FAM152B in the TwinsUK cohort. *Human Molecular Genetics.* 2014 Jun 15;23(12):3343-8. PMID 24518671

Copy number variation at chromosome 5q21.2 is associated with intraocular pressure. *Investigative Ophthalmology & Visual Science.* 2013 May 1;54(5):3607-12. PMID 23599335

Common genetic determinants of intraocular pressure and primary open-angle glaucoma. *PLoS Genetics.* 2012;8(5):e1002611. PMID 22570627.

Pediatric cataract, myopic astigmatism, familial exudative vitreoretinopathy and primary open-angle glaucoma co-segregating in a family. *Molecular Vision.* 2011;17:2118-28. PMID: 21850187.

UW Health

Department of Ophthalmology and Visual Sciences
UNIVERSITY OF WISCONSIN
SCHOOL OF MEDICINE AND PUBLIC HEALTH

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Making a Difference

Participating in a Pediatric Glaucoma Genetic Research Study

Department of Ophthalmology and Visual Sciences
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What is primary congenital glaucoma?

Primary congenital glaucoma (PCG) is a serious eye disease that is usually detected before age one. In PCG, fluid builds up inside the eyes and causes high pressure. Children with PCG can have high pressure inside one or both eyes. Symptoms can include eyes that look larger than normal, sensitivity to light, watery eyes without crying, and swelling and cloudiness of the front of the eye. Without early recognition and treatment, PCG causes permanent vision loss. The vision loss happens because the high pressure damages the optic nerve in the back of the eye.

Why study families?

Primary congenital glaucoma may be inherited in families. It is important to identify the genes involved in primary congenital glaucoma in order to develop therapies to prevent vision loss and other complications. All ages are eligible to participate in the study.

You may be eligible for this research study if you:

- Have been diagnosed with primary congenital glaucoma
- Have one or more family members diagnosed with primary congenital glaucoma

What have we learned?

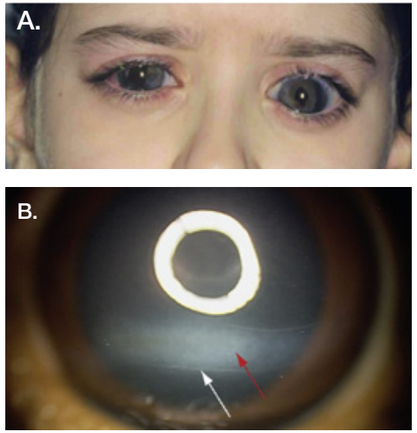
There are genes that we already know cause primary congenital glaucoma when they are not working normally. Changes in the CYP1B1 gene are the most common cause of PCG worldwide. Other genes include LTBP2, MYOC, and FOXC1. We have recently identified a new gene, TEK.

- However, many people have PCG that is not caused by changes in any of these genes because other important genes have not yet been identified.
- Eyes have drainage systems inside them that work to keep eye pressure at healthy levels.
- Severe problems can cause PCG and milder problems may cause glaucoma later in life.
- Our lab is researching which genes are important for making these drainage systems flow normally.

This research study requires you to:

- Complete a short questionnaire, either in person or over the phone
- Provide a sample of DNA from a small blood or saliva sample

Qualified participants can be of any age and will receive a free eye examination.

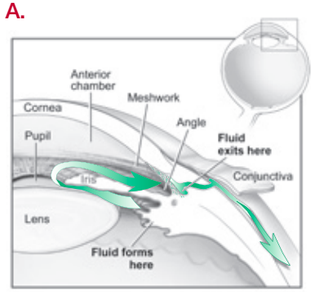


Primary congenital glaucoma

A. *Buphthalmos (enlargement of the eye, left eye) in a child with a history of primary congenital glaucoma. Pupils are altered after multiple surgical procedures.*

B. *Haab striae may lead to corneal edema. The white arrow points to edge of Haab striae, and the red arrow points to the center of Haab striae with corneal edema and opacification. (©2016 American Academy of Ophthalmology)*

A.



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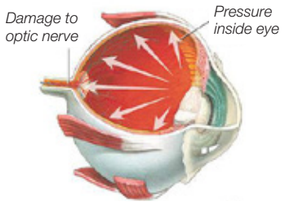
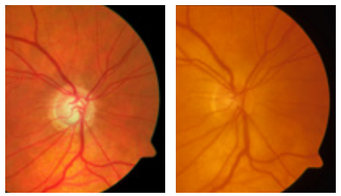


C.



Glaucomatous optic nerve *Normal optic nerve*

Glaucoma

A. *Normal fluid drainage pathway in the eye.*

– National Eye Institute

B. *Abnormalities in fluid drainage can lead to increased pressure inside the eye and damage to the optic nerve.*

– National Library of Medicine

C. *The glaucomatous optic nerve shows increased cupping compared to the normal optic nerve, indicating optic nerve damage that may lead to blindness.*

Photos courtesy of the University of Wisconsin Fundus Photograph Reading Center

Contact us today

If you are interested in participating in this study, please contact study coordinator Nickie Stangel at nstangel@wisc.edu or (608) 263-8783.