MedicalXpress

Finding hope in the dark

June 10 2015

Advances in stem cell transplantation and gene therapy have been pioneered in vision research. An international team of researchers from Bristol, Toronto, Pittsburgh, Dallas and Montreal have identified a gene that could be responsible for some cases of human night blindness.

Leber's congenital amaurosis (LCA) is a group of hereditary retinal diseases that result in severe loss of vision in early childhood and is estimated to affect around 1 in 80,000 of the population. Recent clinical trials of gene therapy for LCA have shown early promising results in treated patients although their improvements in vision appear to be temporary. Similarly, stem cell transplantation in patients with a range of inherited and acquired retinal disorders is currently undergoing clinical trials and the results are eagerly anticipated after encouraging results in animal models.

The research team recently identified a gene PRDM8 that is also linked to the early loss of night vision in animal models. Often genes that affect the function or survival of the principal light-sensitive cells in the retina, the rod and cone photoreceptors, are responsible for early onset night blindness.

In contrast, PRDM8 appears to influence the inner neural circuitry of the retina that connects the photoreceptors with the rest of the central nervous system. PRDM8 affects the survival of neurons within this retinal circuitry so that they are permanently lost. Consequently, signals from the photoreceptors are not processed correctly through the circuitry of the inner retina causing night blindness.

Dr Denize Atan, Consultant Senior Lecturer in Ophthalmology from the University of Bristol's School of Clinical Sciences and first author on the paper, said: "Our findings suggest that PRDM8 might be responsible for some cases of human night blindness and that PRDM8 is a potential candidate for gene therapy. In addition, our discovery of the importance of this gene to inner retinal circuitry could help researchers in their efforts to generate these neurons for transplantation studies."

More information: 'Transcription factor PRDM8 is required for rod bipolar and type 2 OFF-cone bipolar cell survival and amacrine subtype identity' by Cynthia C. Jung, Denize Atan, David Ng, Lynda Ploder, Sarah E. Ross, Martin Klein, David G. Birch, Eduardo Diez and Roderick R. McInnes in *Proceedings of the National Academy of Sciences* (*PNAS*). DOI: 10.1073/pnas.1505870112

Provided by University of Bristol