Pharmaceutical drugs

In addition to Luxturna® gene therapy for *RPE65*-associated IRD, there is one drug approved for use in people with an inherited retinal disease (IRD) – an antioxidant called Raxone® for people with Leber hereditary optic neuropathy. It has been approved in Europe and Israel but is not available in Australia.

There are also a number of other research studies into pharmaceutical drugs underway which may open up opportunities in the future.

If you would like to know if a pharmaceutical clinical trial is an option for you or your family member, please speak to your ophthalmologist. Please do not take the medications described below without close medical monitoring.

Nutritional supplements and vitamins

It is known that a healthy diet can help protect your eyes. In particular, there has been research showing that lutein, zeaxanthin, and omega-3 fatty acids can slow down disease in some subtypes of age-related macular degeneration (AMD).[1] However, the data on nutritional supplements is complicated. Not all subtypes of AMD benefit from the supplements. In particular, there is no evidence that they can help people with the earliest signs of disease. A systematic review has also shown that omega-3 supplements are not helpful for people with IRDs.[2]

In short, there is evidence that nutritional supplements can help some people with retinal disease. However, they can be dangerous in other IRDs; for example, people with Stargardt disease should not take vitamin A supplementation.[3] As always, you should speak with your doctor before considering any medication, even if they are vitamins.

For now, these results provide good evidence that a healthy diet with leafy green vegetables, should be helpful for people with retinal disease. However, please note that some IRDs have specific dietary requirements (for example, Refsum disease), and so you should always seek personal medical advice from your doctor.

Antioxidant pharmaceutical drugs

The one approved IRD pharmaceutical drug on the market is called idebenone (Raxone®), which is an antioxidant used for the acute treatment of people with Leber hereditary optic neuropathy (LHON). In LHON, people present with sudden vision loss and the use of oral idebenone 900 mg/day for 24 weeks has been shown to have persistent beneficial effects in preventing further vision impairment and promoting vision recovery.[4] Idebenone has been approved in Europe and Israel but is not available in Australia.

Clinical studies are also currently underway in both Australia and the USA on an antioxidant called NAC (N-acetyl cysteine) or NACA (N-acetyl cysteine amide).[5] These trials are recruiting people with retinitis pigmentosa (USA) and Usher syndrome (Australia) to see if the antioxidants can slow the progression of vision loss. Although NAC is available over the counter already, this is a different dosage. As the clinical trials are still incomplete, there is not yet evidence to suggest that people with an IRD should take these pharmaceutical compounds. Results are anticipated in 2024.

There are a number of clinical trials investigating antioxidant drugs from food sources, such as safranal[6] (an extract from saffron) and sulforaphane[7] (found in broccoli) for retinal diseases. Again, there have not yet been clinical trials to show safety and efficacy in people with IRD.

Tinlarebant

Clinical trials are currently underway for a drug called tinlarebant, which is an oral medication that is aiming to slow down progression of Stargardt disease. The trial, called DRAGON, is currently running in Sydney, Melbourne and Perth. The drug works to reduce the accumulation of toxins in the eye, with the aim of protecting the retina and preserving vision.

Optogenetics and molecular photoswitches

These treatment modalities function in different ways, but work towards the same desired end result. In simplistic terms, the therapies aim to change the function of the remaining cells in a diseased retina (such as in IRD), to make them become light-sensitive. In other words, the treatment changes cells to replace the function of the dead photoreceptors.

Clinical trials are underway in Australia for a drug called KIO-301. This trial involves people with no light perception, or bare light perception only, from retinitis pigmentosa and choroideremia to test the medication. This treatment involves ongoing injections, similar to the treatments available for neovascular (wet) age-related macular degeneration.

Antisense oligonucleotides

Antisense oligonucleotides (ASOs) are a class of drugs which bind to ribonucleic acid, or RNA. RNA is a molecule which acts as the messenger that helps carry instructions from our genes (which are made of DNA) to tell the cell what to do. ASO drugs block the activity of RNA, and so stop production of proteins. ASO compounds are currently being tested for a number of -IRDs, including retinitis pigmentosa type 11.

ASOs can be thought of as a form of gene therapy, but rather than targeting the initial genetic code (the DNA), these treatments target the next part of the process, the RNA. There are a number of clinical trials for ASOs internationally, which are expected to come to Australia in 2024.

If you would like to keep up with the IRD clinical trials available in Australia, you can subscribe to Retina Australia's newsletters for updates here.

Neuroprotective pharmaceutical drugs

Given that there are over 300 different genes that cause IRDs, many scientists are working on more general treatments for IRDs, that are not specific to any one gene. One such category of research is in neuroprotective pharmaceutical compounds or drugs. These medications are designed to protect the retinal cells, particularly the photoreceptors, to slow down disease progression.

Retina Australia proudly funded Dr Rabab Rashwan in 2022 on a project to investigate a drug called SAHA. In her study, Dr Rashwan injected SAHA into the eyes of mice with retinitis pigmentosa. She found that the drug did rescue some of the central retina cells and may have a net protective effect, but it did not improve the vision of the mice significantly. The next step is to see whether the drug worked better for some genetic mutations than others – this work is underway.

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