Genetic Analysis of Familial Exudative Vitreoretinopathy

Principal Investigator: Johane Robitaille
Departments of Ophthalmology and Pathology, Dalhousie University
Ophthalmologist, IWK Health Centre

Groundbreaking genetic research at the IWK Health Centre in conjunction with Xenon Genetics, Inc. has led to the discovery of the gene that causes a rare, hereditary eye disease that leads to blindness in children and teenagers. Familial exudative vitreoretinopathy (FEVR), or Criswick-Schepens Syndrome, is associated with an abnormal development of blood vessels in the retina, leading to a detached retina causing severely decreased vision and, in some cases, blindness. With the identification of the gene, testing and earlier intervention is now possible for those with a family history of FEVR. The discovery may also eventually have an impact on similar, more common eye conditions.

Dr. Johane Robitaille, an ophthalmologist at the IWK Health Centre, teamed up with researchers in the United States and Vancouver, BC (Xenon Genetics, Inc.) to lead the research into the discovery of the FEVR gene. Although there is no treatment currently available to prevent the progression of this hereditary disease, treatment is possible for certain complications of FEVR that have potential for blindness. There is a clear advantage in treating complications of FEVR at an early stage as the chances for success are improved. Early detection and treatment can prevent blindness and reduce its impact on the individual and the cost to society. Improved counseling will result from either confirmation of an unclear diagnosis or from testing children at risk (i.e., those who have family members with this condition).

Further research into the FEVR gene may reveal that it also plays a role in other more common conditions, such as diabetic retinopathy and retinopathy of prematurity, that affect the eyes in a similar manner to FEVR. Increased efficiency in screening methods, improved timing of known interventions, and the development of better treatments will reduce the risks of blindness, the medical costs related to wide-scale screening, and the cost of the burden of the disease to society. Before specific treatments can be discovered to prevent the development of complications altogether and eradicate blindness caused by this condition, more research is necessary to understand how this gene functions in the eye.

-30-

Contact information:
Johane Robitaille
Ophthalmology Department
Dalhousie University
Phone: 902-470-8008