

Discovery of a new mechanism that can lead to blindness

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An important scientific breakthrough by a team of IRCM researchers led by Michel Cayouette, PhD, is being published today by *The Journal of Neuroscience*. The Montréal scientists discovered that a protein found in the retina plays an essential role in the function and survival of light-sensing cells that are required for vision. These findings could have a significant impact on our understanding of retinal degenerative diseases that cause blindness.

The researchers studied a process called compartmentalization, which establishes and maintains different compartments within a cell, each containing a specific set of proteins. This process is crucial for neurons (nerve cells) to function properly.

"Compartments within a cell are much like different parts of a car," explains Vasanth Ramamurthy, PhD, first author of the study. "In the same way that gas must be in the fuel tank in order to power the car's engine, proteins need to be in a specific compartment to properly exercise their functions."

A good example of compartmentalization is observed in a specialized type of light-sensing neurons found in the retina, the photoreceptors, which are made up of different compartments containing specific proteins essential for vision.

"We wanted to understand how compartmentalization is achieved within photoreceptor cells," says Dr. Cayouette, Director of the Cellular

Neurobiology research unit at the IRCM. "Our work identified a new mechanism that explains this process. More specifically, we found that a protein called Numb functions like a traffic controller to direct proteins to the appropriate compartments."

"We demonstrated that in the absence of Numb, photoreceptors are unable to send a molecule essential for vision to the correct compartment, which causes the cells to progressively degenerate and ultimately die," adds Dr. Ramamurthy, who carried out the project in Dr. Cayouette's laboratory in collaboration with Christine Jolicoeur, research assistant. "This is important because the death of [photoreceptor cells](#) is known to cause retinal [degenerative diseases](#) in humans that lead to blindness. Our work therefore provides a new piece of the puzzle to help us better understand how and why the cells die."

"We believe our results could eventually have a substantial impact on the development of treatments for retinal degenerative diseases, like retinitis pigmentosa and Leber's congenital amaurosis, by providing novel drug targets to prevent photoreceptor degeneration," concludes Dr. Cayouette.

According to the Foundation Fighting Blindness Canada, millions of people in North America live with varying degrees of irreversible vision loss because they have an untreatable, degenerative eye disorder that affects the retina. Research aiming to better understand what causes vision loss could lead to preserving and restoring sight.

More information: *The Journal of Neuroscience*
www.jneurosci.org/content/34/42/13976.short

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