RETINA australia

GENE THERAPY

What are genes?

Chromosomes contain the recipe for making a living thing. They are found in almost every cell's nucleus and are made from strands of DNA. Segments of DNA called "genes" add specific proteins to the recipe. These proteins build, regulate and maintain your body. For instance, they build bones, they enable muscles to move, they control digestion, and they keep your heart beating. It is thought that we have about 20,000 genes in our cells. They make us what we are.

Genes are passed from one generation to the next via children. We have 46 chromosomes in total: each child receives 23 chromosomes from its mother and 23 from its father. Unfortunately, genes can become damaged. If this happens we can suffer illness or even pass an illness or condition to the next generation.

Scientists have discovered that many retinal degenerative conditions are hereditary.

What is gene therapy?

Gene therapies could treat inherited retinal disease by repairing abnormal genes. A disease-causing faulty gene is replaced by putting a "normal" copy into an affected person's cells. Often this can be done by using a harmless virus that has been genetically modified to carry human DNA.

The eye is an ideal organ for gene therapy as it is well protected from the body's immune response. Early successes at clinical trial stage are paving the way for treatments for both inherited and non-inherited forms of blindness in the near future.

In November 2012 a milestone was reached when Glybera®, which is for the treatment of a rare metabolic disorder known as lipoprotein lipase deficiency (LPLD), became the first gene therapy to be granted regulatory approval in Europe. Although this is not directly related to retinal research, it represents a huge step towards the development of registered and approved retinal gene therapies.

What conditions can benefit from gene therapy?

Any condition where the faulty gene that is responsible for the disorder has been identified has the potential to benefit from gene therapy. Examples are some forms of retinitis pigmentosa and Leber congenital amaurosis. Other conditions like age-related macular degeneration that are thought to be partially affected by genes and partially by our environment (e.g. lifestyle, exposure to sunlight) may also benefit from gene therapy in the future.

Blindness is only one disease which researchers are hoping gene therapy will cure. Others include severe combined immunodeficiency, haemophilia, Parkinson's disease, cancer and even HIV.

Gene therapy projects supported by Retina Australia

Retina Australia has recognised the potential of gene therapy for the treatment of inherited retinal diseases and has funded multiple studies in this area. These studies have looked at the processes involved in retinal cell degeneration, including through the use of mouse models.