Scientist identifies critical role for night blindness gene

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A scientist from the Florida campus of The Scripps Research Institute has determined how a particular gene makes night vision possible

The study, which was published in the August 10, 2011 edition of *The* Journal of Neuroscience, focuses on a gene called nyctalopin. Mutations in the gene result in inherited "night blindness," a loss of vision in low-light environments.

"Until now, our understanding of the role of this gene in the visual signaling pathway has been very limited," said Kirill Martemyanov, an associate professor on the Florida campus of The Scripps Research Institute. "This is the first time we have uncovered a functional role for it-and we linked that function to a much larger molecular complex that's needed for low-light vision."

Quick as a Flash

Our vision begins when photons hit light-sensitive photoreceptor cells in the retina. When excited by light, photoreceptors generate a response that needs to be rapidly transmitted to the downstream neurons (nerve cells) for the signal to be processed and sent to the brain, which then interprets the visual picture. The hand off of the information occurs at the specialized contact points called synapses.

"The proper function of a particular type of synapse between rod photoreceptors and bipolar cells is absolutely critical for the transduction of the visual signal," Martemyanov explained. "Even if rods generate response to light but are unable to properly transmit the signal, this results in an inability to see in the dark. Without this signaling, we'd have a tough time surviving in the world where it is dark half of the time."

In addition, the transmission across the synapse must occur rapidly. "The quickness of our signaling response to light creates a clear temporal resolution of what we see," he said. "For example, when you turn your head suddenly, you see different objects clearly, not just a blur. We couldn't drive a car without it."

In the new research, the scientists searched for proteins associated with nyctalopin in the mouse retina. Scientists had known for a decade that the gene encoding nyctalopin is one of the most frequent culprits of night blindness, but its function had remained a mystery. The results showed that the protein expressed by the gene serves as a kind of molecular glue that holds together key elements of the signal transduction machinery at the synapse, allowing for the rapid and intact transmission of these sensory signals.

In molecular terms, the study strongly suggests that nyctalopin coordinates the assembly and precise delivery to the synapse of the macromolecular complex consisting of mGluR6, a neurotransmitter receptor protein, which directly communicates with rod photoreceptors and TRPM1, a protein channel that generates the response, making vision possible.

While the new findings are relevant to the processing of low-light vision, Martemyanov said, the role of nyctalopin might go far beyond the eye. Proteins similar to nyctalopin exist in the central nervous system, and it is possible that they coordinate synaptic signaling in a manner similar to the retina. Indeed, communication between neurons across synapses is fundamental to the nervous system function and disruption of this process is thought to be the main factor contributing to a range of the neuropsychiatric diseases.

Provided by The Scripps Research Institute