CENTRE FOR GENOMICS ENHANCED MEDICINE

Patient Impact Story

The Gift of Seeing Colour

For those of you who are parents, think back to that emotional day in the hospital when your child was born. Now imagine that prior to delivery, your doctor tells you there's a 50-50 chance that your child could have FEVR, a gene mutation that causes blindness.

This is the exact news faced by Matthew's parents. But thanks to the work of the Centre for Genomics Enhanced Medicine (CGEM) member Dr. Johane Robitaille, a leading ophthalmologist, and her team, Matthew received immediate laser treatment when he was just six days old. This treatment stopped FEVR from progressing and let him retain some sight.

The CGEM is a multi-disciplinary collaboration of physicians and researchers specializing in inherited orphan diseases. Dr. Johane Robitaille, MD, a pediatric ophthalmologist, is director of the CGEM Research Institute.

Years earlier, Dr. Robitaille saw a little girl named Tarah who was born with FEVR, which stands for familial exudative vitreoretinopathy. With no medical technologies to help her, Tarah, now 17, is totally blind. Tarah reminisces about her early years when she could see people and things. She says she feels lucky because, in her words, it would be "...really difficult to explain colour."

Tarah's joie de vivre is what inspires Dr. Robitaille and her team to better understand this disease. The research began when Tarah and 106 of her relatives across Nova Scotia and Ontario volunteered to have their DNA sequenced. The results identified 30 family relatives with FEVR. The team built one of the largest databases of FEVR patients in North America and used the data to identify FEVR genes – mutations that prevent blood vessels from growing in the retina.

> *"By identifying the genes, we can accurately diagnose FEVR, provide families with counselling and treat our patient's eyes sooner," says Dr. Robitaille. "Now we're working on even better, more targeted treatments."*

The CGEM's ultimate goal is to find a treatment that will reverse the underlying defect in FEVR to prevent the blinding complications. The research team is developing a new drug that targets the Frizzled-4 pathway. This pathway is critical for blood vessel development. Results of preclinical studies show the drug candidate is able to bypass this genetic impairment and help blood vessels grow in the retina. The result is mice with near normal vessel development.

In the future, babies born with FEVR could potentially be treated with this new drug at birth, before their eyes have fully developed. The hope is to fix the problem at the source by promoting blood vessel growth in the retina and preventing blindness caused by FEVR. This treatment could lead to life-long normal vision for babies born with FEVR.

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