

# Discovery of a new retinal gene involved in childhood blindness

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The team of Dr. Robert Koenekoop which includes Dr. Irma Lopez from the Research Institute of the MUHC at the Montreal Children's Hospital played a crucial role in the international collaboration that led to the discovery of a new gene that causes Leber congenital amaurosis (LCA) and retinitis pigmentosa (RP), two devastating forms of childhood blindness.

This finding of this new gene, called SPATA7, is remarkable because it identifies a new retinal metabolic disease pathway that may be crucial for many patients. It also opens a new avenue for a potential genetic therapy. Gene therapy targeting different genes has recently proved successful for the same disease in human subjects. The study will be published on March 5th, 2009 in the *American Journal of Human Genetics*.

## New cell mechanism at play

Researchers have now identified a total of fifteen genes involved in LCA, but SPATA7 is the first gene with a mutation that disrupts the protein transport between two important compartments of the cell: the endoplasmic reticulum and the Golgi apparatus. All proteins in every cell have to pass through this transport pathway; thus SPATA7 plays a major role and its mutation may affect many aspects of vision.

"Until now we were not aware that this cellular mechanism played a role

in LCA or any other eye disease. This is a very important step that opens up a number of new research avenues, particularly in our understanding of the specific cellular processes involved in blindness. This finding also increases the number of potential therapeutic targets and therefore the chances of finding a treatment. We are extremely motivated by all of these new possibilities," explained Dr. Koenekoop.

## **First step towards gene therapy**

"This is an incredible discovery that gives great hope to LCA patients and their families, that gene based therapies can and will be developed to restore sight," said Sharon Colle, President and CEO of The Foundation Fighting Blindness, the leading private charity for vision research. "We are proud to fund such important discoveries involving prominent Canadian researchers and institutions."

A careful assessment of patients with some specific genetic types of LCA also demonstrated that their retinal cells (specifically the rod and cone photoreceptors), although not functional for vision, were still present and in relatively good condition. This critical observation will allow researchers to continue on the path towards gene therapies. Therapies targeting different genes for the same disease have already shown success in the United Kingdom and in the US, meaning that LCA patients can now enjoy hope for the future.

## **A new and innovative technology**

SPATA7 was identified using an innovative technology developed in the different laboratories involved in this international collaboration. "We started this protocol about two years ago, and it has already helped us to identify four new genes associated with LCA and RP before we discovered SPATA7," explained Dr Koenekoop.

The technique is based on DNA-chips and involves three steps: first the genetic material of the patient is screened to find mutations in 14 specific LCA and RP genes. The LCA and RP patients that are negative for this detailed screen are then subjected to a second DNA chip, this one to identify significant stretches of homozygosity in SNP markers. SNPs are single nucleotide repeats, which are natural variations in the human genome. These homozygous regions may contain new genes and are carefully probed based on functional information and then subjected to sequencing. "This method is indeed both very powerful and very promising for the future," said Dr. Koenekoop.

Source: McGill University Health Centre

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