Achromatopsia

What is Achromatopsia

Achromatopsia is a rare inherited retinal disease (IRD) which leads to the cone photoreceptors not working well (incomplete achromatopsia) or at all (complete achromatopsia). This results in poor or no colour vision, as well as low vision.

It is known as a stationary IRD. This means that, unlike conditions like retinitis pigmentosa, the condition is not progressive. Most people with achromatopsia will maintain some level of vision through life.

What are the symptoms of achromatopsia?

The condition is often first noticed in a young child by their parents, as children with achromatopsia may dislike bright lights (known as photophobia).

Nystagmus is another symptom of the condition, where their eyes may involuntarily move and "dance" from side to side.

Other symptoms of achromatopsia include:

- colour blindness
- poor central vision and
- high levels of refractive error (requiring high-prescription spectacles or contact lenses).

What is the cause of achromatopsia and how is it inherited?

Achromatopsia is very rare and is thought to affect around 1 in 30,000 people around the world.[1] In Australia, this is roughly 880 people.

Researchers have identified six genes which are known to cause achromatopsia.[2] Two of the most common genes linked to the condition (*CNGB3* and *CNGA3*) account for 75% of achromatopsia cases.[2]

The condition is inherited in an autosomal recessive manner. This means that each parent is a carrier, but is not affected by the disease themselves. Each of their children will have a 25% chance of being affected, with males and females having an equal chance of being affected. The children have a 50% chance of being a carrier (having one healthy and one mutated copy of the gene).

What treatments are available?

Due to the high rates of photophobia (glare intolerance) in people with achromatopsia, it is highly recommended for people with this condition to wear tinted sunglasses and/or brimmed hats to reduce brightness.

There are currently no specific treatments for achromatopsia, but a number of emerging treatments are being developed that may assist people with achromatopsia in the future.

References

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(2)Michalakis S, Gerhardt M, Rudolph G, Priglinger S, Priglinger C. Achromatopsia: Genetics and Gene Therapy. Mol Diagn Ther. Jan 2022;26(1):51-59. doi:10.1007/s40291-021-00565-z

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