Dear BCM Families:

14 March, 2011

We are contacting you because you or your child has been given a diagnosis of blue cone monochromacy (BCM) and therefore may be eligible to participate in a research study led by Samuel G. Jacobson, MD, PhD. The ultimate goal of this study is to identify treatments for individuals with blue cone monochromacy (BCM).

This study is being conducted at the Scheie Eye Institute in Philadelphia, PA. Study participation involves at least one evaluation by Dr. Jacobson during which participants will have simple non-invasive tests of vision and retinal structure. Dr. Jacobson has been working in the field of genetic retinal disorders for his entire career, which now spans three decades. He and his colleagues are currently conducting a clinical trial for therapy of another retinal condition and there have been exciting, positive results. The intention is to extend the success to BCM. This must begin by getting to know in detail the vision and visual potential of patients with the condition.

The study is accompanied by research-based genetic testing to confirm the prior clinical diagnostics and to determine the individual genetic cause of the disease. Genetic testing is carried out at the Molecular Genetics Laboratory, Institute of Ophthalmic Research, University Clinics Tübingen, Germany, under the supervision of Dr. Susanne Kohl and Prof. Bernd Wissinger. Both have a longstanding record in the genetic analysis of inherited retinal disorders and have established a registry for BCM patients. The genetic testing requires blood samples to be shipped to Tuebingen and takes 8-10 weeks for the analysis. If you or your child have already had a genetic testing for BCM in Tübingen, there is no need for another testing.

If you are interested in participating, please call Sharon Wolfe-Schwartz, MS, CGC, Genetic Counselor in Dr. Jacobson’s office at 215-662-9981 or e-mail chrd@mail.med.upenn.edu.

For genetic testing, please contact Dr. Susanne Kohl by phone +49 7071 2980702 or by e-mail: susanne.kohl@uni-tuebingen.de

Sincerely,

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