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SCHOOL OF MEDICINE

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To: Cerebroretinal Vasculopathy (CRV) Kindreds

From: John P. Atkinson, M.D. and his laboratory

RE: New information about the disease

I am writing to update the families who have members afflicted with cerebroretinal vasculopathy (CRV).

First, we thank Kimberly Morey and Sharon Bevers and their families and friends who recently have raised funds that allow our investigators to continue selected aspects of our studies on CRV. Their efforts proved especially critical since we are no longer funded by the two-year stimulus grant from the National Institutes of Health and my personal research funds and those provided by the Bob Clark family have now been depleted. In addition, I thank Madonna Bogacki in our office who continues to bring her devotion to patients and organizational skills to help us carry out many types of activities with family members. She is also the person to contact for those considering genetic testing.

This past year we have achieved a much sought-after milestone, the creation of a mutant mouse carrying the exact genetic alteration found in the mutant gene of afflicted family members. We have accomplished this in collaboration with scientists at the National Institutes of Health in Bethesda, MD and scientists at a biotechnology company (Ozgene) in Australia. These mice are potentially of enormous importance. They will provide us with a powerful tool for the detailed study of the CRV disease process (pathogenesis) and for the development of therapeutic strategies. We can analyze these mice in similar ways that we study human CRV. Thus, we have in place techniques and equipment to examine the eyes of mice identical to what you would experience on a visit to an ophthalmologist. We can also perform imaging of mouse brains (MRI and CT scans) with the same type of equipment that is used with humans. However, using this mouse model, our analyses of CRV will be greatly accelerated and far more detailed than otherwise possible in patients.

We have also attained a second major advance, the production of highly specific CRV antibodies. Antibodies are remarkable tools that enable the detection of specific targets. We have produced antibodies that allow us to identify the normal protein as well as the mutated CRV protein that is responsible for the disease. With these new antibodies, we can track the mutant protein in cells and tissues of both mouse and man. The information we get with these antibodies will be very important.

We are excited about these developments, achieved by Parul Kothari, M.D., Kathy Liszewski and Paula Bertram in our laboratory. We have also been working to raise the awareness of CRV among our fellow biomedical scientists and clinicians. To that end, we are preparing a comprehensive report on clinical and pathologic aspects of disease. Over the past decade we have also learned that changes in the CRV gene are related to at least three other human diseases. It is likely that advances in the understanding and treatment of these diseases will prove useful in our quest to understand CRV.

The creation of a CRV animal disease model and the preparation of antibodies to detect the mutant CRV protein were the two major aims of our research effort. We are thrilled that, with your support, our investigations have made these advances possible. Our next goal is to characterize the CRV disease process in the mouse so we can devise ways to eventually treat CRV. Thank you for all of your cooperation and support.

Sincerely,

John Atkinson

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