

The blind and visually impaired can help researchers by getting their genes tested

October 17 2019, by Ruanne Vent-Schmidt



Credit: AI-generated image ([disclaimer](#))

Blind and partially sighted people no longer have to wait passively for a research breakthrough in hope of treatment options. In fact, people living with genetic eye conditions can now actively drive vision research forward —by enrolling in a patient registry and getting their genes tested.

There are [2.2 billion people living with visual impairment globally](#). Some are living with inherited [retinal diseases](#) that are progressive and can lead to complete blindness. Up until recent years, blind and visually impaired people were told that no treatment is available. This is changing as [genetic testing is paving the way for a surge of gene therapies](#).

My passion for vision research is personal

My [doctoral dissertation](#) at the University of British Columbia was on [drug therapy](#) for [retinitis pigmentosa](#). This progressive, blinding eye condition is the most common type of inherited retinal [disease](#).

In people affected by [retinitis pigmentosa](#), the light sensing cells in their retina—photoreceptors—die early. Unlike skin cells that regenerate, the body does not make more photoreceptors once they are damaged.

As a vision scientist affected by retinitis pigmentosa, I am passionate about finding the truth about the disease. Why do photoreceptors die? How can we stop it? How can science and medicine help?

When I was 12 years old, I realized while at summer camp that my night vision was disappearing. In the last two decades, I lost my peripheral vision, contrast sensitivity and depth perception.

I worked in [Dr. Orson Moritz's lab](#) at the UBC department of ophthalmology and visual sciences, which focuses on research using tadpoles that contain known human mutations for retinitis pigmentosa to understand the disease.

[I made an alarming discovery in our animal model](#): knowing the genetic cause of retinitis pigmentosa is [vital for treatment with one class of drugs—histone deacetylase inhibitors](#). These determine how [genes](#) are switched "on" or "off."

A similar [study in mice](#) showed that the same drug reacted differently to variations in a single mutant gene that also causes retinitis pigmentosa.

Treating retinitis pigmentosa is like extinguishing fire. To stop a fire, you need to know whether it's water-based or grease-based. If you try to use water to stop a grease fire, the damage gets worse.

Enrol in a patient registry

Blind and visually impaired people can advocate for eye health by enrolling in a patient registry. Participation in a registry [benefits researchers by offering more information](#) about the disease.

In Canada, individuals can self-refer to [Fighting Blindness Canada's](#) secure, clinical [patient registry](#). This database is dedicated to connecting people living with retinal eye diseases to clinical trials and research.

When a gene therapy trial arises, researchers draw participants from this database. Since [gene therapy aims to correct an underlying genetic mistake in DNA that causes disease](#), knowing the genetic cause of a disease is a criteria for most gene therapy trials.

Globally, other registries include [My Retina Tracker](#) in the United States, [Target 5000](#) in Ireland, [MyEyeSite](#) in the United Kingdom, the [Australian Inherited Retinal Disease Registry](#) and [Japan Eye Genetics Consortium](#). In New Zealand, [Dr. Andrea Vincent](#) has established the Genetic Eye Disease Investigation Unit. There is even a [Blue Cone Monochromacy Patient Registry](#) for one rare eye condition.

Blossoming gene therapy trials

In the last two decades, the number of gene therapy trials has blossomed.

Currently, [250 genes on inherited retinal diseases have been identified](#). In [2017, the first gene therapy for inherited retinal disease](#) —Luxturna —was [approved by the United States Federal Drug Administration](#).

To date, there are trials for: [retinitis pigmentosa](#); [Usher syndrome](#), a condition that involves hearing and vision loss; [achromatopsia](#), a disease that causes colour blindness; [X-linked retinoschisis](#), a dystrophy that causes splitting of the retina and affects mostly in males; and [age-related macular degeneration](#), the [third-largest cause of vision loss worldwide](#), caused by the interplay between [genetics and environment](#).

Enrolment in a patient registry and [genetic testing](#) advance the design of gene [therapy](#) trials. This in turn benefits blind and visually impaired people.

Research advancement is a concerted effort across the globe—blind and partially sighted people should know they have the power to push it forward.

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