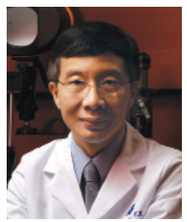
# Simple and Powerful Test

Research led by physician-scientists at Bascom Palmer has produced a breakthrough discovery in diagnosing retinitis pigmentosa, a blinding disease that affects about 1 in 4,000 people in the United States.

Dr. Rong Wen



Dr. Byron Lam

Rong Wen, M.D., Ph.D, and Byron Lam, M.D., professors of ophthalmology, in collaboration with biochemist Ziqiang Guan, Ph.D., a research associate professor at Duke University Medical School, discovered a key marker in blood and urine that can identify people who carry genetic mutations in a gene responsible for retinitis pigmentosa (RP). "A simple urine test can tell who has the RP-causing mutations," says Wen. "Collecting urine is non-invasive and easy, especially from young children."

The first mutation in this gene, named DHDDS, was identified in 2011 by scientists at the University of Miami Miller School of Medicine, including Stephan Zuchner, M.D., Ph.D., Wen, Lam, and Margaret A. Pericak-Vance, Ph.D. on behalf of a South Florida couple who was searching for the reason why three of their children were blinded by RP. Mutations in this gene are more common in persons of Ashkenazi Jewish heritage than in the general population. RP is a group of inherited eye diseases that cause progressive vision loss and blindness due to degeneration of the retina, the layer of light-sensitive tissue at the back of the eye.

"It is our vision that every patient who is affected with an inherited eye disease like RP should have access to a clinician who is knowledgeable about the diseases, as well as to affordable diagnostic testing and counseling," says Lam, director of Bascom Palmer's hereditary eye disease center. "This diagnostic test is a powerful tool that will help in developing treatments for RP caused by DHDDS mutations."

DHDDS, or dehydrodolichol diphosphate synthase, is a key enzyme for producing dolichols, an important type of lipid in cells in the body. The DHDDS mutation has special meaning to the Lidsky family of South Florida. Three of the four Lidsky children, who are now in their 30s, began to lose their sight in their teens. "The fact that a simple blood or urine test can identify the genetic defect that causes this form of RP is very important," says Betti Lidsky, mother of the children, and a founder of Hope for Vision, a non-profit organization dedicated to promoting retinal research. "I have tremendous hope in the doctors and scientists doing this life-changing work and am confident that RP is one step closer to being treated."

Results of this research are published in a paper titled "Aberrant dolichol chain lengths as biomarkers for retinitis pigmentosa caused by impaired dolichol metabolism" in the *Journal of Lipid Research*.

Lam and Wen also serve as directors of the Adrienne Arsht Hope for Vision Center of Retinal Degeneration Research at Bascom Palmer. Their work has been supported by Adrienne Arsht Hope for Vision funds and by the National Eye Institute/ National Institutes of Health.

"We are hopeful that the Bascom Palmer research that identified the genetic cause of retinitis pigmentosa will one day lead to a cure for our family and generations of others." *Betti and Carlos Lidsky*

**Betti and Carlos Lidsky (center) pictured with their children and grandchildren.**


**Betti Lidsky and Adrienne Arsht at the research laboratory dedication.**