RETINA
SOUTH AFRICA

a Cure in Sight for Blindness

A BASIC GUIDE TO

Retinal Conditions

Developed by Retina South Africa

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Knowledge is power. The more you know about your eye condition, the better you will be able to manage all aspects that could affect the health of your eyes.

This booklet is the first step in the journey to obtaining that vital knowledge.

Dr H. Tayob
Ophthalmologist, Pretoria

About Retina South Africa

Retina South Africa is the only patient-led advocacy and support charity for individuals with a wide range of retinal conditions in South Africa.

This booklet is designed as a guide for individuals and families impacted by a retinal condition, providing an overview of common retinal conditions.

This guide should not be considered a substitute for professional advice from your eye care provider. If you suspect a retinal eye condition, it is important to consult an eye care doctor.

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1 Basic Anatomy of the Eye

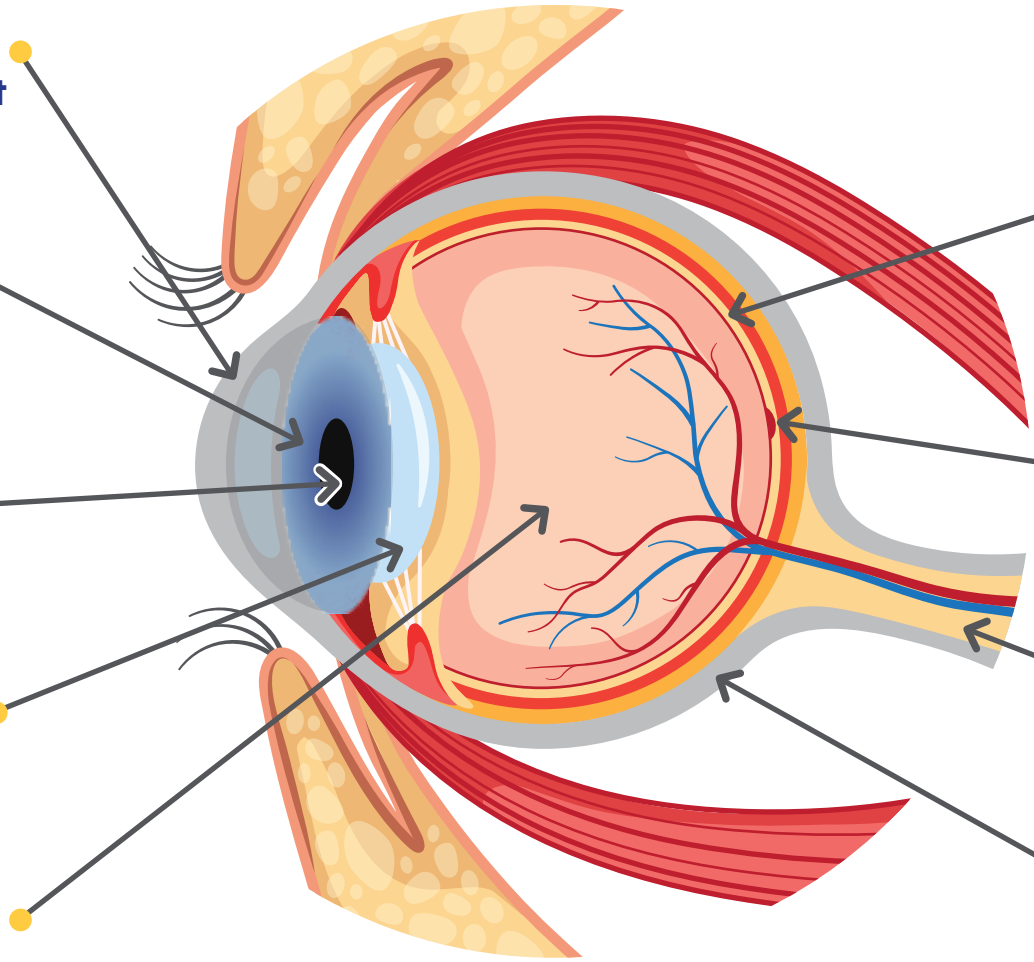
Cornea
Clear front surface that helps protect the eye and focus incoming light

Iris
Coloured part of the eye that controls the size of the pupil

Pupil
Changes size to control the amount of light entering the eye

Lens
Focuses light rays onto the retina

Vitreous (gel)
Transparent area that maintains the shape of the eyeball and supplies nutrients



Retina
Thin layer of cells (cones and rods) that react to light and send signals to the brain to create the image we see

Macula
Part of the retina that creates the sharpest, most detailed part of the image we see

Optic nerve
Carries light signals to the brain

Sclera
Tough, white outer layer that covers the eyeball

What is the retina?

The retina, located at the back of the eye, includes a layer of cells (cones and rods) that react to light and send signals to the brain to create the image we see.

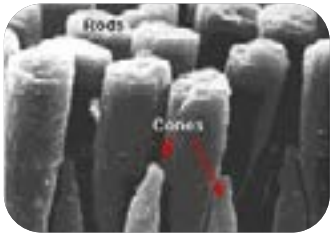
Cones

- Cones are found at the centre of the retina
- They allow us to see colour and focus on fine details, which helps close-up work like reading and recognising faces
- Cones function best in bright light conditions

Rods

- Rods are found around the edges of the retina
- They allow us to see in low light conditions and out of the corners of our eyes
- Unlike cones, rods can only detect black, white and grey, and produce images that are fuzzier

Clinical retinal image of a healthy retina



Magnified image of cones and rods

There are many conditions that can affect the retina, each of which affects different people in different ways.

This booklet provides an overview of the following common retinal conditions:

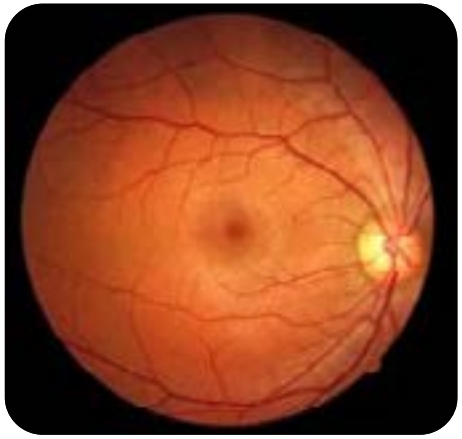
4 Age-Related Macular Degeneration

What is age-related macular degeneration (AMD)?

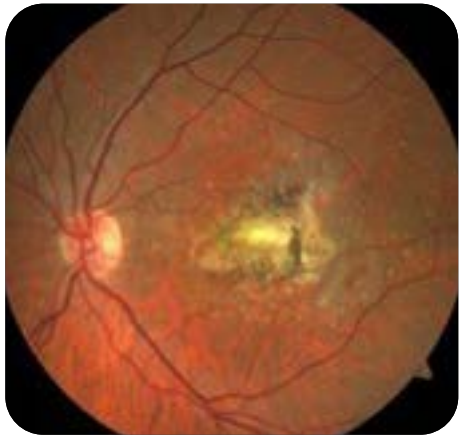
The macula is the part of the retina that creates the sharpest, most detailed part of the image we see. When our macula becomes damaged, it may be due to AMD. People with AMD often find that activities requiring clear, detailed vision, such as reading, writing, driving and recognising faces, may become difficult.

AMD is a common cause of visual loss in people older than 50. Some types of AMD develop rapidly and require immediate attention. If you notice any central vision loss, consult your eye care doctor as soon as possible.

See below for clinical retinal images of AMD:



Clinical retinal image of a healthy retina



Clinical retinal image of AMD

What types of AMD are there?

There are two main types of AMD – dry and wet. Dry AMD is more common: around 8 in 10 people with AMD have dry AMD.

1 Dry AMD

Dry AMD usually develops slowly. It is caused by a build-up of fatty deposits in the retina. People with dry AMD may gradually lose their sight as these fatty deposits grow.

2 Wet AMD

Wet AMD can develop quickly. It is caused by unusual growth of tiny, fragile blood vessels behind the macula. People with wet AMD can lose vision as these vessels leak or experience dramatic vision loss if they burst. When this happens, people need immediate treatment.

What factors could increase the risk of AMD?

- Older age
- Smoking andvaping
- Pollution
- Exposure to blue light (screens like TVs and computers)
- Family history of AMD
- Diet lacking essential nutrients
- Over-exposure to sunlight

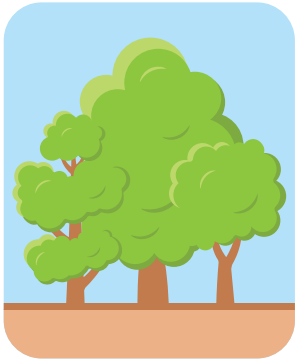
4 Age-Related Macular Degeneration

What are the symptoms of AMD?

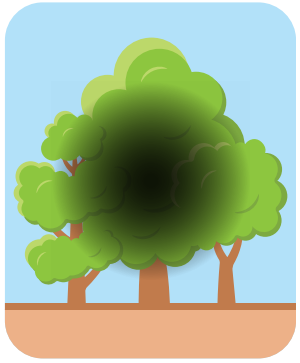
At first, people with AMD may hardly notice any effect on their vision. However, as time passes and the condition progresses, their central vision may become blurry. This means reading, recognising faces and other close-up tasks become harder.

People with wet AMD may notice more sudden vision changes, such as straight lines appearing wavy or distorted, and spots of vision loss.

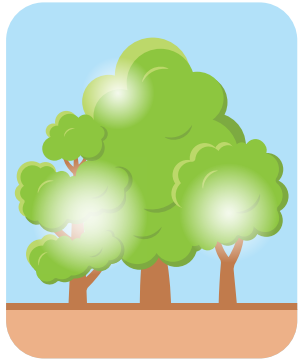
Illustrations of possible vision loss experienced by someone with AMD are shown below:



Normal vision



Blind spot in centre of the field of vision



Unusual fuzzy or distorted vision

The Amsler Grid (page 14) is a square-shaped grid that can be used to check for changes in your central vision, such as:

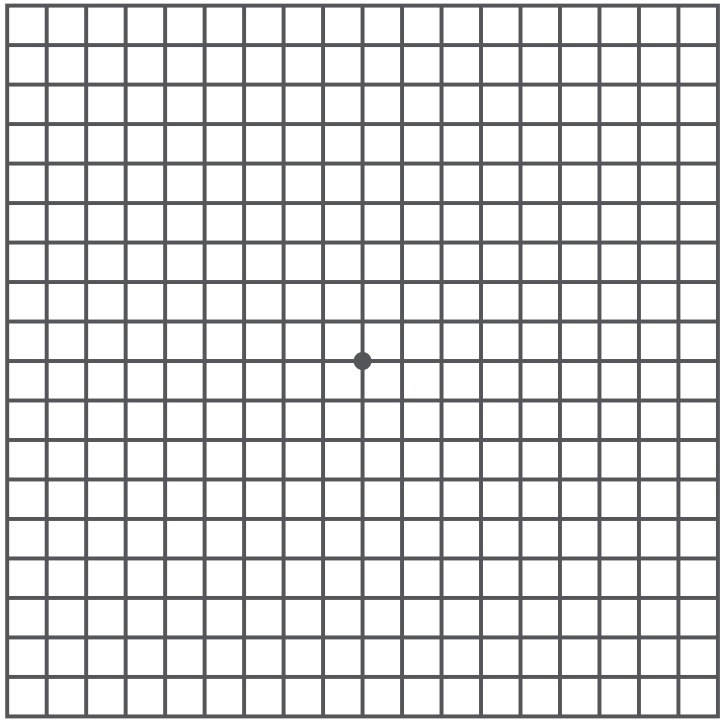
- Wavy, bent or distorted lines
- Missing or blank spots in what you see

Check your vision regularly with the Amsler Grid as guided by your eye care team. If you notice any changes, contact an eye doctor immediately.

4 Age-Related Macular Degeneration

How to use the Amsler Grid:

1. Hold the grid at a comfortable reading distance (approximately 30 cm). Wear your reading glasses or contact lenses
2. Cover one eye and focus on the black dot in the middle of the grid. Cover the other eye and repeat the test
3. If the lines appear to be wavy, broken, dim or fuzzy, schedule an appointment with an eye care doctor urgently



How to manage AMD?

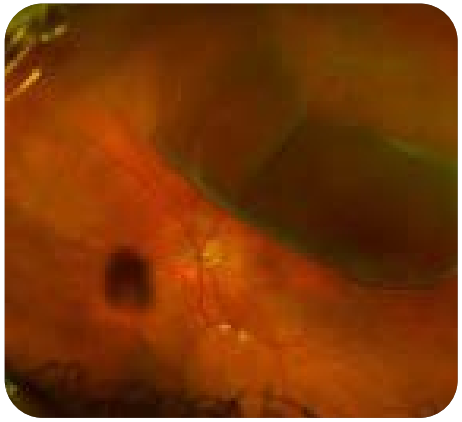
- Have regular eye examinations – especially if you are over the age of 40 or have a family history of AMD. Ask your eye care professional to examine your macula at every visit
- Maintain a healthy body weight
- Eat a balanced, colourful diet. Include a variety of “rainbow” fruits and vegetables, particularly green leafy vegetables, as well as fish, avocados and olives. Limit foods that are high in saturated fat
- Exercise regularly
- Ask your eye care professional whether a nutritional supplement may be beneficial for you. There are new supplements that could assist you
- Do not smoke. Smoking is one of the most important modifiable risk factors for AMD
- Protect your eyes from the sun. Wear sunglasses that provide UV protection and/or a wide-brimmed hat when outdoors
- If you have wet AMD, effective treatments are available. Special anti-VEGF injections can slow or stop vision loss and, in some people, may improve vision. Your ophthalmologist will advise you on whether these injections are appropriate and how often they are needed

5 Diabetic Retinopathy

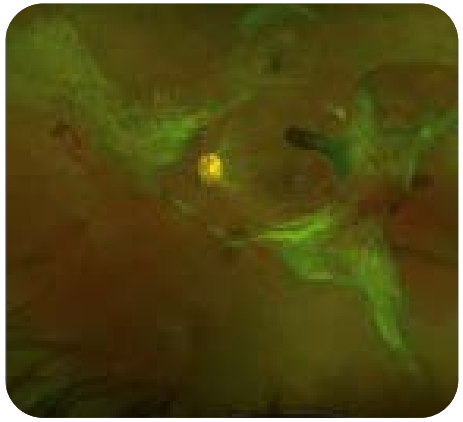
What is diabetic retinopathy?

Diabetic retinopathy, or diabetes-related vision loss, occurs when high blood sugar (glucose) damages the tiny blood vessels in the retina. It is one of the more common complications of diabetes and is a leading cause of blindness in working-age people worldwide.

See below for clinical retinal images of diabetic retinopathy:



Clinical retinal image of diabetic retinopathy with bleeding

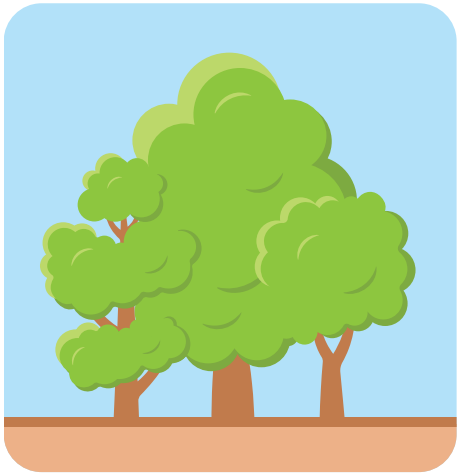


Clinical retinal image of diabetic retinopathy with abnormal blood vessels

Causes of diabetic retinopathy

The risk of diabetic retinopathy is higher when blood glucose is not well-controlled, but damage to the eye may start before a diagnosis of type 2 diabetes has even been made. High blood glucose can weaken and damage tiny blood vessels in the eye. Raised cholesterol levels can also cause or add to this damage. The damaged blood vessels can leak into the retina, harming the light-sensing cells (cones and rods). Sometimes, the blood vessels can also burst and cause a bleed (haemorrhage).

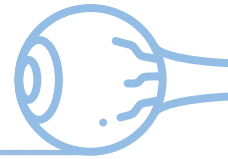
Illustrations of possible vision loss experienced by someone with diabetic retinopathy are shown below:



Normal vision

Blurry vision with dark spots or “floaters”

5 Diabetic Retinopathy



Stages of diabetic retinopathy

Diabetic retinopathy has four stages.

1 Mild non-proliferative retinopathy

Tiny weak areas, called microaneurysms (small, balloon-like bulges) develop in the retina's tiny blood vessels.

2 Moderate non-proliferative retinopathy

During this phase, blood vessels that feed the retina become blocked, causing areas to be deprived of nourishment.

3 Severe non-proliferative retinopathy

More blood vessels become blocked, leading to more areas of the retina becoming deprived of oxygen and nourishment. The retina sends signals for help to ensure that the area is oxygenated and nourished.

4 Proliferative retinopathy

At this advanced stage, the signals sent by the retina for nourishment trigger the growth of new blood vessels. These new blood vessels are abnormal and fragile and can start to leak or burst, causing vision loss.

Who is at risk?

Anyone who has diabetes can develop diabetic retinopathy, but the risk may increase due to:

- Pregnancy
- Tobacco use
- High blood pressure
- Long-standing diabetes (including undiagnosed or 'pre-diabetes')
- Being of African descent
- High cholesterol
- Poor blood glucose control

Prevention of diabetic retinopathy

You can't always prevent diabetic retinopathy. However, regular eye examinations, good control of your blood sugar and blood pressure, and early intervention for vision problems can help prevent severe vision loss.

If you have diabetes, you can reduce your risk of getting diabetic retinopathy by doing the following:

Blood glucose control

- Always take your medications for diabetes as directed by your doctor
- Make sure that you are aware of factors that could impact your blood glucose level

- Try to make healthy food choices (brightly coloured foods such as fruits and vegetables)

Blood pressure and cholesterol control

- Eating a brightly coloured diet (i.e. fruits and vegetables), engaging in regular physical activity and taking prescribed medications on time are essential to keep you healthy

Smoking and/or vaping

- Avoid smoking and/or vaping, as both increase your risk of diabetes-related complications, including diabetic retinopathy

Alcohol

- Avoid excess alcohol

Pay attention to vision changes

- If your vision changes (e.g. by becoming blurry, wavy or spotty), contact your eye care doctor immediately

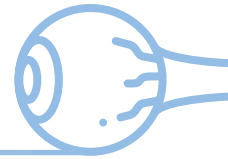
What is an inherited retinal condition?

Retinal conditions can sometimes be inherited, which means they are passed down from parents to their children. This is because we all receive a set of genetic instructions from both our parents. This 'set of instructions' is stored in our DNA, which is organised into 46 packets of information called chromosomes.

These 46 chromosomes are arranged in 23 pairs. We inherit one copy of chromosomes 1 to 22 from each parent. The final pair is the sex-chromosome pair. If we are female, we inherit an X chromosome from both parents, and if we are male, we inherit an X chromosome from our mother and a Y chromosome from our father.

DNA is divided into smaller sections called genes. Each gene gives the body instructions to make a specific protein that helps our cells work properly. Sometimes changes (called mutations) occur. Some changes are harmless, but major changes can cause disease. Changes in genes can be passed down in different ways, known as patterns of inheritance. For retinal conditions, three main patterns are seen: recessive, dominant and X-linked inheritance.

6 Inherited Retinal Conditions



What is recessive inheritance?

The most common pattern by which retinal conditions are inherited is called recessive inheritance. Here, a child needs to inherit two mutated copies of the same gene (one from each parent) to develop the disease. A child who has only one mutated copy is called a ‘carrier’ and usually does not have symptoms.

What is dominant inheritance?

In dominant inheritance, only one changed copy of the gene, inherited from either parent, is enough for the child to inherit the disease.

What is X-linked recessive inheritance?

In X-linked recessive inheritance, the gene change is on the X chromosome. Mothers who carry the change on one of their X chromosomes usually do not have symptoms because they have a second working X chromosome. Their daughters may also inherit the changed X chromosome and become carriers. However, if their sons inherit the X chromosome with the change, they may develop the disease because they only have one X chromosome.

Genetic counselling

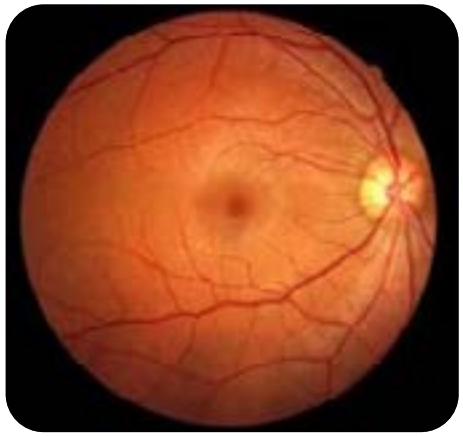
A visit to a genetic counsellor is advised to ensure that you understand your specific condition and the risks to future children and other family members. Genetic counselling costs are usually reimbursed by medical aids. Genetic counselling is available at some state hospitals. A genetic test to identify the specific gene mutation may be recommended. Ask Retina South Africa for more details.

Retinitis pigmentosa (RP)

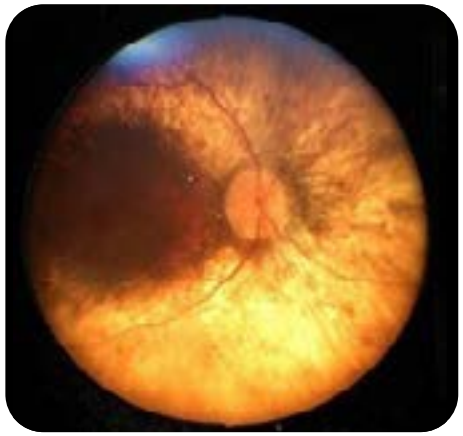
What is RP?

RP is a group of inherited retinal conditions that cause progressive vision loss due to deterioration of cones and rods in the retina. RP initially affects the rods but cones may deteriorate later in the disease. There are over 130 gene mutations that can cause some form of RP; the age of onset and rate of degeneration differ greatly between affected individuals. RP can be inherited in either a recessive, dominant or X-linked mode. The type of inheritance may also influence the course of the disease. It is thought to affect around 1 in 3,000 people.

See below for a clinical retinal image of RP:



Clinical retinal image of a healthy retina



Clinical retinal image of RP

Illustrations of possible vision loss experienced by someone with RP are shown below:

Normal vision

Loss of peripheral vision (tunnel vision)

How to manage RP

A diet rich in antioxidants (found in many fruits and vegetables) may slow the course of the disease, and special supplements are available. Please discuss any supplementation with your eye care doctor. Additionally, rehabilitation programmes, such as low vision support and training to help with everyday tasks and mobility, can be beneficial at any stage of the condition.

Usher syndrome

What is Usher syndrome?

Usher syndrome is RP accompanied by loss of hearing. It is a recessively inherited condition, and is thought to affect 1 in 10,000 to 1 in 12,000 people.

There are three different types of Usher syndrome:

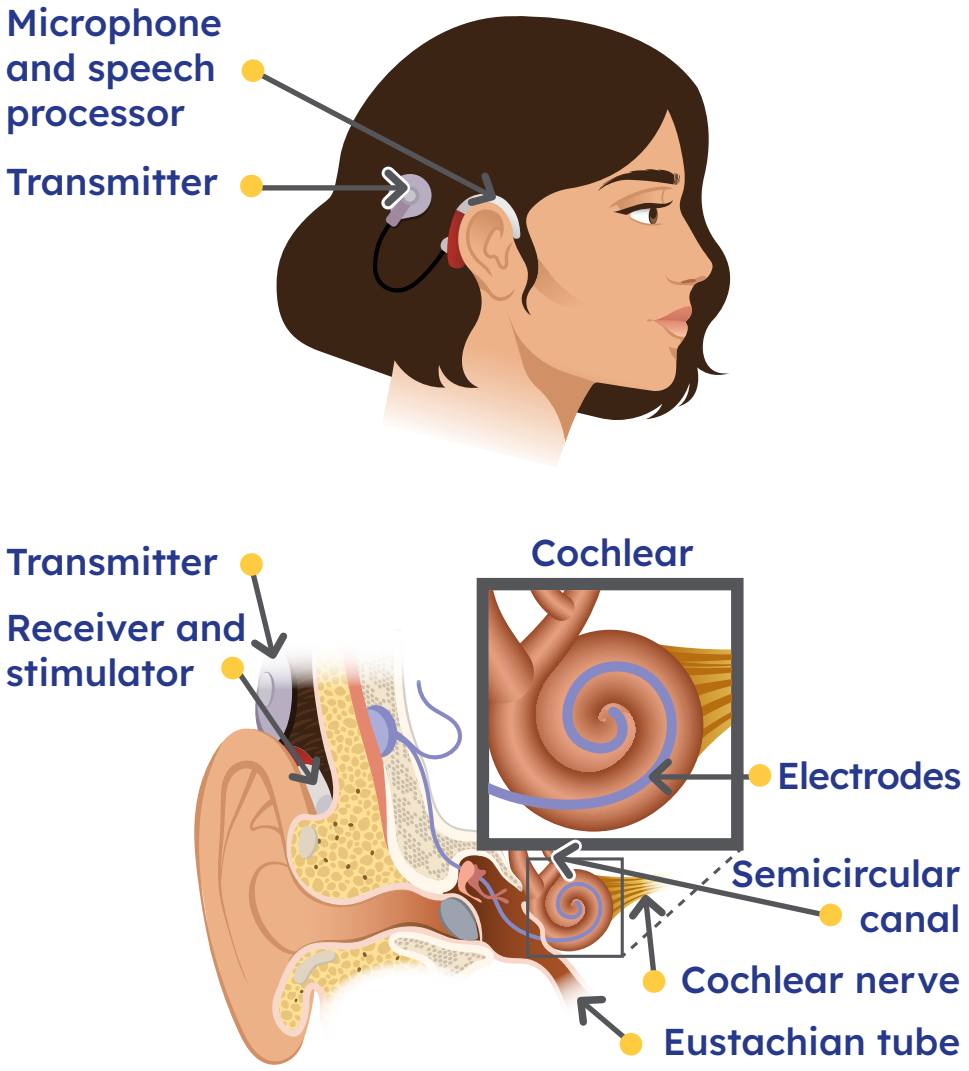
Usher type 1 is profound loss of hearing from birth with later retinal vision loss.

Usher type 2 is less severe hearing loss and a variable age of retinal vision loss.

Usher type 3 is rare and used to describe a later onset of both hearing and vision loss.

How to manage Usher syndrome

Cochlear implants, shown in the image on page 27, are commonly used in people with Usher syndrome to improve hearing. A diet rich in antioxidants (found in many fruits and vegetables) may slow the course of vision loss, and special supplements are available. Please discuss any supplementation with an eye care doctor. Additionally, rehabilitation programmes, including support for both hearing and vision as well as training for communication, mobility and daily activities, can be effective at any stage of the condition.



A cochlear implant system (hearing device)

Stargardt disease

What is Stargardt disease?

Stargardt disease causes progressive loss of central vision, usually beginning in childhood or adolescence. It starts with the build-up of a waste product (called lipofuscin) in the retinal pigment epithelial layer. Over time, this damages cones (the cells responsible for sharp, detailed vision needed for activities like reading and writing), though colour vision is not always compromised.

Stargardt disease is generally a recessively inherited condition and is thought to affect 1 in 10,000 people. In most cases, it is caused by a change in a gene called ABCA4 on Chromosome 1. This change affects how the retina uses vitamin A.

There are other less common genetic forms of Stargardt disease, and a definitive diagnosis of Stargardt disease may only be possible via a genetic test. Retina South Africa can help facilitate this.

Illustrations of possible vision loss experienced by someone with Stargardt disease are shown below:

Normal vision | Dark spots or patches in field of view | Progressive loss of central vision

How to manage Stargardt disease

Children with early-onset Stargardt disease may require additional support or adaptations to ensure their education is not compromised. Assistive technology, dark-lined paper, and black thin-liner koki pens can all be helpful. Additionally, people with Stargardt disease are usually advised to avoid extra vitamin A, such as vitamin A supplements and foods that are very high in vitamin A (for example, animal liver products). Foods containing beta-carotene (a vitamin A precursor found in many orange and green vegetables) may be eaten in moderation.

Cone-rod dystrophy

What is cone-rod dystrophy?

Cone-rod dystrophy is a group of conditions that affect cones and rods in the retina. It can look very similar to Stargardt disease, especially in the early stages. In some people, cone-rod dystrophy is also caused by a change in the ABCA4 gene. However, changes in several other genes can also cause cone-rod dystrophy, including CRX, GUCY2D and RPGR.

The major difference between cone-rod dystrophy and Stargardt disease is the early involvement of the rods. Because of this, people often notice difficulty with seeing and navigating in low-light conditions.

How to manage cone-rod dystrophy

As for Stargardt disease, children with cone-rod dystrophy should avoid Vitamin A supplementation.

Leber congenital amaurosis (LCA)

What is LCA?

LCA is an early-onset condition that affects the retina from birth or very early in life; it is often diagnosed in infancy or early childhood. Severe vision loss may be accompanied by rapid eye movements, called nystagmus. There are over 20 different genetic forms of LCA. In most cases, LCA follows a recessive pattern of inheritance.

Illustrations of possible vision loss experienced by someone with LCA are shown below:

Normal vision

Blind spot at peripheral vision

Progressive vision loss

Other genetic retinal conditions

There are dozens of other rare conditions not covered in this booklet. Reach out to Retina South Africa for more information concerning these, including:

- Achromatopsia
- Retinoschisis
- Refsum disease
- Choroideremia
- Malattia Leventinese
- Gyrate atrophy
- Macular dystrophies
- Rod-cone dystrophy
- Sorsby fundus dystrophy
- Bardet-Biedl syndrome
- Fundus albipunctatus
- Leber hereditary optic neuropathy
- Best vitelliform macular dystrophy
- Blue cone monochromacy

Global research efforts over many years have increased the understanding of these complex eye conditions. One gene therapy for a type of LCA has been approved in the USA and Europe. Although this treatment is extremely expensive, access may be possible for eligible patients through national healthcare systems or insurance coverage. Another treatment for LCA type 4 has just been announced.

Many clinical trials are showing great promise [www.clinicaltrials.gov], and the major areas are:

Gene therapies

- Achromatopsia, AMD, Battens disease, Choroideremia, LCA, RP, Retinoschisis

Cell-based therapies [including stem cells]

- AMD, RP, Usher syndrome

Small molecules, proteins and inhibitors

- AMD, LCA, RP, Stargardt disease, Usher syndrome

Contact Retina South Africa or an eye care doctor for more information.

8 Assistance

Retina South Africa has trained counsellors and social workers who will give you advice and information on your condition.

Many service providers can give you advice on assistive devices and computer-based technologies.

Orientation and mobility training and the use of free smartphone apps can transform your life.

You can contact Retina South Africa using the following details:

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