

2021

# Symptoms Related to Eye Diseases and Conditions

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## Symptoms List

- **Blurred and/or distorted vision:** [Age-Related Macular Degeneration \(AMD\)](#); [Aniridia](#); [Best Disease](#); [Birdshot Uveitis](#); [Cataracts](#); [Coat's disease](#); [Corneal Dystrophies](#); [Fuch Dystrophy](#); [Keratokonius](#); [Nystagmus \(children\)](#); [Cone/Rod Dystrophy](#); [Acanthamoeba Keratitis](#); [Behcet's Disease](#); [Blepharitis](#); [Central Serous Retinopathy \(CSR\)](#); [Corneal Disease](#); [Diabetic Macular Edema \(DME\)](#) and [Diabetic Retinopathy \(DR\)](#); [Dry Eye Disease](#); [Leber Hereditary Optic Neuropathy \(LHON\)](#); [Macular holes](#); [Meesmann Corneal Dystrophy](#); [Retinal Detachment](#); [Stroke](#); [Uveal Melanoma \(anterior\)](#); [Uveitis](#); [Meibomian Gland Dysfunction](#); [Retinal Vein Occlusion](#), [Central Serous Retinopathy \(CSR\)](#); [Corneal Disease](#); [Macular holes](#).
- **Burning, itchy, Irritation, watery eyes:** [Allergic](#) or [Chronic](#) or [Bacterial](#) or [Viral Conjunctivitis](#); [Conjunctival Hyperemia](#); [Dry Eye Disease](#); [Meesmann Corneal Dystrophy](#); [Ocular Mucous Membrane Pemphigoid](#); [Pterygium](#); [Uveitis](#); [Thyroid Eye Disease](#); [Meibomian Gland Dysfunction](#).
- **Double vision and/or patchy vision:** [Graves' Disease](#); [Keratokonius](#); [Giant Cell Arthritis](#); [Ocular Myasthenia Gravis](#); [Stroke](#); [Thyroid Eye Disease \(TED\)](#).
- **Difficulty looking straight ahead:** [Amblyopia](#); [Duane's Retraction Syndrome \(DRS\)](#); [Strabismus](#).
- **Crossed Eyes:** [Retinopathy of Prematurity](#).
- **Eye inflammation or swelling:** [Birdshot Uveitis](#); [Behcet's Disease](#) or Syndrome (systemic); [Graves' disease](#); [Blepharitis](#); [Retinoblastoma](#).
- **Eye pain:** [Birdshot Uveitis](#); [Fuch Dystrophy](#); [Glaucoma](#); [Graves' disease](#); [Acanthamoeba Keratitis](#); [Behcet's Disease](#); [Corneal Eye Disease](#); [Optic Atrophy](#); [Thyroid Eye Disease \(TED\)](#); [Uveitis](#).
- **Eye redness:** [Acanthamoeba Keratitis](#); [Behcet's Disease](#); [Conjunctival Hyperemia](#); [Pterygium](#); [Retinoblastoma](#); [Thyroid Eye Disease \(TED\)](#); [Uveitis](#); [Meibomian Gland Dysfunction](#).
- **Eyelid inflammation:** [Blepharitis](#); [Bacterial Conjunctivitis](#).
- **Eyelid swelling and/or ulceration:** [Eyelid Cancer](#); [Graves' disease](#); [Thyroid Eye Disease \(TED\)](#); [Meibomian Gland Dysfunction](#).
- **Dry/itchy/gritty and/or sore eyelids:** [Meibomian Gland Dysfunction](#).

- **Flashing lights (Photopsia) and/or floaters (black spots):** [Azoor](#); [Birdshot Uveitis](#); [Central Serous Retinopathy](#); [Diabetic Macular Edema \(DME\)](#) and [Diabetic Retinopathy \(DR\)](#); [Retinal Detachment](#); [Uveal Melanoma](#); [Uveitis](#).
- **Hallucinations:** [Charles Bonnet Syndrome](#).
- **Headaches; fatigue:** [Refractive error](#); [Strabismus](#).
- **Hearing and sight loss (ref Retinitis Pigmentosa):** [Usher Syndrome](#).
- **Involuntary eye movements (Jittery vision):** [Stroke](#); [Strabismus](#); [Leber Congenital Amrosis](#); [Nystagmus](#); [Retinopathy of Prematurity](#); [Congenital Blindness](#).
- **Lack of eyes or small eyes:** [Anophthalmia and Microphthalmia](#).
- **Lack of responsiveness:** [Leber Congenital Amaurosis \(LCA in infants\)](#); [Nystagmus](#): (Infants).
- **Loss of acuity and detail:** [Aniridia](#); [Diabetic Retinopathy](#); [Stargardt disease](#); [Leber Hereditary Optic Neuropathy LHON](#); [Uveal Melanoma](#); [Sorsby Fundus Dystrophy](#); [Congenital Blindness](#).
- **Partial or complete loss of central vision:** [Age-Related Macular Degeneration \(AMD\)](#); [Coat's Disease](#); [Leber's Hereditary Optic Neuropathy \(LHON\)](#); [Stargardt](#); [Sorsby's Fundus Dystrophy](#); [Diabetic Macular Edema \(DME\)](#) and [Diabetic Retinopathy \(DR\)](#); [Stargardt Macular Degeneration](#); [Retinal Vein Occlusion](#).
- **Lack of (total or partial) colour vision:** [Achromatopsia](#) or [Colour Blindness](#).
- **Loss of colour perception:** [Birdshot Uveitis](#); [Diabetic Macular Edema \(DME\)](#) and [Diabetic Retinopathy \(DR\)](#); [Leber Hereditary Optic Neuropathy LHON](#); [Optic Atrophy](#); [Stargardt Macular Degeneration](#); [Wolfram Syndrome](#); [Sorsby's Fundus Dystrophy](#).
- **Loss of depth perception:** [Amblyopia](#) ("Lazy eye"); [Birdshot Uveitis](#); [Stroke](#); [Central Serous Retinopathy](#); [Eye cancer](#) (Loss of 1 eye).

- **Loss of field of vision:** [Azoor](#); [Giant Cell Arthritis](#); [Leber Hereditary Optic Neuropathy](#); [Optic Atrophy](#); [Proliferative Vitreoretinopathy \(PVR\)](#); [Uveal Melanoma](#); [Wolfram Syndrome](#); [Stroke](#); [Uveal Melanoma](#); [Uveitis](#).
- **Loss of peripheral vision:** [Choroideremia](#); [Retinitis Pigmentosa \(RP\)](#); [Glaucoma](#); [Diabetic Retinopathy](#); [Retinal Detachment](#); [Best Disease](#); [Birdshot Uveitis](#); [Wolfram Syndrome](#) (Primary Optic Atrophy and/or [Leber Hereditary Optic Atrophy](#)); [Sorsby's Fundus Dystrophy](#).
- **Muscle twitching:** [Dystonia/Blepharospasm](#).
- **Night blindness; difficulty in low light:** [Retinitis Pigmentosa \(RP\)](#); [Birdshot Uveitis](#); [Choroideremia](#); [Dry Eye Disease](#); [Stargardt Macular Degeneration](#); [Sorsby Fundus Dystrophy](#).
- **No light perception:** [Optic Nerve Hypoplasia](#).
- **Sensitivity to light and glare (photophobia):** [Amblyopia](#) (Lazy eye); [Nystagmus](#); [Age-Related Macular Degeneration \(AMD\)](#); [Albinism](#); [Alstrom Syndrome](#) (systemic); [Coloboma](#); [Dystonia/Blepharospasm](#); [Keratokonus](#); [Acanthamoeba Keratitis](#); [Birdshot Uveitis](#); [Conjunctivitis](#).
- **Loss of quick response:** [Cerebral \(or Cortical\) Visual Impairment \(CVI\)](#).
- **Squinting and/or eye strain:** [Amblyopia](#); [Refractive error](#); [Strabismus](#).
- **Visual processing** (e.g object/facial/currency recognition): [CVI](#); See Systemic disease.

*Author's note, [Julian Jackson](#)*

Assistive technology continues to help me work, communicate, and retain a measurable degree of mobility and independence. I am amazed by the ability of technologies and devices to evolve and make life just that little bit easier. Assistive technology is certainly not a panacea for sight loss. It does not pretend to prevent, treat or even cure the symptoms of the eye diseases and conditions listed here. However, I strongly believe that it should be considered as a useful friend in times of crisis or specific need. I would urge all eye health/healthcare professionals and anyone experiencing temporary or permanent sight loss to explore the wonders of assistive technology and seek guidance and support from an ethical, trusted and experienced national distributor such as [Sight and Sound Technology](#).

### **Acanthamoeba Keratitis**

Acanthamoeba keratitis, or AK, is a rare but serious infection of the eye that can cause permanent vision loss or blindness. This infection is caused by a tiny ameba (single-celled living organism) called Acanthamoeba. Acanthamoeba causes Acanthamoeba keratitis when it infects the cornea, the clear dome that covers the coloured part of the eye.

Symptoms of AK include:

Sensation of something in the eye

Eye pain

Eye redness

Blurred vision

Sensitivity to light

Excessive tearing

If you experience any of these symptoms, remove your contact lenses (if you wear them) and call your eye doctor right away. AK is a rare condition, but if left untreated it can result in vision loss or blindness.

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### **Achromatopsia**

Achromatopsia is a condition characterized by a partial or total absence of colour vision. People with complete achromatopsia cannot perceive any colours; they see only black, white, and shades of grey. Incomplete achromatopsia is a milder form of the condition that allows some colour discrimination.

Possible co-morbidities: Achromatopsia can also involve other problems with vision, including an increased sensitivity to light and glare (photophobia), involuntary back-and-forth eye movements (nystagmus), and significantly reduced sharpness of vision (low visual acuity). Affected individuals can also have farsightedness (hyperopia) or, less commonly, near-sightedness (myopia). These vision problems develop in the first few months of life.

Achromatopsia is different from the more common forms of colour vision deficiency (also called [colour blindness](#)), in which people can perceive colour but have difficulty distinguishing between certain colours, such as red and green.

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## **Age-Related Macular Degeneration (AMD)**

### **Dry age-related macular degeneration**

Dry AMD is a slow deterioration of the cells of the macula, often over many years, as the retinal cells die off and are not renewed. The term 'dry' does not mean the person has dry eyes, just that the condition is not wet AMD.

The progression of dry AMD varies, but people often carry on as normal for some time.

What are the symptoms?

Macular disease affects people in different ways.

- Gaps or dark spots (like a smudge on glasses) may appear in your vision, especially first thing in the morning. Objects in front of you might change shape, size or colour, or seem to move or disappear.
- Colours can fade.
- You may find bright light glaring and uncomfortable, or find it difficult to adapt when moving from dark to light environments.
- Words might disappear when you are reading.
- Straight lines such as door frames and lampposts may appear distorted or bent.

There is currently no treatment for dry AMD. That means that you might not be referred to hospital, unless the optometrist needs to confirm their diagnosis, or thinks you need to use the hospital's low vision service.

### **Wet age-related macular degeneration**

Wet age-related macular degeneration (AMD) develops when abnormal blood vessels grow into the macula. These leak blood or fluid which leads to scarring of the macula and rapid loss of central vision. Wet AMD can develop very suddenly, but it can now be treated if caught quickly. Fast referral to a hospital specialist is essential.

What are the symptoms

Macular disease affects people in different ways:

- Gaps or dark spots (like a smudge on glasses) may appear in your vision, especially first thing in the morning. Objects in front of you might change shape, size or colour or seem to move or disappear.
- Colours can fade.
- You may find bright light glaring and uncomfortable or find it difficult to adapt when moving from dark to light environments.
- Words might disappear when you are reading.
- Straight lines such as door frames and lampposts may appear distorted or bent.

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## **Albinism**

Albinism refers to a group of conditions in which people have little or no pigment in their eyes, skin or hair. Albinism occurs due to inherited altered genes which do not make the usual amounts of pigment called ‘melanin’. Based on the amount of melanin in the eyes, different types of albinism can be distinguished, and all are associated with vision problems.

Vision problems with albinism result from abnormal development of the retina and abnormal patterns of nerve connections between the eye and the brain. Albinism affects people of all ethnic backgrounds.

Eye problems in albinism will often include:

Nystagmus: regular horizontal back and forth movement of the eyes.

Strabismus: muscle imbalance of the eyes, “crossed” or “lazy” eyes.

Photophobia: sensitivity to bright lights and glare.

Far or near sightedness.

Underdevelopment of the optic nerve.

Discolouration or whiteness of the eyes due to lack of pigmentation.

What to expect from Albinism

The treatment of albinism mainly consists of visual rehabilitation. Surgery can help reduce some of the symptoms, such as strabismus and nystagmus. Although vision cannot be restored completely, there are assistive devices available to help in a variety of daily tasks.

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## **Alstrom Syndrome**

Alstrom Syndrome is a very rare recessively inherited genetic disorder which means that both parents will carry the gene although probably be unaffected themselves.

Eye and heart problems are often the first symptoms to appear. During the first few weeks of life a number of babies collapse with congenital heart failure due to dilated cardiomyopathy.

Early treatment is often successful and babies appear to recover. Lifelong monitoring of the child’s heart is now considered essential. A number of young people develop the dilated cardiomyopathy during their teenage years.

Also during the first few weeks of life it becomes evident that the baby has an intense dislike of bright lights (photophobia) and that the babies eyes appear to wobble (nystagmus). Most children affected by Alström Syndrome will have severe sight loss and are registered blind.

Babies and young children gain weight quickly despite healthy diets and obesity is a great problem. (Early blood tests may show that the child has high insulin levels, hyperinsulinemia). A large percentage of children develop Diabetes Type Two and should be regularly checked for this.

Hearing loss can occur at any age so it is important to have your child's hearing checked regularly.

Urological problems can develop in the teenage years which cause urinary retention and/or incontinence overflow. The symptoms may be hard to spot at first but severe abdominal pain and soreness around the genitals would be early signs as well as infrequency in visiting the loo.

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### **Amblyopia (Lazy Eye)**

A lazy eye is when the vision of one of your eyes doesn't develop the way it should. Doctors also call this amblyopia.

Without treatment, your brain will learn to ignore the image that comes from the weaker eye. That could cause permanent vision problems.

Signs of a Lazy Eye

Amblyopia starts in childhood, usually between ages 6 and 9. Identifying and treating it before age 7 brings the best chances of fully correcting the condition.

Common symptoms include:

- Trouble telling how near or far away something is (depth perception)
- Squinting or shutting one eye
- Head tilting

Lazy Eye Causes

Doctors don't always know what's behind some cases of amblyopia. Causes may include:

Refractive Errors:

One eye might have much better focus than the other. The other eye could be near-sighted or far-sighted. Or it could have astigmatism (distorted or blurry vision). When your brain gets both a blurry image and a clear one, it starts to ignore the blurry one. If this goes on for months or years, vision in the blurry eye will get worse.

[Strabismus:](#)

This is when your eyes don't line up the way they should. One could turn in or out. People who have strabismus can't focus their eyes together on an image, so they often see double. Your brain will ignore the image from the eye that isn't aligned.

Cataracts. A cloudy lens inside your eye can make things look blurry. The vision in that eye might not develop the way it should.

Droopy eyelid (ptosis). A sagging eyelid can block your vision.

Lazy Eye Risk Factors

A child might be more likely to have a lazy eye if they:

Were born early (premature)



Were smaller than average at birth  
Have a family history of amblyopia or other eye conditions  
Have developmental disabilities  
Lazy Eye Diagnosis

#### Lazy Eye Complications

If treatment starts too late, the vision loss of amblyopia might be permanent because links in the body's visual system don't form the way they should.

#### Lazy Eye Outlook

With early diagnosis and treatment, most children will regain almost all their vision. Make sure your child gets eye exams early on. Follow your doctor's advice about treatment, even when it's hard to make your child do things like wear a patch every day.

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### **Aniridia**

Most cases of aniridia are inherited, which means the faulty gene which causes it is passed from parent to child. The gene which causes aniridia is the PAX6 gene. If this gene is faulty or missing, it stops the eye, and in particular the iris, developing fully when a baby is in the womb.

Some children with aniridia may only have mild blurred vision and others may have quite a lot of sight loss. Sight loss can really vary from child to child. Generally speaking, how much your child's vision is affected depends on what other parts of the eye may have been affected by aniridia and to what extent.

Other eye conditions can be linked with aniridia. These other eye conditions might be present at birth or develop later on when a child is a bit older. Your child's specialist will examine their eyes on a regular basis. How often the specialist will examine your child's eyes usually depends on how much vision your child has and what parts of their eye have been affected. These regular eye examinations can help to protect your child's useful vision and lets the specialist give any additional treatments that may be necessary.

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### **Anophthalmia and Microphthalmia**

Anophthalmia and microphthalmia are birth defects of a baby's eye(s). Anophthalmia is a birth defect where a baby is born without one or both eyes. Microphthalmia is a birth defect in which one or both eyes did not develop fully, so they are small.

### What is Anophthalmia and Microphthalmia?

Anophthalmia and microphthalmia develop during pregnancy and can occur alone, with other birth defects, or as part of a syndrome. Anophthalmia and microphthalmia often result in blindness or limited vision.

### Causes and Risk Factors

The causes of anophthalmia and microphthalmia among most infants are unknown. Some babies have anophthalmia or microphthalmia because of a change in their genes or chromosomes. Anophthalmia and microphthalmia can also be caused by taking certain medicines, like isotretinoin (Accutane®) or thalidomide, during pregnancy. These medicines can lead to a pattern of birth defects, which can include anophthalmia or microphthalmia. These defects might also be caused by a combination of genes and other factors, such as the things the mother comes in contact with in the environment or what the mother eats or drinks, or certain medicines she uses during pregnancy.

### Diagnosis

Anophthalmia and microphthalmia can either be diagnosed during pregnancy or after birth. During pregnancy, doctors can often identify anophthalmia and microphthalmia through an ultrasound or a CT scan (special x-ray test) and sometimes with certain genetic testing. After birth, a doctor can identify anophthalmia and microphthalmia by examining the baby. A doctor will also perform a thorough physical exam to look for any other birth defects that may be present.

### Treatment

There is no treatment available that will create a new eye or that will restore complete vision for those affected by anophthalmia or microphthalmia. A baby born with one of these conditions should be seen by a team of special eye doctors.

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### **Azoor**

Azoor is a rare eye disease, marked by typical characteristics of photopsias (flashes of light), visual field defects, abnormal ERG, and subtle changes to the fundus. It is often found in young myopic females, similar to other white dot syndromes of MEWDS and PIC. It was first described by Gass, who described AZOOR as minimal fundus or fluorescein angiographic changes, loss of zones of outer retinal function, irreversible ERG abnormalities, and permanent visual field loss. Late disease changes include atrophy of the photoreceptors, narrowing of arterioles, and pigment migration in a bone spicule pattern. Pathologically, the primary lesion in AZOOR is a photoreceptor outer segment degeneration. Often the pathology and decreased vision are present in a unilateral presentation; however concomitant involvement of the fellow eye is observed in some individuals.

- AZOOR is a syndrome with one or more zones of visual dysfunction, usually with photopsias, reduced outer retinal function on ERG, and with occasional photoreceptor cell loss that occurs in young women.

- The pathogenesis is unknown at this point with hypotheses supporting causation from a viral infection and from a specific autoimmune process.
- There is no clear evidence for a treatment to improve visual function, although corticosteroids have been tried with occasional success.

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(SYSTEMIC)

### **Behcet's Syndrome**

Behcet's (beh-CHETS) disease, also called Behcet's syndrome, is a rare disorder that causes blood vessel inflammation throughout your body.

The disease can lead to numerous signs and symptoms that can seem unrelated at first. They can include mouth sores, eye inflammation, skin rashes and lesions, and genital sores.

-Symptoms

Behcet's disease symptoms vary from person to person, can come and go or become less severe over time. Signs and symptoms depend on which parts of your body are affected.

Areas commonly affected by Behcet's disease include:

**Eyes.** Inflammation in the eye (uveitis) causes redness, pain and blurred vision, typically in both eyes.

**Mouth.** Painful mouth sores that look similar to canker sores are the most common sign of Behcet's disease. They begin as raised, round lesions in the mouth that quickly turn into painful ulcers. The sores usually heal in one to three weeks, though they do recur.

**Skin.** Some people develop acne-like sores on their bodies. Others develop red, raised and tender nodules on their skin, especially on the lower legs.

**Genitals.** Red, open sores can occur on the scrotum or the vulva. The sores are usually painful and can leave scars.

In people with Behcet's disease, the condition can come and go.

**Joints.** Joint swelling and pain often affect the knees in people with Behcet's disease. The ankles, elbows or wrists also might be involved. Signs and symptoms can last one to three weeks and go away on their own.

**Blood vessels.** Inflammation in veins and arteries can cause redness, pain, and swelling in the arms or legs when a blood clot results. Inflammation in the large arteries can lead to complications, such as aneurysms and narrowing or blockage of the vessel.

**Digestive system.** A variety of signs and symptoms can affect the digestive system, including abdominal pain, diarrhea and bleeding.

**Brain.** Inflammation in the brain and nervous system can cause headache, fever, disorientation, poor balance or stroke.

Causes

Behcet's disease might be an autoimmune disorder, which means the body's immune system mistakenly attacks some of its own healthy cells. It's likely that genetic and environmental factors play a role.

The signs and symptoms of Behcet's disease are considered to be due to inflammation of the blood vessels (vasculitis). The condition can involve arteries and veins of all sizes, damaging them throughout the body.

Several genes have been found to be associated with the disease. Some researchers believe a virus or bacterium can trigger Behcet's disease in people who have certain genes that make them susceptible to Behcet's.

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### **Best Disease**

Best disease, also called juvenile Best macular degeneration, juvenile Best disease and vitelliform macular degeneration, is an inherited eye condition which can affect both men and women. It usually occurs in both eyes (binocular) but it may not affect vision to the same extent in each eye. Sometimes it only affects one eye (monocular).

Best disease only affects the eyes so is not caused by, or linked to, a problem or disease in any other part of the body.

The sight loss caused by Best disease can take many years to develop and some people with Best disease can continue to read into their forties, fifties or well beyond.

Adult-onset macular vitelliform dystrophy is a slightly different eye condition to Best disease. In adult-onset macular vitelliform dystrophy there are less changes at the back of the eye, the changes begin much later in life and they do not progress in the same way. Adult-onset macular vitelliform dystrophy does not usually affect sight until around the age of 40 to 60 with very mild or moderate changes in vision. The change to vision can be so small that often it is detected by chance through a routine eye test. In general adult-onset macular vitelliform dystrophy has less impact on vision than Best disease.

The symptoms of Best disease vary from person to person, but usually the first problems people notice are with their ability to see detail. You may have problems reading small print, or you may find that there is a slight smudge in your sight or that your vision has a small blurred area in the centre. Straight lines may look distorted or wavy or as if there's a little bump in them. People may only notice these changes in one eye.

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**Birdshot Uveitis** (chorioretinopathy)

What is birdshot chorioretinopathy?

Birdshot chorioretinopathy (or retinochoroidopathy), normally shortened to ‘birdshot’, is a rare, potentially blinding, posterior uveitis. This is chronic inflammation of the choroid, which also tends to affect the retina and retinal vessels. It affects both eyes. Birdshot chorioretinopathy is characterised by inflammation of the vitreous (clear jelly in the eye) which causes orange, yellow, or cream coloured oval shaped spots at the back of your eye on your retina. These affect the macula (an area near the centre of the retina used for detailed vision) and can cause vision loss. The reason this disease is called ‘birdshot’ is because these spots look like the pattern seen when you fire birdshot pellets from a shotgun. What causes birdshot? It is believed to be due to an autoimmune disease. An autoimmune disease is an illness that occurs when the body tissues are attacked by its own immune system, which causes chronic inflammation. It is most likely to develop in people aged between 45 and 55, although it can also occur in much younger and older people. Birdshot is a relatively new disease. It was first discovered in 1949 and only given the title ‘birdshot’ in 1980. It is still widely misunderstood and often goes unrecognised and undiagnosed.

-What are the symptoms?

Usually, the initial symptoms of birdshot are floaters (black dots or wispy lines that move across your field of vision) and/or blurred vision, but there is often little noticeable effect on your ability to see. However, there are cases where symptoms have appeared very rapidly; these included painful eyes, difficulty in seeing in the dark or low light, flashing lights and sensitivity to bright light.

The progress of the disease is usually gradual, with a slow and painless loss of vision in both eyes. In the initial stages, you may be able to continue to see well but might begin to experience night vision problems and difficulty seeing different colours.

Birdshot is usually chronic (it lasts a long time) and needs treatment to prevent on-going inflammation, which can lead to permanent loss of vision. If birdshot is left untreated and you continue to have inflammation, it can lead to macular oedema (a swelling of the macula layer in the eye) which causes blindness.

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**Blepharitis**

Blepharitis is an inflammation of the eyelids in which they become red, irritated and itchy with dandruff-like scales that form on the eyelashes. It is a common eye disorder caused by either bacteria or a skin condition, such as dandruff of the scalp or rosacea.

### Anterior blepharitis

Anterior blepharitis occurs at the outside front edge of the eyelid where the eyelashes attach.

### Posterior blepharitis

Posterior blepharitis affects the inner edge of the eyelid that touches the eyeball.

### Causes & risk factors

Anterior blepharitis is commonly caused by bacteria (staphylococcal blepharitis) or dandruff of the scalp and eyebrows (seborrheic blepharitis). These bacteria are commonly found on the face and lids, but if they become excessive, or the lid area reacts poorly to their presence, an infection may occur. Less commonly, allergies or a mite infestation of the eyelashes can cause anterior blepharitis.

Posterior blepharitis can occur when the glands of the eyelids irregularly produce oil (meibomian blepharitis). This creates a favourable environment for bacterial growth. Posterior blepharitis can also develop as a result of other skin conditions, such as rosacea and scalp dandruff.

### Symptoms

People with blepharitis may experience a gritty or burning sensation in their eyes, excessive tearing, itching, red and swollen eyelids, dry eyes or crusting of the eyelids. For some people, blepharitis causes only minor irritation and itching. However, it can lead to more severe symptoms, such as blurring of vision, missing or misdirected eyelashes, and inflammation of other eye tissue, particularly the cornea. By touching and rubbing the irritated area, a secondary infection can also result.

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## **Central Serous Retinopathy (CSR)**

Central serous retinopathy is a medical condition where fluid builds up behind the retina in the eye. It can cause sudden or gradual vision loss as the central retina detaches. This central area is called the macula.

-Central serous retinopathy is a build-up of fluid underneath the retina inside the eye.

The retina is responsible for translating light taken into the eye as images the brain can understand. The build-up of liquid can cause the retina to detach, and this can cause vision problems.

In some cases, no medical intervention is required, and the person will recover their vision after a short period.

However, people should see their doctor immediately if they start to notice changes in vision.

-Symptoms

Blurry vision is a common symptom.

A person may also notice that the area around their central vision starts to darken or becomes blurry. In most cases, the vision issue is limited to one eye.

It is possible that a person may develop the condition in each eye at separate points throughout their life.

Additional symptoms of central serous retinopathy may include:

objects appear farther away

whites may appear duller

lines appear crooked

a dark spot in the centre of vision

Central serous retinopathy does not always produce symptoms. It is possible that fluid may build up in areas that are not around the macula, which is responsible for clear central vision. If this happens, a person may have the condition without knowing it because they do not have any symptoms.

-Risk factors and causes

Doctors do not know the exact causes of central serous retinopathy, but the following factors may contribute to its development:

Stress

Stress is a likely cause of central serous retinopathy. Stress causes the body to produce a hormone called cortisol.

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## **Cerebral Visual Impairment**

Cerebral visual impairment (sometimes called cortical visual impairment or CVI) is a disorder caused by damage to the parts of the brain that process vision. It's most common in babies and young children, but can continue into adulthood.

A child with CVI has vision problems that are caused by their brain that can't be explained by a problem with their eyes. Normally, the eyes send electrical signals to the brain, and the brain turns those signals into the images you see. If you have CVI, your brain has trouble processing and understanding these signals.

What are the symptoms of CVI?

CVI can cause a variety of visual problems that can range from mild to severe. Kids with CVI may have trouble:

Responding to the things they see

Seeing certain parts of what is in front of them, like busy moving scenes

Recognizing faces and objects

Recognizing things in cluttered spaces

Reaching for something while they're looking at it

Understanding what they're looking at

Parents may also notice that their child with CVI:

Reacts slowly to visual cues

Prefers to look at things that are moving

Prefers to look at things in a certain part of their vision, like with their peripheral (side) vision

Some kids with CVI tend to stare at light (like lamps or the sun), while others are sensitive to light.

Possible co-morbidities can include:

Developmental disabilities

Cerebral palsy (a brain disorder that causes movement problems)

Epilepsy (a brain disorder that causes seizures)

Hearing loss

What causes CVI?

CVI is caused by an injury to the brain. Most of the time, these injuries happen before, during, or shortly after birth. Common causes of CVI in babies and young children include:

Lack of oxygen or blood supply to the brain — often because of a stroke

Hydrocephalus (when fluid builds up in the brain)

Infections that reach the brain e.g Meningitis

Head injury

Certain genetic conditions

Babies who are born prematurely (early) are more likely to have CVI.

What's the treatment for CVI?

There's no cure for CVI, but vision rehabilitation can help people with CVI make the most of their vision. For some people with CVI, vision problems get better over time on their own.

Experts aren't sure why this happens.

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## **Charles Bonnet Syndrome**

The main cause of CBS is loss of vision and how your brain reacts to this loss. Exactly how sight loss leads to hallucinations isn't really known, but research is slowly revealing more about how the eye and the brain work together.

Current research seems to suggest that when you are seeing real things around you, the information received from your eyes actually stops the brain from creating its own pictures. When you lose your sight, however, your brain is not receiving as much information from your eyes as it used to. Your brain can sometimes fill in these gaps by releasing new fantasy pictures, patterns or old pictures that it has stored. When this occurs, you experience these images stored in your brain as hallucinations. CBS tends to begin in the weeks and months following a deterioration in your sight.

How you know if you have CBS



If you have lost some sight to a condition like macular degeneration or glaucoma and you start to hallucinate or see things that aren't really there, then you may have CBS.

There isn't one test that your doctor can do to find out whether you have CBS or not. Usually by talking with you and in some cases doing tests, your doctor will be able to rule out the other causes of hallucinations, like mental health problems, Alzheimer's and other conditions. If there are no signs of these other conditions and you have lost sight then it is probable that CBS is the cause of your hallucinations.

It's natural to be worried, confused or frightened when you see things that are not really there. Until you know what's happening, you may be concerned that seeing things is a sign of a mental health problem, or you might think that you have Alzheimer's disease. However, it's important to remember and reassuring to know that CBS is caused by sight loss only and not by any other health problem.

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## **Choroideremia**

Choroideremia is a condition characterized by progressive vision loss that mainly affects males. The first symptom of this condition is usually an impairment of night vision (night blindness), which can occur in early childhood. A progressive narrowing of the field of vision (tunnel vision) follows, as well as a decrease in the ability to see details (visual acuity). These vision problems are due to an ongoing loss of cells (atrophy) in the specialized light-sensitive tissue that lines the back of the eye (retina) and a nearby network of blood vessels (the choroid). The vision impairment in choroideremia worsens over time, but the progression varies among affected individuals. However, all individuals with this condition will develop blindness, most commonly in late adulthood.

### Frequency

The prevalence of choroideremia is estimated to be 1 in 50,000 to 100,000 people. However, it is likely that this condition is underdiagnosed because of its similarities to other eye disorders. Choroideremia is thought to account for approximately 4 percent of all blindness.

### Causes

Mutations in the CHM gene cause choroideremia. The CHM gene provides instructions for producing the Rab escort protein-1 (REP-1). As an escort protein, REP-1 attaches to molecules called Rab proteins within the cell and directs them to the membranes of various cell compartments (organelles). Rab proteins are involved in the movement of proteins and organelles within cells (intracellular trafficking). Mutations in the CHM gene lead to an absence of REP-1 protein or the production of a REP-1 protein that cannot carry out its protein escort function. This lack of functional REP-1 prevents Rab proteins from reaching and attaching (binding) to the organelle membranes. Without the aid of Rab proteins in intracellular trafficking, cells die prematurely.

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## **Coat's Disease**

Coats' disease affects the smaller blood vessels (capillaries) in your retina. Retinal capillaries are important in supplying your retina with blood which carries nutrients to the cells of your retina so that they work correctly. The cells of your retina need to remain healthy for you to be able to see clearly.

Coats' disease causes retinal capillaries to develop incorrectly. They become wider (dilated) and twisted, which make them more noticeable when the inside of your eye is examined. The medical term for these changes is telangiectasia.

As well as the retinal capillaries becoming dilated, they also become weak and leaky. This causes some of the fluid from the blood to leak out of your vessels and into your retina. This fluid, known as exudate, builds up in your retina and causes it to become waterlogged (swollen).

Where there are areas of exudates and telangiectasia your retina won't be able to work properly. This in turn will mean you won't be able to see clearly in this part of your vision.

Most children with Coats' disease don't have any symptoms – the eye doesn't look unusual, isn't painful or red, and your child won't usually be aware that there is a problem. Often children are diagnosed following an unusual finding at an optician's appointment, after failing a school vision screening test, or when the eye has an odd appearance on photographs like a white/pale pupil rather than a red pupil when a flash photograph is taken.

What causes Coats' disease?

The cause of Coats' disease isn't fully known (this is medically known as idiopathic) but it doesn't appear to be hereditary (passed through families) or caused by any other health conditions.

How does Coats' disease affect sight?

How Coats' disease affects a child's vision depends on where in the retina the leaking blood vessels grow. In the early stages of Coats' disease peripheral (side) vision is more likely to be affected. Peripheral vision describes what you see at the side when you are looking straight ahead.

Coats' disease may progress and as more of a child's retina is affected, more of their vision can become affected. The macula is the small central area of your retina. It gives you your central and detailed vision, and this is what you use when you look directly at something, for example when you read. If Coats' disease affects the macula then your child's central vision will be affected.

In the advanced stages of Coats' disease more vision will have been lost because more of the retina has been affected. When a large area of retina has become swollen it can cause the retina to detach from the back of the eye which can cause loss of sight.

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## **Coloboma**

A coloboma can affect the iris which is the coloured part at the front of the eye. It can affect the lens, the part of the eye which helps focus light onto the retina.

Coloboma can also affect the choroid which is a thin network of blood vessels which help to keep the retina healthy. Finally, it can affect the retina at the back of the eye. Very rarely coloboma can also affect the optic disc or the eyelid.

A coloboma forms whilst the baby grows in the womb. The eyes develop early during pregnancy and start off as little buds. Usually the eye folds in on itself as it develops which leaves a small gap called the foetal cleft.

The foetal cleft helps maintain the blood supply to the developing parts of the eye. In the final stage of eye development during pregnancy the cleft seals up from the back of the eye forwards and all the structures of the eye are formed. In an eye with coloboma this gap does not fully close and remains in some of the structures of the eye.

### Diagnosis

If the hospital staff suspect a child has a coloboma, which is normally first noticed by the possible keyhole-like shape of the pupil, then an ophthalmologist (eye doctor) will carry out a full eye examination.

A thorough eye examination at the hospital will help them to find out how much of the eye has been affected by the coloboma. It may be hard to tell how much a child's sight has been affected until they are older. This is because small children aren't able to communicate in words how good their vision is.

### Effects of coloboma on vision

The affect coloboma has on vision depends a lot on which part of the eye is affected and how big the gap is. Normally the gap is at the bottom of the eye (where a 6 is on the face of a clock) and runs from the front to the back of the eye. Coloboma may affect only the front of the eye if most of the cleft has sealed up. It may affect the eye from the front to the centre and back if more of the cleft has failed to close.

Most commonly coloboma only affects the iris. Children with this type of coloboma often have fairly good vision. Their pupil may be shaped a bit like a keyhole. They may have some problems with and dislike bright lights because the iris, which usually limit the amount of light entering the eye by controlling the size of the pupil may not work properly. Too much light entering the eye can cause discomfort and distort the image created.

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## **Colour Blindness**

Most colour blind people are able to see things as clearly as other people but they are unable to fully 'see' red, green or blue light. There are different types of colour blindness and there are extremely rare cases where people are unable to see any colour at all.

The most common form of colour blindness is known as red/green colour blindness and most colour blind people suffer from this. Although known as red/green colour blindness this does not mean sufferers mix up red and green, it means they mix up all colours which have some red or green as part of the whole colour. For example, a red/green colour blind person will confuse a blue and a purple because they can't 'see' the red element of the colour purple.

Similar problems can arise across the whole colour spectrum affecting all reds, greens, oranges, browns, purples, pinks and greys. Even black can be confused as dark green or dark blue.

The effects of colour vision deficiency can be mild, moderate or severe so, for example, approximately 40% of colour blind pupils currently leaving secondary school are unaware that they are colour blind, whilst 60% of sufferers experience many problems in everyday life.

Statistically speaking most people with a moderate form of red/green colour blindness will only be able to identify accurately 5 or so coloured pencils from a standard box of 24 pencil crayons.

See [Achromatopsia](#).

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## **Cone/Rod Dystrophy**

Cone-rod dystrophy (CRD) is a group of inherited eye disorders that affect the light sensitive cells of the retina called the cones and rods. People with this condition experience vision loss over time as the cones and rods deteriorate.[1][2] Initial signs and symptoms that usually occur in childhood may include decreased sharpness of vision (visual acuity) and abnormal sensitivity to light (photophobia). These signs are usually followed by blind spots in the central field of vision (scotomas), loss of colour perception, and loss of peripheral vision. Most individuals with this condition are legally blind by mid adulthood.

There are over 30 types of CRD caused by mutations in several different genes that can be inherited in many different ways including autosomal recessive, autosomal dominant, X-linked

or mitochondrial patterns.[1] CRDs are usually non-syndromic, but they may also be part of several syndromes.

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### **Congenital Blindness**

Congenital blindness refers to a group of diseases and conditions occurring in childhood or early adolescence of below 16 years old, which, if left untreated, result in blindness or severe visual impairment that are likely to be permanent blindness later in life.

•Causes

Premature Birth

Refractive error

Congenital cataract

Retinopathy of prematurity (ROP)

Infection such as Ophthalmia neonatorum that happen during time of conception or intrauterine period

Vitamin A deficiency

Measles

Genetic mutation

•The most frequently affected parts of the eyes are:

Whole globe (36%)

Cornea (36%)

Lens (11%)

Retina (6%)

Optic nerve (5%)

Uvea (2%)

#### Conjunctival disorders

-The conjunctiva is a part of the eye that covers the white of the eye and lines the inside of the eyelid. Irritation or damage to this surface can lead to conjunctival disease.

Causes

The conjunctiva can be damaged by injury, infection, chemical irritation, allergic reactions, dry eye and, rarely, by cancers. Inflammation in the conjunctiva is known as conjunctivitis.

Inflammation may be caused by infection (such as trachoma) or by an ‘auto-immune’ response in which the body attacks itself with the system that usually fights off infection.

Age can also lead to the conjunctiva becoming loose and developing folds.

#### Symptoms

Symptoms of conjunctival disease may range from redness and irritation to discharge, swollen eyelids, a burning sensation and pain.

Some conjunctival conditions can cause significant scarring. The eyelids can become stuck to the eye and cause sight loss because of damage to the cornea (the clear front surface of the eye).

Treatments

Treatment for conjunctival disorders will depend on the condition and the exact symptoms. Options may include antibiotics, steroids and other anti-inflammatories, eye drops and surgery.

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### **Congenital Cataracts**

There are many types of cataract. Some affect vision and others never do. A cataract located towards the centre of the lens is more likely to affect vision and visual system development, than one which is around the edge of the lens, though this will depend on its size and how dense the cataract is.

Very dense cataracts can cause blindness in babies if left untreated. The ophthalmologist (eye doctor) will check your child's eyes and vision and be able to tell you how much the cataract is affecting your child's vision.

Congenital cataracts can continue to develop, although this normally takes months to years. The ophthalmologist would assess how much the cataract is affecting your child's vision and then discuss treatment with you if they feel it is needed.

Causes of congenital cataracts

Around three to four per 10,000 children born in the UK have a cataract which affects vision. About a third of cataracts do not have any cause and are not linked with any other disease or condition.

A unilateral cataract (in one eye only) usually has no known cause and it is something that can just happen for no reason. In some cases they can be linked with other conditions in the eye, eye trauma, or a baby being affected by an infection whilst developing in the womb.

Bilateral cataracts (in both eyes) often run in families which a baby might inherit. Up to 23% of congenital cataracts are inherited. They can also be linked with other conditions or infections, such as rubella, when the baby was growing in the womb. Medical conditions that affect the baby's metabolism (how its body turns food into energy) can also cause congenital cataracts.

If a cataract is passed on to a baby from one or other parent it is usually dominantly inherited. One parent may know that they have cataracts themselves but sometimes they may only have a tiny cataract which does not affect their vision and which they are unaware of. This is why it can be helpful for the ophthalmologist to examine the eyes of the parents of a child with cataract even if they are unaware of a problem with their eyes.

Most children who are born with or develop infantile cataracts do not have other medical problems but some do.

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### **(Allergic) Conjunctivitis**

What is allergic conjunctivitis?

When your eyes are exposed to substances like pollen or mold spores, they may become red, itchy, and watery. These are symptoms of allergic conjunctivitis. Allergic conjunctivitis is an eye inflammation caused by an allergic reaction to substances like pollen or mold spores.

The inside of your eyelids and the covering of your eyeball have a membrane called the conjunctiva. The conjunctiva is susceptible to irritation from allergens, especially during hay fever season. Allergic conjunctivitis is quite common. It's your body's reaction to substances it considers potentially harmful.

What are the types of allergic conjunctivitis?

Allergic conjunctivitis comes in two main types:

**Acute allergic conjunctivitis**

This is a short-term condition that is more common during allergy season. Your eyelids suddenly swell, itch, and burn. You may also have a watery nose.

**Chronic allergic conjunctivitis**

A less common condition called chronic allergic conjunctivitis can occur year-round. It is a milder response to allergens like food, dust, and animal dander. Common symptoms come and go but include burning and itching of the eyes and light sensitivity.

-What are the symptoms of allergic conjunctivitis?

Red, itchy, watery, and burning eyes are common symptoms of allergic conjunctivitis. You may also wake up in the morning with puffy eyes.

How is allergic conjunctivitis diagnosed?

Your doctor will examine your eyes and review your allergy history. Redness in the white of the eye and small bumps inside your eyelids are visible signs of conjunctivitis. Your doctor may also order one of the following tests:

An allergy skin test exposes your skin to specific allergens and allows your doctor to examine your body's reaction, which may include swelling and redness.

A blood test may be recommended to see if your body is producing proteins, or antibodies, to protect itself against specific allergens like mold or dust.

A scraping of your conjunctival tissue may be taken to examine your white blood cells. Eosinophils are white blood cells that become activated when you have allergies.

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### **(Bacterial) Conjunctivitis**

Conjunctivitis is the name for inflammation of the conjunctiva; this is the thin layer of tissue on the inside of the eyelids and covers the white part of the eye. Also known as pink eye, conjunctivitis is often caused by bacteria, a virus or allergies. If pink eye is caused by bacteria, it is called bacterial conjunctivitis. Like all types of pink eye, bacterial conjunctivitis is common but not usually serious.

Symptoms of bacterial conjunctivitis

Bacterial conjunctivitis often begins in one eye and then spreads to the other. Symptoms may include

Pinkness or redness in the eye

Burning, itching, a sensation of grittiness, or mild pain or discomfort in the eye

Increased watering of the eye

Thick, sticky, often yellowish discharge from the eye; this can form a “crust” at night, making the eyes feel as if they are glued shut in the morning

Swollen eyelids

Slight sensitivity to bright light

Swelling of the lymph nodes in front of the ears

Bacterial conjunctivitis usually only causes mild symptoms and does not affect a person’s vision, other than causing slight blurriness when discharge has built up on and around the eye.

Depending on the type of bacteria causing the infection, there may be additional symptoms.

Causes of bacterial conjunctivitis

The most common causes of bacterial conjunctivitis are the following types of bacteria:

Staphylococcus aureus

Staphylococcus epidermidis

Streptococcus pneumonia

Haemophilus influenzae; this is not the same as the flu, which is caused by a virus

Less commonly, the infection can be the result of the following sexually transmitted infections (STIs):

Chlamydia

Gonorrhea

Bacterial conjunctivitis caused by chlamydia or gonorrhea is more serious and requires treatment by a doctor.

Diagnosis of bacterial conjunctivitis

When symptoms are mild, a diagnosis of bacterial conjunctivitis can often be made without seeing a doctor, and the condition can be treated at home. However, if there is any uncertainty or concern over the condition, or the symptoms are severe, seeing a doctor is very important.

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**(Viral) Conjunctivitis**

Viral conjunctivitis is a highly contagious acute conjunctival infection usually caused by adenovirus. Symptoms include irritation, photophobia, and watery discharge. Diagnosis is clinical; sometimes viral cultures or immunodiagnostic testing is indicated. Infection is self-limited, but severe cases sometimes require topical corticosteroids.

**Etiology**

Conjunctivitis may accompany the common cold and other systemic viral infections (especially measles, but also chickenpox, rubella, and mumps). Localized viral conjunctivitis without systemic manifestations usually results from adenoviruses and sometimes enteroviruses or herpes simplex virus.

Epidemic keratoconjunctivitis usually results from adenovirus serotypes Ad 5, 8, 11, 13, 19, and 37 and tends to cause severe conjunctivitis.

**Symptoms and Signs**

After an incubation period of about 5 to 12 days, conjunctival hyperemia, watery discharge, and ocular irritation usually begin in one eye and spread rapidly to the other. Follicles may be present on the palpebral conjunctiva. A preauricular lymph node is often enlarged and painful. Many patients have had contact with someone with conjunctivitis, a recent upper respiratory infection, or both.

In severe adenoviral conjunctivitis, patients may have photophobia and foreign body sensation due to corneal involvement. Chemosis may be present. Pseudomembranes of fibrin and inflammatory cells on the tarsal conjunctiva, focal corneal inflammation, or both may blur vision. Even after conjunctivitis has resolved, residual corneal subepithelial opacities (multiple, coin-shaped, 0.5 to 1.0 mm in diameter) may be visible with a slit lamp for up to 2 years. Corneal opacities occasionally result in decreased vision and significant halos and starbursts.

**Diagnosis****Clinical evaluation**

Diagnosis of conjunctivitis and differentiation between bacterial, viral, and noninfectious conjunctivitis are usually clinical; special tissue cultures are necessary for growth of the virus but are rarely indicated. Nucleic acid amplification tests (NAAT) and other rapid, office-based immunodiagnostic tests, can be useful especially when the inflammation is severe and other diagnoses (eg, orbital cellulitis) must be ruled out. Features that may help differentiate between viral and bacterial conjunctivitis can include purulence of ocular discharge, presence of preauricular lymphadenopathy, and, in epidemic keratoconjunctivitis, chemosis. Patients with photophobia are stained with fluorescein and examined with a slit lamp. Epidemic keratoconjunctivitis may cause punctate corneal staining. Secondary bacterial infection of viral conjunctivitis is very rare. However, if any signs suggest bacterial conjunctivitis (eg, purulent discharge), cultures or other studies may be useful.

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**Conjunctival Hyperaemia**

Hyperaemia refers to increased blood flow in certain bodily organs. Conjunctival hyperaemia occurs during eye inflammation (conjunctivitis). With conjunctivitis, histamines are released into the blood, which causes capillaries to expand. Consequently, the conjunctiva becomes reddened. Conjunctival hyperaemia can be associated with allergy, mechanical damage to the cornea or bacterial or viral infections. Those suffering from conjunctival hyperaemia can experience burning, itching and intraocular pressure.

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**Corneal Eye Disease**

Corneal disease is a serious condition that can cause clouding, distortion, scarring and eventually blindness. There are many types of corneal disease. The three major types are keratoconus, Fuchs' endothelial dystrophy and Bullous Keratopathy.

**Symptoms of corneal disease**

With keratoconus, as the cornea protrudes or steepens, vision becomes increasingly blurred and contact lens wear, which is often an early treatment for the disease, becomes difficult. The contact lens may not stay on the eye due to the irregular shape of the cornea.

A person with Fuchs' endothelial dystrophy or bullous keratopathy may first notice glare with lights at night or in bright sunlight. As these conditions progress, vision may be foggy or blurry in the morning and clear up as the day progresses. As the diseases further progress, vision will stay blurrier later into the day and eventually may not clear at all.

Some corneal diseases can be very painful.

Keratoconus is a weakening and thinning of the central cornea. The cornea develops a cone-shaped deformity. Progression can be rapid, gradual or intermittent. Keratoconus usually occurs in both eyes, but can occur in only one eye.

Fuchs' endothelial dystrophy is a hereditary abnormality of the inner cell layer of the cornea called the endothelium. The purpose of this layer is to pump fluids out of the cornea, keeping it thin and crystal clear. When the endothelium is not healthy, fluids are not pumped out and the cornea develops swelling, causing it to become cloudy and decrease vision.

Bullous keratopathy is a condition in which the cornea becomes permanently swollen. This occurs because the inner layer of the cornea, the endothelium, has been damaged and is no longer pumping fluids out of the tissue.

**Causes of corneal disease**

Infection: Bacterial, fungal and viral infections are common causes of corneal damage.

The cause of keratoconus in most patients is unknown.

Age: Aging processes can affect the clarity and health of the cornea.

Cataract and intraocular lens implant surgery: Bullous keratopathy occurs in a very small percentage of patients following these procedures.

Heredity

Contact lenses

Eye trauma

Certain eye diseases, such as retinitis pigmentosa, retinopathy of prematurity, and vernal keratoconjunctivitis.

Systemic diseases, such as Leber's congenital amaurosis, Ehlers-Danlos syndrome, Down syndrome and osteogenesis imperfecta.

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### **Corneal Dystrophy**

Corneal dystrophies are rare conditions in which the cornea is altered without the presence of any inflammation, infection or other eye disease. The clearness (transparency) of the cornea is affected and vision may or may not be disturbed. Corneal dystrophies tend to run in families. They have been described in many different ways but because each dystrophy will start in a particular layer of the cornea, they are classified as epithelial dystrophies, stromal dystrophies or endothelial dystrophies.

the types are as follows:

1. Recurrent corneal erosion
2. Inherited corneal dystrophies
3. Epithelial dystrophies - Meesman's dystrophy - Epithelial basement membrane dystrophy - Reis-Bücklers' dystrophy.
4. Stromal dystrophies -Granular dystrophy - Macular dystrophy - Lattice dystrophy.
5. Endothelial dystrophies - Fuchs' endothelial dystrophy - Keratoconus.

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### **Diabetic Macular Edema (DME)**

Diabetic macular edema (DME) is a complication of diabetes. People with type 1 or type 2 diabetes can develop DME.

DME occurs when excess fluid starts to build up in the macula of the eye. The macula allows us to focus and see fine details. It's located in the centre of the retina, the lining at the back of the eye that's full of blood vessels.

When excess fluid builds up in the macula, it causes vision problems.

DME generally develops over time. High blood sugar levels can damage the blood vessels in the retina. Damaged blood vessels can leak fluid, which causes swelling and other issues. This damage is called retinopathy.

There are several treatment options for DME. It's easiest to treat when diagnosed early and monitored regularly by an eye care doctor.

### Treating DME

There are effective treatments available for DME. Annual eye exams can detect any changes early. If you have DME, treatments can protect your eyesight and may reverse vision loss.

### Symptoms of DME

In its early stages, there may be no symptoms. If you have diabetes, it's important to see an eye care doctor every year so they can examine your eyes for any changes. If there's any sign of retinopathy or DME, early treatment can prevent or restore vision loss.

Make sure to tell your eye care doctor if you have any of the following symptoms:

- blurry vision
- seeing colours that look washed out
- seeing more floaters in your vision
- double vision

### Causes of DME

Over time, high blood sugar levels can damage small blood vessels in the eyes, increasing the risk of DME.

Working with your healthcare team to keep your blood sugar levels as close to target as possible is a key part of keeping your eyes healthy.

High blood pressure and high cholesterol levels can also contribute to blood vessel damage.

In some cases of diabetes, pregnancy can increase the risk of developing DME. Your doctor may recommend more frequent eye exams during pregnancy.

### Types of DME

DME is sometimes classified based on the amount of swelling seen in the retina. A thicker retina means there's more swelling, and this usually means greater vision loss.

It may also be defined by the location of damage to the blood vessels. In some cases, it's confined to one area. In other cases, the damage is more widespread throughout the retina.

When you have an eye exam, your eye care doctor may perform several tests on your eyes. The tests assess any vision loss and show any damage to blood vessels or amount of fluid build-up (swelling) in the retina.

## Prevention

It's never too late to discuss treatment options with your doctor. If you've received a diagnosis of DME, starting treatment quickly can help prevent long-term eye damage and vision loss.

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## **Diabetic Retinopathy**

People who have had diabetic retinopathy for a long time are likely to have this condition. The blood vessels in the retina are only mildly affected, when they swell they sometimes leak blood or fluid. The macula remains undamaged and vision will be normal.

Maculopathy: People who have had background diabetic retinopathy are likely to develop maculopathy. When the blood vessels in the retina begin to leak, the macula becomes affected and central vision will become gradually worse. It is very rare for someone with maculopathy to lose all of their sight as peripheral vision will be preserved. It may become difficult to recognise people's faces from a distance or see detail. The loss of central vision varies between people.

Proliferative diabetic retinopathy: People who have been dependent on insulin for a long period of time are more likely to develop proliferative diabetic retinopathy. Diabetes can cause blood vessels in the retina to become blocked. As a consequence new blood vessels will form in the eye, which is nature's way of trying to correct the problem as the retina needs a new blood supply. These new blood vessels are weak and grow on the surface of the retina and the vitreous gel. They can scar very easily and cause scar tissue to form in the eye. The scarring pulls and distorts the retina out of position.

Eyesight may become blurred or patchy as retinal bleeding obscures vision. Retinal bleeding or detachment can cause sudden and severe sight loss. If proliferative retinopathy is not treated, total loss of vision might occur.

Complications with diabetic retinopathy can be reduced by having good control of diabetes. It is important to monitor diabetes, and treat high blood pressure to prevent sight loss from diabetes.

Smoking can raise blood pressure and blood sugar levels and can increase chances of nerve damage, kidney and cardiovascular disease in people with diabetes.

Reduce the risk of diabetic retinopathy by having eyes checked regularly, not smoking, and controlling sugar levels, blood pressure and cholesterol.

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**Dry Eye Syndrome**

Dry eye disease is a common condition that occurs when your tears aren't able to provide adequate lubrication for your eyes. Tears can be inadequate and unstable for many reasons. For example, dry eyes may occur if you don't produce enough tears or if you produce poor-quality tears. This tear instability leads to inflammation and damage of the eye's surface.

Signs and symptoms, which usually affect both eyes, may include:

A stinging, burning or scratchy sensation in your eyes

Stringy mucus in or around your eyes

Sensitivity to light

Eye redness

A sensation of having something in your eyes

Difficulty wearing contact lenses

Difficulty with night-time driving

Watery eyes, which is the body's response to the irritation of dry eyes

Blurred vision or eye fatigue

When to see a doctor

See your doctor if you've had prolonged signs and symptoms of dry eyes, including red, irritated, tired or painful eyes. Your doctor can take steps to determine what's bothering your eyes or refer you to a specialist.

Causes

Tear glands and tear ducts

Tear glands and tear ducts

Dry eyes are caused by a variety of reasons that disrupt the healthy tear film. Your tear film has three layers: fatty oils, aqueous fluid and mucus. This combination normally keeps the surface of your eyes lubricated, smooth and clear. Problems with any of these layers can cause dry eyes.

Reasons for tear film dysfunction are many, including hormone changes, autoimmune disease, inflamed eyelid glands or allergic eye disease. For some people, the cause of dry eyes is decreased tear production or increased tear evaporation.

Decreased tear production

Dry eyes can occur when you're unable to produce enough water (aqueous fluid). The medical term for this condition is keratoconjunctivitis sicca.

Common causes of decreased tear production include:

Aging

Certain medical conditions including Sjogren's syndrome, allergic eye disease, rheumatoid arthritis, lupus, scleroderma, graft vs. host disease, sarcoidosis, thyroid disorders or vitamin A deficiency

Corneal nerve desensitvity caused by contact lens use, nerve damage or that caused by laser eye surgery, though symptoms of dry eyes related to this procedure are usually temporary

Increased tear evaporation

The oil film produced by small glands on the edge of your eyelids (meibomian glands) might become clogged. Blocked meibomian glands are more common in people with rosacea or other skin disorders.

Common causes of increased tear evaporation include:

Posterior blepharitis (meibomian gland dysfunction)

Blinking less often, which tends to occur with certain conditions, such as Parkinson's disease; or when you're concentrating during certain activities, such as while reading, driving or working at a computer

Eyelid problems, such as the lids turning outward (ectropion) and the lids turning inward (entropion)

Eye allergies

Preservatives in topical eyedrops

Wind, smoke or dry air

Vitamin A deficiency

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### **Duane Retraction Syndrome**

Duane syndrome, also called Duane retraction syndrome (DRS), is a congenital and non-progressive type of strabismus due to abnormal development of the 6th cranial nerve. It is characterized by difficulty rotating one or both eyes outward (abduction) or inward (adduction). There may also be changes of eyelid position on attempted movement of the eyes.

Cause of Duane Syndrome

Duane syndrome is due to miswiring of nerves to the eye muscles.

In Duane syndrome, the 6th cranial nerve that controls the lateral rectus muscle (the muscle that rotates the eye out towards the ear) does not develop properly. Why the nerve does not develop is not fully understood. Thus, the problem is not primarily with the eye muscle itself, but with the nerve that controls to the muscle. There can also be associated with miswiring of the 3rd cranial nerve, which normally controls the medial rectus muscle (the muscle that rotates the eye toward the nose). This is why abnormalities may be found in both left gaze and right gaze.

Characteristics of Duane Syndrome

Strabismus: the eyes may be misaligned and point in different directions some or all of the time

Head position: patients often maintain an abnormal head posture or head turn to keep the eyes straight

Amblyopia (reduced vision in the affected eye): occurs in 10% of patients

Eyelid narrowing: the affected eye may appear smaller than the other eye

Upshoot or downshoot: with certain eye movements, the eye may occasionally deviate upward or downward

When is Duane Syndrome treated?

For the majority of patients, Duane syndrome does not require surgical treatment. Surgery for Duane syndrome is indicated for one of four reasons:

To reduce a significant deviation in normal straight-ahead position

To eliminate a significant abnormal head position.

To eliminate a significant upshoot or downshoot.

To eliminate disfiguring abnormal eyelid position.

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### **Dystonia/Blepharospasm**

Blepharospasm usually starts gradually. First symptoms may include eye irritation, sensitivity to light and increased blinking. The frequency and severity of the muscle spasms generally increases over a period of one to two years. Sometimes eye dystonia is experienced together with mouth, jaw or tongue dystonia (oromandibular). The medical term for this is Meige's syndrome.

Symptoms usually appear between the ages of 50 and 70 but it does sometimes affect younger people. It affects around 7,000 adults in the UK.

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**Eyelid Cancer**

Eyelid cancer is a general term for a cancer that occurs on or in the eyelid. It is broadly categorized as an epithelial tumour, which is on the outer surface. An eyelid tumour can begin from sebaceous (fat), sweat, or apocrine glands, which is a type of sweat gland.

The most common types of cancer occurring on the eyelid are:

**Basal cell carcinoma.** Under the squamous cells (flat, scale-like cells) in the lower epidermis are round cells known as basal cells. About 80% of skin cancers arise from this layer in skin, and they are directly related to exposure to the sun. Basal cell carcinoma is the most common type of eyelid cancer. It usually appears in the lower lid and occurs most often in individuals with fair or pale skin.

**Sebaceous carcinoma.** Mostly occurring in middle age to older adults, sebaceous carcinoma is the second most common eyelid cancer. It may start from meibomian glands, which are glands of the eyelids that discharge a fatty secretion that lubricates the eyelids. Less frequently, it starts from glands of Zeis, the sebaceous glands at the base of the eyelashes. Sebaceous carcinoma is an aggressive cancer that normally occurs on the upper eyelid and is associated with radiation exposure, Bowen's disease, and Muir-Torre syndrome. A large sebaceous carcinoma or one that returns after treatment may require surgical removal of the eye.

**Squamous cell carcinoma.** Squamous cells make up most of the top layer of the epidermis. Approximately 10% to 30% of skin cancers begin in this layer. These skin cancers usually arise from sun exposure. They may also appear on skin that has been burned, damaged by chemicals, or exposed to x-rays. Squamous cell carcinoma is much less common than basal cell carcinoma, but it behaves more aggressively and can more easily spread to nearby tissues.

**Melanoma.** The deepest layer of the epidermis contains scattered cells called melanocytes, which produce the melanin that gives skin colour. Melanoma starts in melanocytes, and it is the most serious of the three skin cancer types.

**Symptoms and signs**

-People with eyelid cancer may experience the following symptoms or signs. Sometimes people with eyelid cancer do not show any of these symptoms. Or, these symptoms may be caused by a medical condition that is not cancer.

A change in appearance of the eyelid skin

Swelling of the eyelid

Thickening of the eyelid

Chronic infection of the eyelid

An ulceration (area where skin is broken) on the eyelid that does not heal

A spreading, coloured mass on the eyelid

**Treatment****Surgery**

Surgery is the removal of the tumour and some surrounding healthy tissue during an operation. Eye surgery is typically performed by an ophthalmologist. Different types of surgical

procedures are used depending on the size of the cancer and where it is located. Learn more about the basics of cancer surgery.

Extensive surgery may result in scarring and deformity of the eyelid, enucleation (removal of the eye), and/or may cause problems with tear drainage. Talk with your doctor before surgery about the possible side effects from your surgery, including changes to your vision and appearance, as well as physical and psychological support services available to you for your recovery.

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### **Fuch Dystrophy**

The endothelium is the innermost layer of the cornea and acts as a pump which maintains the correct amount of fluid in the cornea. In Fuchs' Dystrophy the endothelium becomes irregular and doesn't function normally. As a result, the cornea begins to take on too much fluid and starts to swell. The swelling means that the cornea becomes hazy and this is why the vision is affected. As the swelling increases, the cornea becomes slightly wrinkled, affecting the vision even more. Pain also becomes a problem because the epithelium, the outermost layer of the cornea filled with nerve endings, is also affected. At this stage it may be necessary to use a soft contact lens as a bandage to protect the epithelium.

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(SYSTEMIC)

### **Giant Cell Arteritis**

Giant cell arteritis is an inflammation of the lining of your arteries. Most often, it affects the arteries in your head, especially those in your temples. For this reason, giant cell arteritis is sometimes called temporal arteritis.

Giant cell arteritis frequently causes headaches, scalp tenderness, jaw pain and vision problems. Untreated, it can lead to blindness.

Prompt treatment with corticosteroid medications usually relieves symptoms of giant cell arteritis and might prevent loss of vision. You'll likely begin to feel better within days of starting treatment. But even with treatment, relapses are common.

You'll need to visit your doctor regularly for check-ups and treatment of any side effects from taking corticosteroids.

### Symptoms

Inflamed temporal arteries in giant cell arteritis

The most common symptoms of giant cell arteritis are head pain and tenderness — often severe — that usually affects both temples. Head pain can progressively worsen, come and go, or subside temporarily.

Generally, signs and symptoms of giant cell arteritis include:

Persistent, severe head pain, usually in your temple area

Scalp tenderness

Jaw pain when you chew or open your mouth wide

Fever

Fatigue

Unintended weight loss

Vision loss or double vision, particularly in people who also have jaw pain

Sudden, permanent loss of vision in one eye

Pain and stiffness in the neck, shoulders or hips are common symptoms of a related disorder, polymyalgia rheumatica. About 50 percent of people with giant cell arteritis also have polymyalgia rheumatica.

### Causes

With giant cell arteritis, the lining of arteries becomes inflamed, causing them to swell. This swelling narrows your blood vessels, reducing the amount of blood — and, therefore, oxygen and vital nutrients — that reaches your body's tissues.

Almost any large or medium-sized artery can be affected, but swelling most often occurs in the arteries in the temples. These are just in front of your ears and continue up into your scalp.

What causes these arteries to become inflamed isn't known, but it's thought to involve abnormal attacks on artery walls by the immune system. Certain genes and environmental factors might increase your susceptibility to the condition.

### Risk factors

Several factors can increase your risk of developing giant cell arteritis, including:

**Age.** Giant cell arteritis affects adults only, and rarely those under 50. Most people with this condition develop signs and symptoms between the ages of 70 and 80.

**Sex.** Women are about two times more likely to develop the condition than men are.

**Race and geographic region.** Giant cell arteritis is most common among white people in Northern European populations or of Scandinavian descent.

**Polymyalgia rheumatica.** Having polymyalgia rheumatica puts you at increased risk of developing giant cell arteritis.

**Family history.** Sometimes the condition runs in families.

### Complications

Giant cell arteritis can cause serious complications, including:

**Blindness.** Diminished blood flow to your eyes can cause sudden, painless vision loss in one or, rarely, both eyes. Loss of vision is usually permanent.

**Aortic aneurysm.** An aneurysm is a bulge that forms in a weakened blood vessel, usually in the large artery that runs down the centre of your chest and abdomen (aorta). An aortic aneurysm might burst, causing life-threatening internal bleeding.

Because this complication can occur even years after the diagnosis of giant cell arteritis, your doctor might monitor your aorta with annual chest X-rays or other imaging tests, such as ultrasound and CT.

**Stroke.** This is an uncommon complication of giant cell arteritis.

**Diagnosis**

Giant cell arteritis can be difficult to diagnose because its early symptoms resemble those of other common conditions. For this reason, your doctor will try to rule out other possible causes of your problem.

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## **Glaucoma**

Glaucoma is often (but not always) linked to high pressure in the eye. Healthy eyes hold their shape using fluid (aqueous humour) in the middle part of the eye. It stays at the right pressure because some drains away as more is made.

But if the eye can't drain well enough, pressure inside the eye will rise. This can squeeze and damage the cells that together form the optic nerve (retinal ganglion cells).

Older age and family history can increase the risk of glaucoma. People of recent African origin also have a higher risk.

Glaucoma can sometimes develop when eye pressure rises due to another eye condition. This is known as secondary glaucoma. Babies may be born with 'congenital' glaucoma, but this is rare.

Some people with glaucoma have normal or low eye pressure. This can be linked to low blood pressure or other conditions such as sleep apnoea. Women and people with Japanese heritage are more at risk of this type of glaucoma.

Sight loss in glaucoma usually happens very slowly, over time. It may be so slow that people don't notice until the condition is quite severe.

Primary open angle glaucoma (POAG) is the most common type of glaucoma. Sight loss starts around the edge of the field of view and slowly moves inwards. The effect is like looking through a tube and is often called 'tunnel vision'.

Acute angle closure glaucoma is much rarer. It involves a sudden blockage and a much quicker build-up of pressure. This can cause great pain. Vision may seem misty and rainbow-coloured rings around white lights might appear.

Sight loss in glaucoma is irreversible. This means that research is focused on preventing optic nerve damage with treatment or by spotting the earliest signs.

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### **Keratokonius**

Keratoconus can be difficult to detect, as it usually develops slowly. Nearsightedness and problems with how the eye is able to focus light may accompany this disease, causing additional problems with distorted and blurred vision and making a clear diagnosis more difficult.

Keratoconus often appears in the teens or early twenties. There may be a family history of Keratoconus, but this is fairly rare. Keratoconus is common in patients with atopic dermatitis, connective tissue disorder, retinitis pigmentosa and Down Syndrome.

Symptoms of Keratoconus may include:

Distorted and/or blurred vision

Glare and light sensitivity

Frequent prescription changes

Itchy eyes

Frequent rubbing of the eyes

Seeing double with one eye covered and/or multiple images

What effect does Keratoconus have?

In the mildest form of Keratoconus, glasses or soft contact lenses may help. As the disease progresses and the cornea thins and changes shape, these will no longer be sufficient to correct your vision. When the usual glasses or lenses are no longer usable, surgery to flatten the cornea or a corneal transplant may offer a solution. Discuss the best options for your specific situation with your eye care professional.

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**Leber Congenital Amaurosis**

Leber congenital amaurosis (LCA) is a group of inherited retinal diseases characterized by severe impairment vision or blindness at birth. Some retinal experts consider LCA to be a severe form of retinitis pigmentosa (RP). The condition is caused by degeneration and/or dysfunction of photoreceptors, the cells in the retina that make vision possible. Photoreceptors capture light, converting it to electrical signals which are sent to the back of the brain to create the images we see. Mutations in one of more than two dozen genes can cause LCA.

**Symptoms**

Often within an affected infant's first few months of life, parents notice a lack of visual responsiveness and roving eye movements, known as nystagmus. Eye examinations of infants with LCA sometimes reveal normal-appearing retinas. In other cases, several abnormalities are observed. Regardless, an electroretinogram (ERG), which measures retinal function, detects little if any activity in the retina. ERG tests are often essential to establishing a diagnosis of LCA. A genetic test can often provide a definitive diagnosis

Many children with LCA habitually press their eyes with their fists or fingers. This habitual pressing on the eyes is known clinically as oculodigital reflex. The eyes of individuals with LCA can also appear sunken or deep set. Keratoconus (cone shape to the front of the eye) and cataracts (clouding of the lens through which light passes) can occur with the disease.

In some cases, other body systems (e.g., kidneys) can be affected by the genetic defects that cause LCA.

**Inheritance**

LCA is almost always passed down through the autosomal recessive pattern of inheritance. In this type of inheritance, both parents, called carriers, have one mutated copy of the gene and one normal gene. They are unaffected carriers of LCA. Each of their children has a 25 percent chance of inheriting the two LCA gene copies (one from each parent) needed to cause the disorder.

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**Leber's Hereditary Optic Neuropathy**

The occurrence of LHON in most populations is still unclear. It has been estimated that there are between 1 in 30000 to 1 in 50000 affected individuals in the UK. Although this condition usually begins in a person's teens or twenties, rare cases may appear in early childhood or later in adulthood. LHON vision loss mostly affects men around age 15 - 25. Women tend to be affected at an older age, often around times of oestrogen loss. For unknown reasons, males are affected much more often than females and there is approximately a 50% chance of losing vision if you are male and a 10% chance if you are female. Male carriers are therefore at a much higher risk of being affected and the reason for this gender bias is still unclear.

**Cause**

LHON is an inherited form of vision loss. This inheritance applies to genes contained in mitochondrial DNA. Mitochondria produce most of the energy that cells need to function and these inherited mutations disrupt the mitochondria and cause cells in the retina to stop working or die. These ganglion cells are required to relay visual information from the eyes to the brain (the optic nerve). Egg cells contribute mitochondria to the developing embryo so only females pass mitochondrial conditions to their children (maternal inheritance). Mitochondrial conditions can appear in every generation of a family and can affect both males and females, but fathers do not pass mitochondrial traits to their children. LHON is the most common mitochondrial condition and about 45 mutations have been linked to LHON.

Those who have lost their central vision due to LHON are often referred to as ‘affected’ and those with a LHON mutation gene but without vision loss are known as ‘carriers’. In some rare cases, patients do experience a significant recovery of vision and this is more likely to happen if you have a certain mutation. About 40% of individuals affected with LHON do not have a clear family history of this condition. A person may carry a mitochondrial DNA mutation without experiencing any signs or symptoms of vision loss therefore it is hard to predict which members of a family who carry a mutation will eventually become affected. However, it is important to realise that most people do not go completely blind and their peripheral vision allows them to lead an independent life.

Patients with LHON mitochondrial DNA mutations usually do not have any symptoms until early adult life, when a trigger process leads to acute loss of vision.

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### **Macular hole**

A macular hole is a small break in the macula, located in the centre of the eye’s light-sensitive tissue called the retina. The macula provides the sharp, central vision we need for reading, driving, and seeing fine detail.

A macular hole can cause blurred and distorted central vision. Macular holes are related to aging and usually occur in people over age 60.

There are three stages to a macular hole:

Foveal detachments (Stage I). Without treatment, about half of Stage I macular holes will progress.

Partial-thickness holes (Stage II). Without treatment, about 70 percent of Stage II macular holes will progress.

Full-thickness holes (Stage III).

The size of the hole and its location on the retina determine how much it will affect a person’s vision. When a Stage III macular hole develops, most central and detailed vision can be lost. If left untreated, a macular hole can lead to a detached retina, a sight-threatening condition that should receive immediate medical attention.

Symptoms

Macular holes often begin gradually. In the early stage of a macular hole, people may notice a slight distortion or blurriness in their straight-ahead vision. Straight lines or objects can begin to look bent or wavy. Reading and performing other routine tasks with the affected eye become difficult.

#### Cause

Most of the eye's interior is filled with vitreous, a gel-like substance that fills about 80 percent of the eye and helps it maintain a round shape. The vitreous contains millions of fine fibres that are attached to the surface of the retina. As we age, the vitreous slowly shrinks and pulls away from the retinal surface. Natural fluids fill the area where the vitreous has contracted. This is normal. In most cases, there are no adverse effects. Some patients may experience a small increase in floaters, which are little "cobwebs" or specks that seem to float about in your field of vision.

However, if the vitreous is firmly attached to the retina when it pulls away, it can tear the retina and create a macular hole. Also, once the vitreous has pulled away from the surface of the retina, some of the fibres can remain on the retinal surface and can contract. This increases tension on the retina and can lead to a macular hole. In either case, the fluid that has replaced the shrunken vitreous can then seep through the hole onto the macula, blurring and distorting central vision.

Macular holes can also occur in other eye disorders, such as high myopia (near-sightedness), injury to the eye, retinal detachment, and, rarely, macular pucker.

What's the treatment for a macular hole?

Although some macular holes can seal themselves and require no treatment, surgery is necessary in many cases to help improve vision. In this surgical procedure – called a vitrectomy – the vitreous gel is removed to prevent it from pulling on the retina and replaced with a bubble containing a mixture of air and gas. The bubble acts as an internal, temporary bandage that holds the edge of the macular hole in place as it heals. Surgery is performed under local anesthesia and often on an out-patient basis.

Following surgery, patients must remain in a face-down position, normally for a day or two but sometimes for as long as two-to-three weeks. This position allows the bubble to press against the macula and be gradually reabsorbed by the eye, sealing the hole. As the bubble is reabsorbed, the vitreous cavity refills with natural eye fluids.

Maintaining a face-down position is crucial to the success of the surgery. Because this position can be difficult for many people, it is important to discuss this with your doctor before surgery.

What are the risks of surgery?

The most common risk following macular hole surgery is an increase in the rate of cataract development. In most patients, a cataract can progress rapidly, and often becomes severe enough to require removal. Other less common complications include infection and retinal detachment either during surgery or afterward, both of which can be immediately treated.

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**Meesmann Corneal Dystrophy**

Meesmann corneal dystrophy is an eye disease that affects the cornea, which is the clear front covering of the eye. This condition is characterized by the formation of tiny round cysts in the outermost layer of the cornea, called the corneal epithelium. This part of the cornea acts as a barrier to help prevent foreign materials, such as dust and bacteria, from entering the eye.

In people with Meesmann corneal dystrophy, cysts can appear as early as the first year of life. They usually affect both eyes and increase in number over time. The cysts usually do not cause any symptoms until late adolescence or adulthood, when they start to break open (rupture) on the surface of the cornea and cause irritation. The resulting symptoms typically include increased sensitivity to light (photophobia), twitching of the eyelids (blepharospasm), increased tear production, the sensation of having a foreign object in the eye, and an inability to tolerate wearing contact lenses. Some affected individuals also have temporary episodes of blurred vision.

**Frequency**

Meesmann corneal dystrophy is a rare disorder whose prevalence is unknown. It was first described in a large, multi-generational German family with more than 100 affected members. Since then, the condition has been reported in individuals and families worldwide.

**Causes**

Meesmann corneal dystrophy can result from mutations in either the KRT12 gene or the KRT3 gene. These genes provide instructions for making proteins called keratin 12 and keratin 3, which are found in the corneal epithelium. The two proteins interact to form the structural framework of this layer of the cornea. Mutations in either the KRT12 or KRT3 gene weaken this framework, causing the corneal epithelium to become fragile and to develop the cysts that characterize the disorder. The cysts likely contain clumps of abnormal keratin proteins and other cellular debris. When the cysts rupture, they cause eye irritation and the other symptoms of Meesmann corneal dystrophy.

**Inheritance**

This condition is inherited in an autosomal dominant pattern, which means one copy of an altered KRT12 or KRT3 gene in each cell is sufficient to cause the disorder. In most cases, an affected person inherits the condition from an affected parent.

**Other names for this condition**

corneal dystrophy, juvenile epithelial of Meesmann  
 corneal dystrophy, Meesmann epithelial  
 juvenile hereditary epithelial dystrophy  
 MECD  
 Meesman's corneal dystrophy  
 Meesmann corneal epithelial dystrophy  
 Meesmann epithelial corneal dystrophy

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## **Meibomian Gland Dysfunction**

Meibomian gland dysfunction (MGD) is a term used to describe a group of disorders, both congenital and acquired, linked by functional abnormalities of the meibomian glands. MGD can lead to altered tear film composition, ocular surface disease, ocular and eyelid discomfort, and evaporative dry eye.

Meibomian refers to a particular type of gland in the eyelids. Meibomian glands are named after Heinrich Meibom, the German doctor who first described and made drawings of them way back in 1666.

There are about 25 to 40 meibomian glands in the upper eyelid and 20 to 30 in the lower eyelid. The function of these glands is to secrete oils onto the surface of the eye. These oils help keep the tears from evaporating too quickly.

Meibomian gland dysfunction (MGD) is blockage or some other abnormality of the meibomian glands so they don't secrete enough oil into the tears. Because the tear film on the surface of the eye then evaporates too quickly, MGD is associated with dry eye syndrome. It also is connected with an eyelid problem called blepharitis.

Another name for meibomian gland dysfunction is "meibomianitis."

-MGD risk factors

There are several factors that can increase your risk of getting meibomian gland dysfunction.

Like the risk of dry eyes, the risk of MGD increases with age. People over age 40 have a significantly greater risk of developing it than children or young adults.

-Meibomian gland dysfunction (MGD) often is the underlying cause of dry eyes.

Our ethnic background also plays a role. An extensive review of published research concerning MGD showed that some studies have found that up to 69% of Asian populations in Thailand, Japan and China have meibomian gland dysfunction. By comparison, other studies have found that only up to 20% of non-Hispanic whites in the U.S. and Australia have MGD.

Wearing eye makeup is another contributing cause of MGD. Eyeliner and other makeup can clog the openings of meibomian glands. This is especially true if you don't thoroughly clean your eyelids and remove all traces of eye makeup before sleep.

Some researchers believe wearing contact lenses also may increase the risk of MGD. Recent research has shown that alterations of the meibomian glands are associated with contact lens wear, and that discontinued use of contacts for up to six months doesn't eliminate these changes.

But it's unclear whether contact lens wear actually causes meibomian gland dysfunction and most researchers say additional study is needed to determine if people who wear contacts have a greater risk of MGD.

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## **Nystagmus**

What causes nystagmus?

Early onset nystagmus will appear in very young babies. It can also be called congenital nystagmus. Acquired nystagmus is when the condition appears later in childhood.

The condition might be caused by a developmental problem of the eye or brain, or the pathway between the two. Sometimes the condition can be caused by a stroke or head injury. The majority of children with the condition do not have any other health problems. When the cause is unknown it is called idiopathic. Some forms of nystagmus can be inherited.

What are the signs and symptoms of nystagmus?

The most obvious sign that a child has nystagmus is their eye or eyes will be moving randomly. The child may not be aware of this.

Children with nystagmus often have poorer vision and problems with balance. They will also find it more difficult to follow fast movements.

How is nystagmus normally diagnosed?

The condition can be diagnosed by various eye tests. These will include a normal sight test and monitoring their eye movements.

Nystagmus is defined according to the direction of movement of the eyes, how far they move and how often. Both eyes can move together or independently of each other.

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## **Ocular Mucous Membrane Pemphigoid**

Ocular mucous membrane pemphigoid is a chronic, bilateral, progressive scarring and shrinkage of the conjunctiva with opacification of the cornea. Early symptoms are hyperemia and irritation; progression leads to eyelid and corneal damage and sometimes blindness. Diagnosis is sometimes confirmed by biopsy. Treatment often requires systemic immunosuppression.

Symptoms and signs

Usually beginning as a chronic conjunctivitis with nonspecific hyperemia without discharge in certain quadrants, ocular mucous membrane pemphigoid progresses as follows:

Subconjunctival fibrosis

Conjunctival shrinkage with loss of the inferior fornix

Symblephara (adhesions between the tarsal and bulbar conjunctiva)

Keratoconjunctivitis sicca

Trichiasis (in-turning eyelashes)

Corneal epithelial defects and bacterial corneal infection

Corneal neovascularization, opacification, keratinization, and blindness

Oral mucous membrane involvement with ulceration and scarring is common, but skin involvement, characterized by scarring bullae and erythematous plaques, is uncommon.

Diagnosis

Unexplained symblephara or biopsy findings

Diagnosis of ocular mucous membrane pemphigoid is suspected clinically in patients with conjunctival scarring plus corneal changes, symblephara, or both. The differential diagnosis of progressive conjunctival scarring includes previous radiation exposure and atopic disease.

Therefore, the clinical diagnosis of cicatricial pemphigoid is made when there is progression of a symblepharon without a history of local radiation or severe perennial allergic conjunctivitis.

Diagnosis can be confirmed by conjunctival biopsy showing linear antibody deposition on the basement membrane. A negative biopsy result does not rule out the diagnosis.

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### **Ocular Myasthenia Gravis**

How is ocular myasthenia gravis different from generalized myasthenia gravis?

Ocular myasthenia gravis only affects the muscles that move the eyes and eyelids. The symptoms of ocular myasthenia gravis include double vision (seeing two images instead of one), trouble focusing, and drooping eyelids. On the other hand, generalized myasthenia gravis affects muscles throughout the body. In addition to visual symptoms, generalized myasthenia gravis may cause trouble speaking, trouble swallowing, and weakness in the arms or legs.

Why does myasthenia gravis typically cause double vision?

The brain finely controls the eye muscles in order to keep the eyes aligned properly. Weakness of the eye muscles leads to misalignment of the eyes, which causes the eyes to perceive the same object in two different locations. Double vision is a common symptom of myasthenia gravis because this condition very frequently affects the strength of the eye muscles.

How often does myasthenia gravis only affect the eyes?

For about one half of patients with myasthenia gravis, the first symptoms are visual. About 15% of these patients will remain only having visual symptoms, even years after their diagnosis. In the other 85% of patients, however, symptoms of weakness will develop in another part of the body, usually within the next three years. At that time, these patients are considered to have generalized myasthenia gravis.

Myasthenia gravis is a condition that causes weakness of specific muscles in the body.

Normally nerves send a signal to muscles using a chemical called acetylcholine, which tells the muscles when to move. In patients with myasthenia gravis, the body's immune system mistakenly interferes with the muscles' receptors for acetylcholine. When these receptors

cannot work properly, the affected muscles tire easily. The amount of weakness typically fluctuates and may be worse at the end of the day.

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## **Optic Atrophy**

Common causes of optic atrophy:

Primary optic nerve disease

- Chronic glaucoma.
- Retrobulbar optic neuritis - eg, due to multiple sclerosis.
- Traumatic optic neuropathy.
- Lesions compressing the optic nerve (eg, tumour, aneurysms, Paget's disease of bone).

Primary retinal disease

- Central retinal artery occlusion or central retinal vein occlusion

History and Physical:

Patients who develop optic atrophy often complain of loss of vision with the segmental or diffuse blurring of the visual field. History should be directed to the suspected cause of visual impairment. Certain important points in history include the nature of the presenting illness; visual and ocular history; family, medical and surgical history; medication and social history; and hospital or institutional admission history. History of systemic infections and diseases such as diabetes and thyroid disorders, dietary disturbances, addictions to alcohol, tobacco or recreational drugs, trauma, and other factors should be elicited.

Optic neuritis is an important cause of optic atrophy. It usually occurs in individuals between 10-50 years of age. Patients typically present with sudden, the usually severe visual loss associated with pain on ocular movements. AION occurs in individuals above 50 years of age with headache and tenderness of the temporal artery. In optic atrophy due to tumours, there is an insidious history of slowly progressive visual impairment. However, hemorrhage within or due to the tumour eroding surrounding vessels would cause a sudden visual loss.

Reduced colour saturation or contrast sensitivity may develop before the occurrence of defective vision. Red colour desaturation is seen in optic neuritis. While defects in identifying blue-yellow colour may be an early sign of dominant optic atrophy, the normal linear association between stereoacuity and Snellen visual acuity could also be lost in optic atrophy.

On examining a case of optic atrophy, the observer may notice reduced visual acuity and contrast sensitivity.

Optic atrophy can be classified using different parameters. These can be clinical, pathological, and those based upon the extent and etiology.

**Clinical Types**

Primary optic atrophy occurs without any preceding swelling of the optic nerve head. The condition is caused by lesions in the anterior visual system extending from the RGCs to the

lateral geniculate body (LGB). The etiology of primary optic atrophy varies from conditions such as pituitary or optic nerve tumours and aneurysms, hereditary- and traumatic- optic neuropathies, toxic- and nutritional-optic neuropathies, following retrobulbar neuritis to multiple sclerosis. In this condition, the axons degenerate in an orderly manner. Subsequently, the resolution is characterized by the laying down of columns of glial cells.

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### **Optic Nerve Hypoplasia**

Optic nerve hypoplasia (ONH) is a congenital disorder characterized by underdevelopment (hypoplasia) of the optic nerves. The optic nerves transmit impulses from the nerve-rich membranes lining the retina of the eye to the brain. Most people with ONH have abnormal eye movements ([nystagmus](#)) and vision can range from no light perception to good functional vision, or even full vision in one eye.

Children with ONH may have brain malformations and pituitary problems.

#### Signs & Symptoms

ONH is present at birth, but many symptoms may not be apparent until childhood, or even adolescence. Most infants with ONH have involuntary, rapid eye movements (nystagmus) and/or mild to severe visual impairment of one or both eyes. Vision often improves modestly in early childhood even though there is no growth of the optic nerves after birth. Due to underdevelopment of the optic nerves, the optic disk is smaller than normal size in one or both eyes when viewed by a doctor using an ophthalmoscope. Also referred to as the “blind spot,” the optic disk is the structure in which nerve fibres from the retina combine to form the optic nerve before leaving the back of the eye. The optic nerves meet to form the optic chiasm and optic tracts at the base of the hypothalamus.

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### **Proliferative Vitreoretinopathy**

Proliferative vitreoretinopathy (PVR), a major complication of rhegmatogenous retinal detachment (RRD), is an abnormal process whereby proliferative, contractile cellular membranes form in the vitreous and on both sides of the retina, resulting in tractional retinal detachment with fixed retinal folds. However, it is increasingly being recognised that PVR may be intraretinal also, which causes retinal shortening. Research suggests that membranes form in response to cytokines and inflammatory mediators that arise following anatomic disruption and tissue damage caused by rhegmatogenous retinal detachment (RRD) and resultant

inflammation. Treatment is principally surgical and often requires multiple procedures that, in fact, yield a high rate of retinal reattachment; nevertheless, many anatomically successful eyes do not recover good visual function likely due to the long-standing macular detachment.

#### Diagnosis

Diagnosis of PVR is made via an in-depth patient history, i.e. evidence of longstanding primary RRD or of recent retinal reattachment surgery, and via physical examination, most importantly recognition of retinal detachment with fixed retinal folds.

#### Prognosis

With current surgical techniques, most eyes with PVR can now be reattached. However, despite recent advances, visual results of surgery for PVR remain poor. For example, Pastor reports that in 40-80% of anatomically successful cases, patients will maintain only ambulatory vision, defined as vision 5/200 or better<sup>40</sup>. Such a prognosis is largely a function of post-operative abnormalities, e.g. macular edema, macular pucker, subretinal membranes, optic atrophy, retinal atrophy, etc<sup>41</sup>. In addition, many eyes will retain silicone oil long after surgery with the possibility of late visual loss associated with chronic retention of silicone oil.

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### **Pterygium**

A pterygium is a growth of the conjunctiva or mucous membrane that covers the white part of your eye over the cornea. The cornea is the clear front covering of the eye. This benign or noncancerous growth is often shaped like a wedge. A pterygium usually doesn't cause problems or require treatment, but it can be removed if it interferes with your vision. The exact cause of pterygium isn't known. One explanation is that too much exposure to ultraviolet (UV) light can lead to these growths. It occurs more often in people who live in warm climates and spend a lot of time outdoors in sunny or windy environments. People whose eyes are exposed to certain elements on a regular basis have a higher risk of developing this condition. These elements include:

- pollen
- sand
- smoke
- wind

What are the symptoms?

A pterygium doesn't always cause symptoms. When it does, the symptoms are usually mild. Common symptoms include redness, blurred vision, and eye irritation. You might also feel a burning sensation or itchiness. If a pterygium grows large enough to cover your cornea, it can interfere with your vision. Thick or larger pterygium can also cause you to feel like you have a foreign object in your eye. You might not be able to continue wearing contact lenses when you have a pterygium due to discomfort.

How serious is it?

A pterygium can lead to severe scarring on your cornea, but this is rare. Scarring on the cornea needs to be treated because it can cause vision loss. For minor cases, treatment usually involves eye drops or ointment to treat inflammation. In the more serious cases, treatment can involve surgical removal of the pterygium.

How is it diagnosed?

Diagnosing a pterygium is straightforward. Your eye doctor may diagnose this condition based on a physical examination using a slit lamp. This lamp allows your doctor to see your eye with the help of magnification and bright lighting. If your doctor needs to do additional tests, they may include:

Visual acuity test. This test involves reading letters on an eye chart.

Corneal topography. This medical mapping technique is used to measure curvature changes in your cornea.

Photo documentation. This procedure involves taking pictures to track the growth rate of the pterygium.

How is it treated?

A pterygium usually doesn't require any treatment unless it's blocking your vision or causing severe discomfort. Your eye doctor might want to check your eyes occasionally to see if the growth is causing vision problems.

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## **Refractive Errors**

Refractive errors are where the eye's ability to focus light is affected, causing reduced visual capacity.

This is the most common and well-known eye condition there are various types:

Myopia - Spherical errors

Spherical errors occur when the optical power of the eye is either too large or too small to focus light correctly on the retina, (the light-sensitive tissue lining the inner surface of the eye).

Examples of spherical errors are:

- Myopia: myopia is often referred to as near-sightedness. This causes the image that one sees when looking at a distant object to be out of focus, but an image that one sees up close to be in focus.

- Hyperopia: hyperopia is often referred to as farsightedness. This causes the image that one sees when looking at a close object to be out of focus, but an image that one sees in the distance to be in focus.

Cylindrical errors

Cylindrical errors occur when the optical power of the eye is too powerful or too weak.

Examples of cylindrical errors are:



- Astigmatism: astigmatism is caused by an irregularly shaped cornea (the transparent part of the eye that covers the iris, pupil and anterior chamber). In an eye with astigmatism, light fails to come to a single focus point to produce clear vision. Instead, multiple focus points occur.
- Presbyopia: presbyopia causes the flexibility of the lens to decline, resulting in difficulty with focussing on nearby objects.

Refractive errors affect a large part of the population. To some degree almost everyone possesses refractive errors in one eye (asymptomatic) or in both eyes (symptomatic).

What are the symptoms of Refractive Errors

Symptoms of refractive errors vary per individual and may include:

- headache
- fatigue
- eye strain
- squinting
- blurred or distorted images either up close or in the distance

What to expect from Refractive Errors

How refractive errors are treated depends on the amount and the severity of the condition. Glasses and contact lenses are often used to correct the blurred vision resulting from a refractive error. In some cases, refractive surgery may be an option to correct the underlying cause. Discuss the best options for your specific case with your eye care professional.

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## **Retinal Detachment**

Retinal detachment describes an emergency situation in which a thin layer of tissue (the retina) at the back of the eye pulls away from its normal position.

Retinal detachment separates the retinal cells from the layer of blood vessels that provides oxygen and nourishment. The longer retinal detachment goes untreated, the greater your risk of permanent vision loss in the affected eye.

Warning signs of retinal detachment may include one or all of the following: the sudden appearance of floaters and flashes and reduced vision. Contacting an eye specialist (ophthalmologist) right away can help save your vision.

Symptoms

Retinal detachment itself is painless. But warning signs almost always appear before it occurs or has advanced, such as:

The sudden appearance of many floaters — tiny specks that seem to drift through your field of vision

Flashes of light in one or both eyes (photopsia)

Blurred vision

Gradually reduced side (peripheral) vision

A curtain-like shadow over your visual field

Causes

There are three different types of retinal detachment:

Rhegmatogenous - These types of retinal detachments are the most common. Rhegmatogenous detachments are caused by a hole or tear in the retina that allows fluid to pass through and collect underneath the retina, pulling the retina away from underlying tissues. The areas where the retina detaches lose their blood supply and stop working, causing you to lose vision.

The most common cause of rhegmatogenous detachment is aging. As you age, the gel-like material that fills the inside of your eye, known as the vitreous (VIT-ree-us), may change in consistency and shrink or become more liquid. Normally, the vitreous separates from the surface of the retina without any complications — a common condition called posterior vitreous detachment (PVD). One complication of this separation is a tear.

As the vitreous separates or peels off the retina, it may tug on the retina with enough force to create a retinal tear. Left untreated, the liquid vitreous can pass through the tear into the space behind the retina, causing the retina to become detached.

Tractional. This type of detachment can occur when scar tissue grows on the retina's surface, causing the retina to pull away from the back of the eye. Tractional detachment is typically seen in people who have poorly controlled diabetes or other conditions.

Exudative. In this type of detachment, fluid accumulates beneath the retina, but there are no holes or tears in the retina. Exudative detachment can be caused by age-related macular degeneration, injury to the eye, tumours or inflammatory disorders.

Risk factors

The following factors increase your risk of retinal detachment:

Aging — retinal detachment is more common in people over age 50

Previous retinal detachment in one eye

Family history of retinal detachment

Extreme near-sightedness (myopia)

Previous eye surgery, such as cataract removal

Previous severe eye injury

Previous other eye disease or disorder, including retinoschisis, uveitis or thinning of the peripheral retina (lattice degeneration)

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## **Retinal Vein Occlusion**

Retinal vein occlusion is a blockage of the small veins that carry blood away from the retina. The retina is the layer of tissue at the back of the inner eye that converts light images to nerve signals and sends them to the brain.

### Causes

Retinal vein occlusion is most often caused by hardening of the arteries (atherosclerosis) and the formation of a blood clot.

Blockage of smaller veins (branch veins or BRVO) in the retina often occurs in places where retinal arteries that have been thickened or hardened by atherosclerosis cross over and place pressure on a retinal vein.

Risk factors for retinal vein occlusion include:

Atherosclerosis

Diabetes

High blood pressure (hypertension)

Other eye conditions, such as glaucoma, macular edema, or vitreous hemorrhage

The risk of these disorders increases with age, therefore retinal vein occlusion most often affects older people.

Blockage of retinal veins may cause other eye problems, including:

Glaucoma (high pressure in the eye), caused by new, abnormal blood vessels growing in the front part of the eye

Macular edema, caused by the leakage of fluid in the retina

### Treatment

Many people will regain vision, even without treatment. However, vision rarely returns to normal. There is no way to reverse or open the blockage.

You may need treatment to prevent another blockage from forming in the same or the other eye.

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## **Retinitis Pigmentosa**

Retinitis Pigmentosa is an inherited condition affecting both eyes. If it starts in one eye, the other eye generally develops the same condition in a number of years. It is often diagnosed during the teenage years, but may be present at birth. Cases diagnosed at birth are often stable and non-progressive. Cases diagnosed later in life are often mild and may progress more slowly than those diagnosed in the teenage years.

Retinitis Pigmentosa may eventually lead to blindness, depending on the severity and the progression of the condition.

Other inherited diseases share some of the clinical symptoms of Retinitis Pigmentosa. The most common is Usher Syndrome, where both hearing and vision are affected. Other related syndromes include Best Disease, Choroideremia, gyrate-atrophy and Stargardt Disease.

What are the symptoms of Retinitis Pigmentosa

Symptoms of Retinitis Pigmentosa include:

- in the initial stage, difficulty in night vision
- decreased peripheral vision
- tunnel vision

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## **Retinoblastoma**

Retinoblastoma is an eye cancer that begins in the retina — the sensitive lining on the inside of your eye. Retinoblastoma most commonly affects young children, but can rarely occur in adults.

Your retina is made up of nerve tissue that senses light as it comes through the front of your eye. The retina sends signals through your optic nerve to your brain, where these signals are interpreted as images.

A rare form of eye cancer, retinoblastoma is the most common form of cancer affecting the eye in children. Retinoblastoma may occur in one or both eyes.

Symptoms

Because retinoblastoma mostly affects infants and small children, symptoms are rare. Signs you may notice include:

A white colour in the centre circle of the eye (pupil) when light is shone in the eye, such as when taking a flash photograph

Eyes that appear to be looking in different directions

Eye redness

Eye swelling

Causes

Autosomal dominant inheritance pattern

Autosomal dominant inheritance pattern Open pop-up dialog box

Retinoblastoma occurs when nerve cells in the retina develop genetic mutations. These mutations cause the cells to continue growing and multiplying when healthy cells would die. This accumulating mass of cells forms a tumour.

Retinoblastoma cells can invade further into the eye and nearby structures. Retinoblastoma can also spread (metastasize) to other areas of the body, including the brain and spine.

In the majority of cases, it's not clear what causes the genetic mutations that lead to retinoblastoma. However, it's possible for children to inherit a genetic mutation from their parents.

#### Retinoblastoma that is inherited

Gene mutations that increase the risk of retinoblastoma and other cancers can be passed from parents to children.

Hereditary retinoblastoma is passed from parents to children in an autosomal dominant pattern, which means only one parent needs a single copy of the mutated gene to pass the increased risk of retinoblastoma on to the children. If one parent carries a mutated gene, each child has a 50 percent chance of inheriting that gene.

Although a genetic mutation increases a child's risk of retinoblastoma, it doesn't mean that cancer is inevitable.

Children with the inherited form of retinoblastoma tend to develop the disease at an earlier age. Hereditary retinoblastoma also tends to occur in both eyes, as opposed to just one eye.

#### Complications

Children treated for retinoblastoma have a risk of cancer returning in and around the treated eye. For this reason, your child's doctor will schedule follow-up exams to check for recurrent retinoblastoma. The doctor may design a personalized follow-up exam schedule for your child. In most cases, this will likely involve eye exams every few months for the first few years after retinoblastoma treatment ends.

Additionally, children with the inherited form of retinoblastoma have an increased risk of developing other types of cancers in any part of the body in the years after treatment. For this reason, children with inherited retinoblastoma may have regular exams to screen for other cancers.

#### Prevention

In most cases, doctors aren't sure what causes retinoblastoma, so there's no proven way to prevent the disease.

#### Prevention for families with inherited retinoblastoma

In families with the inherited form of retinoblastoma, preventing retinoblastoma may not be possible. However, genetic testing enables families to know which children have an increased risk of retinoblastoma, so eye exams can begin at an early age. That way, retinoblastoma may be diagnosed very early — when the tumour is small and a chance for a cure and preservation of vision is still possible.

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### **Retinopathy of Prematurity**

Retinopathy of prematurity (ROP) is a potentially blinding eye disorder that primarily affects premature infants weighing about 2¾ pounds (1250 grams) or less that are born before 31 weeks of gestation (a full-term pregnancy has a gestation of 38–42 weeks). The smaller a baby is at birth, the more likely that baby is to develop ROP. This disorder — which usually develops in both eyes — is one of the most common causes of visual loss in childhood and can lead to lifelong vision impairment and blindness. ROP was first diagnosed in 1942.

How many infants have retinopathy of prematurity?

Today, with advances in neonatal care, smaller and more premature infants are being saved. These infants are at a much higher risk for ROP. Not all babies who are premature develop ROP. There are approximately 3.9 million infants born in the U.S. each year; of those, about 28,000 weigh 2¾ pounds or less. About 14,000–16,000 of these infants are affected by some degree of ROP. The disease improves and leaves no permanent damage in milder cases of ROP. About 90 percent of all infants with ROP are in the milder category and do not need treatment. However, infants with more severe disease can develop impaired vision or even blindness. About 1,100–1,500 infants annually develop ROP that is severe enough to require medical treatment. About 400–600 infants each year in the US become legally blind from ROP.

Are there different stages of ROP?

Yes. ROP is classified in five stages, ranging from mild (stage I) to severe (stage V):

Stage I — Mildly abnormal blood vessel growth. Many children who develop stage I improve with no treatment and eventually develop normal vision. The disease resolves on its own without further progression.

Stage II — Moderately abnormal blood vessel growth. Many children who develop stage II improve with no treatment and eventually develop normal vision. The disease resolves on its own without further progression.

Stage III — Severely abnormal blood vessel growth. The abnormal blood vessels grow toward the centre of the eye instead of following their normal growth pattern along the surface of the retina. Some infants who develop stage III improve with no treatment and eventually develop normal vision. However, when infants have a certain degree of Stage III and “plus disease” develops, treatment is considered. “Plus disease” means that the blood vessels of the retina have become enlarged and twisted, indicating a worsening of the disease. Treatment at this point has a good chance of preventing retinal detachment.

Stage IV — Partially detached retina. Traction from the scar produced by bleeding, abnormal vessels pulls the retina away from the wall of the eye.

Stage V — Completely detached retina and the end stage of the disease. If the eye is left alone at this stage, the baby can have severe visual impairment and even blindness.

Most babies who develop ROP have stages I or II. However, in a small number of babies, ROP worsens, sometimes very rapidly. Untreated ROP threatens to destroy vision.

Can ROP cause other complications?

Yes. Infants with ROP are considered to be at higher risk for developing certain eye problems later in life, such as retinal detachment, myopia (near-sightedness), strabismus (crossed eyes), amblyopia (lazy eye), and glaucoma. In many cases, these eye problems can be treated or controlled.

What causes ROP?

ROP occurs when abnormal blood vessels grow and spread throughout the retina, the tissue that lines the back of the eye. These abnormal blood vessels are fragile and can leak, scarring the

retina and pulling it out of position. This causes a retinal detachment. Retinal detachment is the main cause of visual impairment and blindness in ROP.

Several complex factors may be responsible for the development of ROP. The eye starts to develop at about 16 weeks of pregnancy, when the blood vessels of the retina begin to form at the optic nerve in the back of the eye. The blood vessels grow gradually toward the edges of the developing retina, supplying oxygen and nutrients. During the last 12 weeks of a pregnancy, the eye develops rapidly. When a baby is born full-term, the retinal blood vessel growth is mostly complete (the retina usually finishes growing a few weeks to a month after birth). But if a baby is born prematurely, before these blood vessels have reached the edges of the retina, normal vessel growth may stop. The edges of the retina — the periphery — may not get enough oxygen and nutrients.

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### **Sorsby Fundus Dystrophy**

Sorsby fundus dystrophy causes similar symptoms to age-related macular degeneration (AMD), although it generally affects people at a younger age.

How is it inherited?

Sorsby fundus dystrophy is inherited in an autosomal dominant fashion. This means that someone who inherits the faulty gene from either of their parents will develop the condition. There is a 50 per cent chance of an affected parent passing the condition on to each of their children. It affects equal numbers of men and women.

What are the symptoms?

One early symptom is night-blindness. Some research suggests that difficulty distinguishing blue from green, and yellow from red can be an early sign of Sorsby fundus dystrophy too. Later (usually when people are in their 40s) macular cells start to die off, and new blood vessels may grow into the retina, causing loss of central vision.

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### **Stargardt Macular Dystrophy**

Stargardt macular degeneration is a genetic eye disorder that causes progressive vision loss. This disorder affects the retina, the specialized light-sensitive tissue that lines the back of the eye. Specifically, Stargardt macular degeneration affects a small area near the centre of the retina called the macula. The macula is responsible for sharp central vision, which is needed for detailed tasks such as reading, driving, and recognizing faces. In most people with Stargardt

macular degeneration, a fatty yellow pigment (lipofuscin) builds up in cells underlying the macula. Over time, the abnormal accumulation of this substance can damage cells that are critical for clear central vision. In addition to central vision loss, people with Stargardt macular degeneration have problems with night vision that can make it difficult to navigate in low light. Some affected individuals also have impaired colour vision. The signs and symptoms of Stargardt macular degeneration typically appear in late childhood to early adulthood and worsen over time.

#### Frequency

Stargardt macular degeneration is the most common form of juvenile macular degeneration, the signs and symptoms of which begin in childhood. The estimated prevalence of Stargardt macular degeneration is 1 in 8,000 to 10,000 individuals.

#### Causes

In most cases, Stargardt macular degeneration is caused by mutations in the ABCA4 gene. Less often, mutations in the ELOVL4 gene cause this condition. The ABCA4 and ELOVL4 genes provide instructions for making proteins that are found in light-sensing (photoreceptor) cells in the retina.

#### Inheritance

Stargardt macular degeneration can have different inheritance patterns.

When mutations in the ABCA4 gene cause this condition, it is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

When this condition is caused by mutations in the ELOVL4 gene, it is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

#### Other Names for This Condition

juvenile macular degeneration  
macular dystrophy with flecks, type 1  
Stargardt disease  
STGD

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## **Strabismus**

To prevent double vision from congenital and early childhood strabismus, the brain ignores the visual input from the misaligned eye, which typically leads to [amblyopia](#) or "lazy eye" in that eye.

Strabismus Symptoms and Signs



The primary sign of strabismus is a visible misalignment of the eyes, with one eye turning in, out, up, down or at an oblique angle.

When the misalignment of the eyes is large and obvious, the strabismus is called "large-angle," referring to the angle of deviation between the line of sight of the straight eye and that of the misaligned eye. Less obvious eye turns are called small-angle strabismus.

Typically, constant large-angle strabismus does not cause symptoms such as eye strain and headaches because there is virtually no attempt by the brain to straighten the eyes. Because of this, large-angle strabismus usually causes severe amblyopia in the turned eye if left untreated.

Less noticeable cases of small-angle strabismus are more likely to cause disruptive visual symptoms, especially if the strabismus is intermittent or alternating. In addition to headaches and eye strain, symptoms may include an inability to read comfortably, fatigue when reading and unstable or "jittery" vision. If small-angle strabismus is constant and unilateral, it can lead to significant amblyopia in the misaligned eye.

Both large-angle and small-angle strabismus can be psychologically damaging and affect the self-esteem of children and adults with the condition, as it interferes with normal eye contact with others, often causing embarrassment and awkwardness.

Newborns often have intermittent crossed eyes due to incomplete vision development, but this frequently disappears as the infant grows and the visual system continues to mature. Most types of strabismus, however, do not disappear as a child grows.

What Causes Strabismus?

Each eye has six external muscles (called the extraocular muscles) that control eye position and movement. For normal binocular vision, the position, neurological control and functioning of these muscles for both eyes must be coordinated perfectly.

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(SYSTEMIC)

### **Stroke-Related Eye Conditions**

Strokes occur when a part of your brain is starved of oxygen.

Vision problems are common after you have a stroke. This is because our eyes send visual information to different parts of the brain involved in seeing.

If a stroke affects certain parts of the brain then this can affect your sight. Strokes can cause vision problems including visual field loss, double or blurry vision and can also affect visual processing.

Stroke can affect the visual pathways of your eye and this can affect your vision in different ways including:

visual field loss  
blurry vision  
double vision  
moving images  
other problems such as dry eye and sensitivity to light.

When stroke affects the areas of your brain that process information you see, it can cause problems such as:

visual neglect  
judging depth and movement  
recognising objects and people  
visual hallucinations.  
Field loss: hemianopia

Hemianopia is where there is a loss of one half of your visual field. This may mean that you're not able to see to either the left or right from the centre of your field of vision in both eyes. If you have a stroke to one side of your brain, you may develop field loss to the opposite side. The extent of field loss can vary and depends on the area of your brain that has been affected by the stroke.

Eye movement problems

A stroke can lead to problems with eye movements resulting in both eyes not working together as a pair. This can make it difficult to focus on specific things because of blurred vision as well as diplopia (double vision).

People may also experience problems with their fast (saccades) or slow (pursuit) eye movements which make it very difficult for the person to focus visually. In addition, their eyes may wobble (a condition known as nystagmus) or they may not be able to move both eyes together in a particular direction (gaze palsy).

Vision processing

This is when you may be able to see an object clearly but the images are not processed by your brain correctly. It can lead to people ignoring objects that are there or being unable to interpret text when reading.

Is there any treatment?

There are different techniques that can be used to try to help deal with the visual effects of stroke. These will depend on how the stroke has affected your vision but can include glasses, prisms, patching, magnifiers and scanning information. There's also computer-based rehab programs which may help improve your ability to scan if you have field loss.

Some people may see some improvement in their vision up to six months following a stroke. Again, this is highly dependent on where the damage in your brain has happened as well as the type of stroke suffered and other existing health problems. Unfortunately for many people, especially those with loss of visual field, sight loss may be permanent.

Part of the rehabilitation program for someone who has had a stroke should include an assessment of their vision and eyes. Orthoptists and low vision specialists can assess and work

with you on visual training with or without optical aids. The stroke team, GP or ophthalmologist can refer you for an orthoptic assessment and/or to the low vision clinic.

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### **Thyroid Eye Disease**

Thyroid eye disease (TED) is a condition in which the eye muscles, eyelids, tear glands and fatty tissues behind the eye become inflamed. This can cause the eyes and eyelids to become red, swollen and uncomfortable and the eyes can be pushed forward ('staring' or 'bulging' eyes).

In some cases there is swelling and stiffness of the muscles that move the eyes so that they no longer move in line with each other; this can cause double vision. Rarely TED can cause reduced vision from pressure on the nerve at the back of the eye or ulcers forming on the front of the eyes if the eyelids cannot close completely.

TED – also known as Graves' Orbitopathy or Ophthalmopathy – is an autoimmune condition. It occurs when the body's immune system attacks the tissue surrounding the eye causing inflammation in the tissues around and behind the eye. In most patients, the same autoimmune condition that causes TED also affects the thyroid gland, resulting in Graves' disease. Graves' disease most commonly causes thyroid overactivity (hyperthyroidism) but can also rarely cause thyroid underactivity (hypothyroidism). TED can occur in people when their thyroid is overactive, underactive or functioning normally. It can also occur after treatment for Graves' disease. People with TED need to be looked after by an eye specialist (ophthalmologist) and a thyroid specialist (endocrinologist).

What are the symptoms of TED?

Change in the appearance of the eyes (usually staring or bulging eyes)

A feeling of grittiness in the eyes or excessive dryness in the eyes

Watery eyes

Intolerance of bright lights

Swelling or feeling of fullness in upper or lower eyelids

New bags under the eyes

Redness of the lids and eyes

Blurred or double vision

Pain in or behind the eye, especially when looking up, down or sideways

Difficulty moving the eyes

If you have puffy eyelids and puffy skin around and under the eyes and you have a severely underactive thyroid, this is probably not TED. It should improve once you are adequately treated with levothyroxine (thyroid hormone replacement treatment).

TED can sometimes be difficult to diagnose and patients may be treated for other conditions such as conjunctivitis, allergy or hayfever months before the diagnosis is made. The signs that the diagnosis may be TED rather than any of these conditions are:

Symptoms may occur in the wrong season for hayfever  
Allergies usually cause itchy eyes, whereas TED does not  
Conjunctivitis usually causes sticky eyes, whereas TED usually does not  
TED often is associated with an ache or pain in or behind the eye, especially when trying to look up or sideways, whereas the other conditions mentioned are not  
TED is sometimes associated with double vision, whereas the other causes of eye symptoms are not

Unfortunately, some people with TED are left with permanent double vision or a change in the appearance of their eyes. Rehabilitative surgery may help once the inflammation has settled.

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## **Usher Syndrome**

What are the symptoms of Usher syndrome?

The major symptoms of Usher syndrome are hearing loss and an eye disorder called retinitis pigmentosa, or RP. Deafness is the first symptom to become apparent, usually from birth. There are broadly three different clinical types of Usher syndrome, type 1, type 2, and type 3. In Ireland, types 1 and 2 are the most common types. People with Usher type 1 develop profound deafness from birth and early childhood symptoms of RP. The deafness is generally so early in onset and so severe that hearing aids may not be of value, (although cochlear implants may be beneficial), and patients fail to develop intelligible speech. Patients with Usher type 2 also have an early onset hearing loss but the deafness is less severe than in type 1 and thus the child will benefit from hearing aids and will develop intelligible speech. In type 2 Usher syndrome the features of RP usually become obvious during their teenage years. Usher type 3 is a very rare form of Usher and is generally found in people with their family origins in Finland.

What is the cause of Usher syndrome and how is it inherited?

The prevalence of Usher syndrome varies from country to country, but it is a rare condition affecting approximately one in 10,000 people. Usher syndrome is a genetic disease that occurs when there are mutations (defects) in genes that are important for the function of both photoreceptors in the retina and hair cells in the cochlea, or inner ear. So far, researchers have found 11 genes that are associated with the three main subtypes of the syndrome. Usher syndrome is always inherited in a recessive pattern. Patients with a combination of early-onset partial deafness and retinitis pigmentosa but due to alterations in the mitochondrial DNA do not fall into the category of Usher syndrome.

If a family member is diagnosed with Usher syndrome, it is strongly advised that other members of the family also have an eye exam by an eye doctor (ophthalmologist) who is specially trained to detect retinal diseases. As the deafness becomes obvious at a much earlier

age than the RP in patients with Usher syndrome, it is particularly important that younger siblings with a hearing problem have a careful eye examination.

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### **Uveal Melanoma**

Uveal melanoma is a rare disease, but the most common primary ocular malignancy. While recent advancements are improving treatment options and supporting more accurate prognoses, the risk for metastasis and mortality remains unchanged.

#### **Symptoms**

The clinical presentation of malignant uveal melanoma is characterized by nonspecific findings associated with the location of the tumour. The more common posterior choroidal tumours have been shown to present with decreased visual acuity, floaters, photopsia, and visual field defects. Tumours involving the iris often present with the patient complaining of iris colour changes or other externally observed iris abnormalities. Both iris and choroidal melanoma have a well documented history in the literature of presenting with blurry vision and visual field defects secondary to angle tumour extension, angle neovascularization, and/or pigment-laden macrophages causing blockage of the trabecular meshwork resulting in glaucoma.

#### **Clinical diagnosis**

Diagnosis of uveal melanoma necessitates the expertise of an ophthalmologist with experience in ocular oncology.

While the above symptoms have been shown to be the first features of this malignancy, uveal melanoma is most frequently caught as an incidental finding on routine ophthalmoscopic examination. It is most commonly seen as a raised, sub-retinal lesion in the posterior pole. The differential such lesions includes benign nevus, metastatic lesions, hemangioma, hamartoma of the retina and retinal pigment epithelium, congenital hypertrophy or reactive hyperplasia of the retinal pigment epithelium, diffuse melanocytic proliferation, and detachment of the pigment epithelium, retina, or choroid.

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### **Uveitis**

Uveitis is a form of eye inflammation. It affects the middle layer of tissue in the eye wall (uvea).

Uveitis (u-vee-I-tis) warning signs often come on suddenly and get worse quickly. They include eye redness, pain and blurred vision. The condition can affect one or both eyes, and it can affect people of all ages, even children.

Possible causes of uveitis are infection, injury, or an autoimmune or inflammatory disease. Many times a cause can't be identified.

Uveitis can be serious, leading to permanent vision loss. Early diagnosis and treatment are important to prevent complications and preserve your vision.

The signs, symptoms and characteristics of uveitis may include:

Eye redness

Eye pain

Light sensitivity

Blurred vision

Dark, floating spots in your field of vision (floaters)

Decreased vision

Symptoms may occur suddenly and get worse quickly, though in some cases, they develop gradually. They may affect one or both eyes. Occasionally, there are no symptoms, and signs of uveitis are observed on a routine eye exam.

The uvea is the middle layer of tissue in the wall of the eye. It consists of the iris, the ciliary body and the choroid. When you look at your eye in the mirror, you will see the white part of the eye (sclera) and the coloured part of the eye (iris).

The iris is located inside the front of the eye. The ciliary body is a structure behind the iris. The choroid is a layer of blood vessels between the retina and the sclera. The retina lines the inside of the back of the eye, like wallpaper. The inside of the back of the eye is filled with a gel-like liquid called vitreous.

The type of uveitis you have depends on which part or parts of the eye are inflamed: Anterior uveitis affects the inside of the front of your eye (between the cornea and the iris) and the ciliary body. It is also called iritis and is the most common type of uveitis.

Intermediate uveitis affects the retina and blood vessels just behind the lens (pars plana) as well as the gel in the centre of the eye (vitreous).

Posterior uveitis affects a layer on the inside of the back of your eye, either the retina or the choroid.

Panuveitis occurs when all layers of the uvea are inflamed, from the front to the back of your eye.

When to seek medical advice

Contact your doctor if you think you have the warning signs of uveitis. He or she may refer you to an eye specialist (ophthalmologist). If you're having significant eye pain and unexpected vision problems, seek immediate medical attention.

Causes

In about half of all cases, the specific cause of uveitis isn't clear, and the disorder may be considered an autoimmune disease that only affects the eye or eyes. If a cause can be determined, it may be one of the following:

An autoimmune or inflammatory disorder that affects other parts of the body, such as sarcoidosis, ankylosing spondylitis, systemic lupus erythematosus or Crohn's disease  
An infection, such as cat-scratch disease, herpes zoster, syphilis, toxoplasmosis or tuberculosis  
Medication side effect  
Eye injury or surgery  
Very rarely, a cancer that affects the eye, such as lymphoma  
Risk factors  
People with changes in certain genes may be more likely to develop uveitis. Cigarette smoking has been associated with more difficult to control uveitis.

Complications

Left untreated, uveitis can cause complications, including:

Retinal swelling (macular edema)  
Retina scarring  
Glaucoma  
Cataracts  
Optic nerve damage  
Retinal detachment  
Permanent vision loss

See [Birdshot Uveitis](#).

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(SYSTEMIC)

### **Wolfram Syndrome**

Wolfram syndrome is an inherited condition that is typically associated with childhood-onset insulin-dependent diabetes mellitus and progressive optic atrophy. In addition, many people with Wolfram syndrome also develop diabetes insipidus and sensorineural hearing loss. Another name for the syndrome is DIDMOAD, which refers to diabetes insipidus, diabetes mellitus, optic atrophy, and deafness. Most cases of Wolfram syndrome are caused by changes (mutations) in the WFS-1 gene. Less severe mutations in the WFS-1 gene cause WFS1-related disorders, in which the affected person has only some of the features of Wolfram syndrome, such as sensorineural hearing loss without diabetes or other features.

Symptoms:

The symptoms and rate of progression of Wolfram syndrome can be quite variable. The primary symptoms of Wolfram syndrome (diabetes mellitus, optic atrophy, diabetes insipidus and hearing loss) can emerge at different ages and change at different rates. If some of these

symptoms never appear at all, the patient's condition would be called a WFS1-related disorder.

Most people affected by Wolfram syndrome develop insulin-dependent diabetes mellitus before the age of 16 (87%). The starches and sugars (carbohydrates) in the foods we eat are normally processed by the digestive system into glucose that circulates in the blood as one energy source for body functions. A hormone produced by the pancreas (insulin) allows muscle and fat cells to take up glucose. In diabetes mellitus, the pancreas does not make enough insulin so the cells cannot take up glucose normally and the blood sugar gets too high. In diabetes mellitus caused by the Wolfram gene, the patient needs daily injections of insulin to control the blood sugar. Symptoms of diabetes may include frequent urination, excessive thirst, increased appetite, weight loss, and blurred vision.

Symptoms: it is thought that almost all of those affected by Wolfram syndrome have primary optic atrophy (OA) and subsequent vision impairment of varying severity before the age of 16 (80%). The optic nerve conducts visual information to the brain for processing. Loss of the nerve fibres and/or their insulation (myelin) results in colour blindness and reduced vision typically beginning in childhood and progressing with age, though some progress quickly and others slowly.

Some people who have Wolfram syndrome also develop diabetes insipidus (42%). This is not related to diabetes or insulin. The only thing it has in common with diabetes is the symptoms of excessive thirst and urination. This condition results in excretion of large quantities of very watery-appearing urine and excessive thirst due to the brain not making enough of a hormone (vasopressin) that causes the kidneys to hold onto water. Patients tend to drink enormous quantities of fluid and urinate very often. Other symptoms may be dehydration, weakness, dryness of the mouth, and sometimes constipation, which may develop rapidly if the loss of fluid is not continuously replaced. Diabetes insipidus can be treated with vasopressin hormone replacement called dDAVP.

Hearing loss is the fourth major symptom of Wolfram syndrome and occurs in approximately 48% of patients. This symptom may occur at any age and may be partial or complete. The hearing loss is due to a loss of sound perception transmitted by nerves (sensorineural). Symptoms may include loss of sound intensity or pitch, or loss of the ability to hear high tones.

#### Related Disorders

The following disorders have symptoms like some of the symptoms of Wolfram syndrome:

Leber Hereditary Optic Atrophy (LHOA) is a rare inherited disorder of the eye that is characterized by the relatively slow, painless, progressive loss of vision. The optic atrophy in LHOA and the optic atrophy in Wolfram syndrome may look the same and have the same symptoms. LHOA can start in one eye or both, but both eyes are usually affected within six months. In most people, vision loss is permanent. LHOA is a genetic disorder that occurs as the result of a mutation in the mitochondrial DNA that is inherited from the mother or arises as a new sporadic mitochondrial DNA mutation. (For

Thiamine-responsive megaloblastic anemia syndrome (TRMA) is an autosomal recessive disorder with features that include megaloblastic anemia, sensorineural hearing loss and diabetes mellitus. Megaloblastic anemia is a blood disorder characterized by anemia, with red



blood cells that are larger than normal, usually resulting from a deficiency of folic acid or of vitamin B-12. The hearing loss and diabetes mellitus in this disorder can look the same as the hearing loss and diabetes mellitus in Wolfram.

#### Diagnosis

Wolfram syndrome is difficult to diagnose. In many instances, people with this disorder and their doctors may be unaware that the various symptoms and complaints are related and indicate a specific disorder. Initially, the focus may be on one symptom, typically diabetes mellitus, and its treatment. Later, the presence of other symptoms may become apparent.

Wolfram syndrome should be considered in anyone with diabetes mellitus and optic atrophy; anyone with low frequency sensorineural hearing loss; anyone with either diabetes mellitus or optic atrophy in addition to hearing loss or diabetes insipidus or bladder dysfunction or loss of sense of smell or a family member with Wolfram syndrome.

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