Vision cells, not brain, to blame for colour blindness

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The real culprits of colour blindness are vision cells rather than unusual wiring in the eye and brain, recent research has shown.

The discovery brings scientists a step closer to restoring full colour vision for people who are colour blind – a condition that affects close to two million Australians, says Professor Paul Martin from The Vision Centre and The University of Sydney.

It may also help pave the way for an answer to one of the most common causes of blindness – age-related macular degeneration (AMD), which accounts for half of the legal blindness cases in Australia.

"There are millions of cones in our eyes – vision cells that pick up bright light and allow us to see colour," Prof. Martin says. "They are nicknamed red, green and blue cones because they are sensitive to different wavelengths of light.

"We now know that in the macular region of the eye, each cone has its own 'private line' into the optic nerve and the brain. Just as a painter can mix from three tubes of paint to produce a wide and vivid palette, our brain uses the 'private lines' from the three cone types to create thousands of colour sensations.

Scientists previously thought that full colour vision depends on specialised nerve wiring in the eye and brain, but animal studies show that the wiring is identical for monkeys whether they have normal or

abnormal colour vision, Prof. Martin says.

"This tells us that there's nothing wrong in the brain – it's only working with the signals that it receives on the 'private lines'," he says. "So the only difference in normal and abnormal colour vision is caused by the first stage of sight, which points to faulty cones. Either they have failed to develop, or else they are picking up abnormal wavelengths.

"Now that we know faulty wiring isn't the cause, we can concentrate on fixing the cones, which are controlled by genes – and thus prone to mutation or mistakes during cell replication. There are already promising results from gene therapy as a way to restore full colour vision in colour blind monkeys."

"While we have still have some way to go, the benefits of this gene therapy – if successful – can potentially extend beyond providing complete colour vision," he says.

"If we can get these genes to work in human eyes, it means that the same approach might be possible for other visual problems – including blinding diseases such as macular degeneration."

"In macular degeneration, energy supplies to the macula can't keep up with demand. So the 'private line' system must be very energy-intensive. Gene therapy could be used to turn down the cones' energy demand, or to increase energy supply from supporting cells to cone cells," Prof. Martin says.

"Together with clinical researchers at the Save Sight Institute, we are now working hard to find out exactly how many 'private lines' there are in humans. That can point us to where energy demand is highest and we can target gene therapy to the right place.

"So animal research on 'private lines' for colour vision has given new clues for understanding one of the most important visual diseases – macular degeneration – in humans."

Provided by University of Sydney