

**Extract from -**

**Macular Disease Foundation Australia**

**Vision Voice Newsletter Autumn Edition 2018**

**LUXTURNA (voretigene-neparvovec-rzyl)**

In late 2017, the first gene therapy for a rare, inherited disease known as biallelic RPE65 mutation-associated retinal dystrophy was approved by the FDA in the USA. Although this condition is very rare, the treatment, known as Luxturna is the first example of a new type of treatment that is likely to have much wider application in the future for a broad range of other serious eye diseases that are caused by a single gene defect. This includes conditions such as Stargardt's disease and Best's disease, which are forms of macular degeneration affecting younger people.

Genes deliver instructions to enable normal development and functioning of the body, however tiny changes or mutations in a gene can result in incorrect instructions being given. The RPE65 gene is involved in the production of a protein essential for normal vision. Mutations in the RPE65 gene result in significant vision loss, which progressively deteriorates over time.

This new therapy is given as a single injection directly under the retina and works by delivering a 'normal' copy of the RPE65 gene to the retina, which takes over the function of the defective gene. Research has shown that this therapy has the potential to significantly improve functional vision.

Gene therapies using similar technology to Luxturna are being developed for other inherited conditions caused by specific gene mutations. However, it is still unknown when these therapies will become available.

Nevertheless, Luxturna's approval is certainly very encouraging, as it

demonstrates that this technology can work.

It should be noted that this therapy is not appropriate for age-related macular degeneration as this is not caused by a single gene mutation, but is influenced by a wide range of many genetic variations. Research is underway to develop other types of gene therapy for age-related macular degeneration and we will keep you abreast of any new developments as they come to light.