

## The Genetic Basis of Glaucoma

### Part One of Three

*The following article is the first in a series of three articles focusing on genes and genetic research in relationship to glaucoma.*

Many characteristics such as height or hair colour are inherited down through the family line via genetic information contained in every cell in your body. In the same way there is also a chance that you may have inherited glaucoma. Research around the world is now clearly establishing the relationship between mutations in certain genes, and the glaucomas.

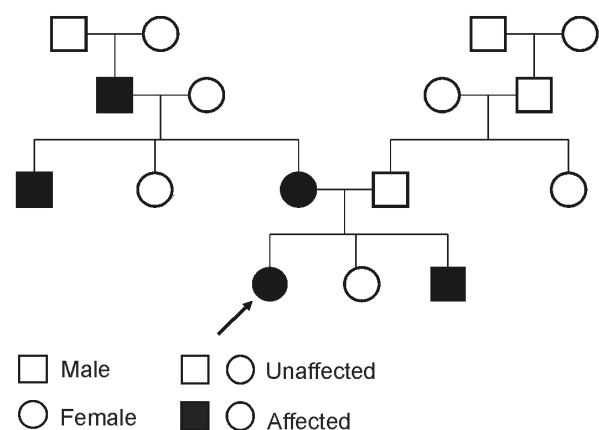
Every cell in our body contains DNA, which is a bit like a blueprint, or a plan, for every component of our physical appearance and our functioning. It contains over 3 billion “letters”, each of which is one of four types (called A, C, G and T) organised in an incredibly complex manner. Every time a cell divides all this information is copied with such exactness that errors are seldom made. Occasionally however, mistakes in this copying process do occur (the equivalent of a “typo”) and these mistakes are called mutations.

Glaucoma has a strong hereditary component, with approximately 40% of all individuals reporting a positive family history. We all carry two copies of every gene and in some cases if one copy of the gene has a mistake in it, this is enough to cause the disease. This mechanism is called autosomal dominant inheritance. As we inherit one copy of a gene from each parent, there is therefore a fifty percent chance that an individual could develop glaucoma, if one of their parents has glaucoma. However new mutations can occur spontaneously.

To date three genes have been identified, accounting for less than 12% of Primary Open Angle Glaucoma (POAG), but research suggests at least six other genes contribute. Identification of glaucoma disease genes will contribute to greater understanding of disease development, and therefore prevention.

So how do we know if we have a mistake in our genes that causes eye disease? The first thing is to understand your family history. If you have a relative (parent, grandparent, aunt or uncle, sibling) who is affected with eye disease it is imperative that you have regular eye checks with an optometrist or an ophthalmologist to detect the disease. At present gene testing is only being done on a research basis here in NZ.

The genes and research will be discussed in further issues of *Eyelight*.



*Figure 1: An example of a pedigree (family tree) with Autosomal Dominant Primary Open Angle Glaucoma*