Division of Human Genetics
University of Cape Town

Inherited Retinal Degenerative Disorders

What is Retinal degeneration?

Retinitis Pigmentosa and retinal degeneration are the terms used for a group of disorders that affects the **retina**. The retina lines the back of the eye. It acts like the film in a camera, receiving and processing everything that a person sees. The retina is made up of light sensitive cells, which **receive the pictures that we see and transmits them to the brain**. Figure 1 illustrates the position of the retina within the eye.

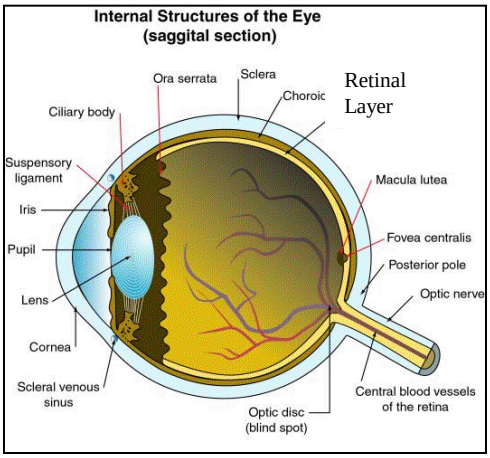

Figure 1: A diagram of the anatomy of the eye. (Picture from MSCD Biology Department Faculty website: clem.mscd.edu/~biology/231course/ bio231info.htm)

The light sensitive cells in the retina are called rods and cones. Rods pick up **movement** and enable a person to **see at night or in poor lighting conditions**. Rods are more numerous towards the **outer edge of the retina**. Cones are used for colour vision and in close precision work, like reading. Cones are concentrated in the centre of the retina (around the macula), but can also be found throughout the retina.

Retinal degeneration may be caused by a **deterioration in the function of the rods or cones** in parts of the retina. When rods predominantly deteriorate, the condition is known as Retinitis Pigmentosa and when cones deteriorate the condition is known as Macular Degeneration.

Individuals affected with Retinitis Pigmentosa usually complain of an increased difficulty in seeing in dimly lit places (often called "night blindness"). This is then followed by loss of peripheral or side vision, resulting in a "tunnel vision" effect. In some cases, total blindness can result. Other symptoms include slow light to dark adaptation times (eyes take longer to adapt to the dark) and poor contrast sensitivity. Within the classical picture of retinal degeneration there is a high degree of variability especially in age of onset, rate of progression of the deterioration and end-stage vision.

What other names are associated with Inherited Retinal Degenerative Disorders?

- Retinitis Pigmentosa
- Macular Degeneration
- Usher Syndromes
- Leber Congenital Amaurosis
- Cone Rod Dystrophy
- Rod Cone Dystrophy
- Sorbus Fundus Dystrophy

How common are Retinal Degenerative Disorders?

It is estimated that 1 in every 80 South Africans are carriers (not affected, but have the gene fault) for Autosomal Recessive Retinitis Pigmentosa. Twenty five percent of Caucasians over the age of 65 are at-risk for age related Macular Degeneration.

How Do People Inherit Retinal Degenerative Disorders?

Retinal degeneration can be inherited or passed on by means of **Autosomal dominant**, **Autosomal recessive or X-linked recessive inheritance.** (Refer to Fact sheets 9,10,11)

What genes are related to Retinal Degenerative Disorders?

There are many genes that have an effect on the functioning of the retina. Research is continuously discovering more genes that cause the various types of retinal degeneration.

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RETINA SOUTH AFRICA
Fighting Blindness

Genetic Testing and Retinal Degenerative Disorders :

The Division of Human Genetics at the University of Cape Town is currently doing research on families with retinal degenerative disorders. Certain faults, known as mutations, have been identified to occur in families with retinal degenerative disorders. If one of these faults can be identified in an affected person (known as a diagnostic test), other blood relatives can be offered genetic testing (known as a predictive test) to see if they have the same mutation. If mutations are not found then linkage analysis may be done (a different test that can be done without knowing where the fault has occurred) depending on the family structure. The DNA can also remain in the research laboratory for further investigation if no mutation has been found.

How long before I get a result?

Screening genes for these faults is a time-consuming task. To streamline the process the research group only focus their attention on a few specific types of retinal degenerative disorders at a time. As new genes are continuously being discovered, samples are retested if no mutations have been found during the previous testing. As a result it may take years to find the mutation that causes a retinal degenerative disorder in a family.

What are the criteria for involvement in the research program for Retinal Degenerative Disorders?

The research program involves individuals (over the age of 18 years) and families affected with different forms of inherited retinal degenerative disease.

What Are The Important Concerns In Genetic testing?

Genetic counselling can provide you with information concerning the chance that you have inherited the mutation (fault) that would result in a retinal degenerative disorder. The counsellor can also discuss the various options for testing that may be available to you and provide support as well as further information about the condition.

RETINA SOUTH AFRICA
Fighting Blindness

Is there treatment available for retinal degenerative disorders?

Currently there is no known treatment that alters the course of the disease, although clinical trials are underway for different gene-based therapies. Knowing the specific gene mutation involved is a prerequisite for participation in any gene-based therapy trial. Pharmacological agents may become available but all potential treatment options are still at the trial phase (http://www.clinicaltrials.gov/).

Where can I read more about Retinal Degenerative Disorders?

You may find the following resources about Retinitis Pigmentosa helpful.

- Retina South Africa: http://www.rpsa.org.za
- Genetics Home Reference website: http://ghr.nlm.nih.gov/
- The British Retinitis Pigmentosa Society: http://www.brps.org.uk/Home_White.html
- Centre for genetics education: http://www.geneclinics.org/profiles
- Foundation Fighting Blindness www.blindness.org
- Division of Human Genetics http://www.humangenetics.uct.ac.za/

Is there a support group for Retinal Degenerative Disorders?

YES-

Retina South Africa
P.O. Box 40432
Cleveland
2022
Tel: (011) 622-4904
Fax: (011) 622-6277
SHARE CALL: 0860 59 59 59
e-mail: national@rpsa.org.za
Website: www.rpsa.org.za

Who do I contact for more information regarding testing?

Division of Human Genetics Molecular Laboratory:
Prof. Jacquie Greenberg: (021) 406-6299
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