# Coloboma

## What is coloboma?

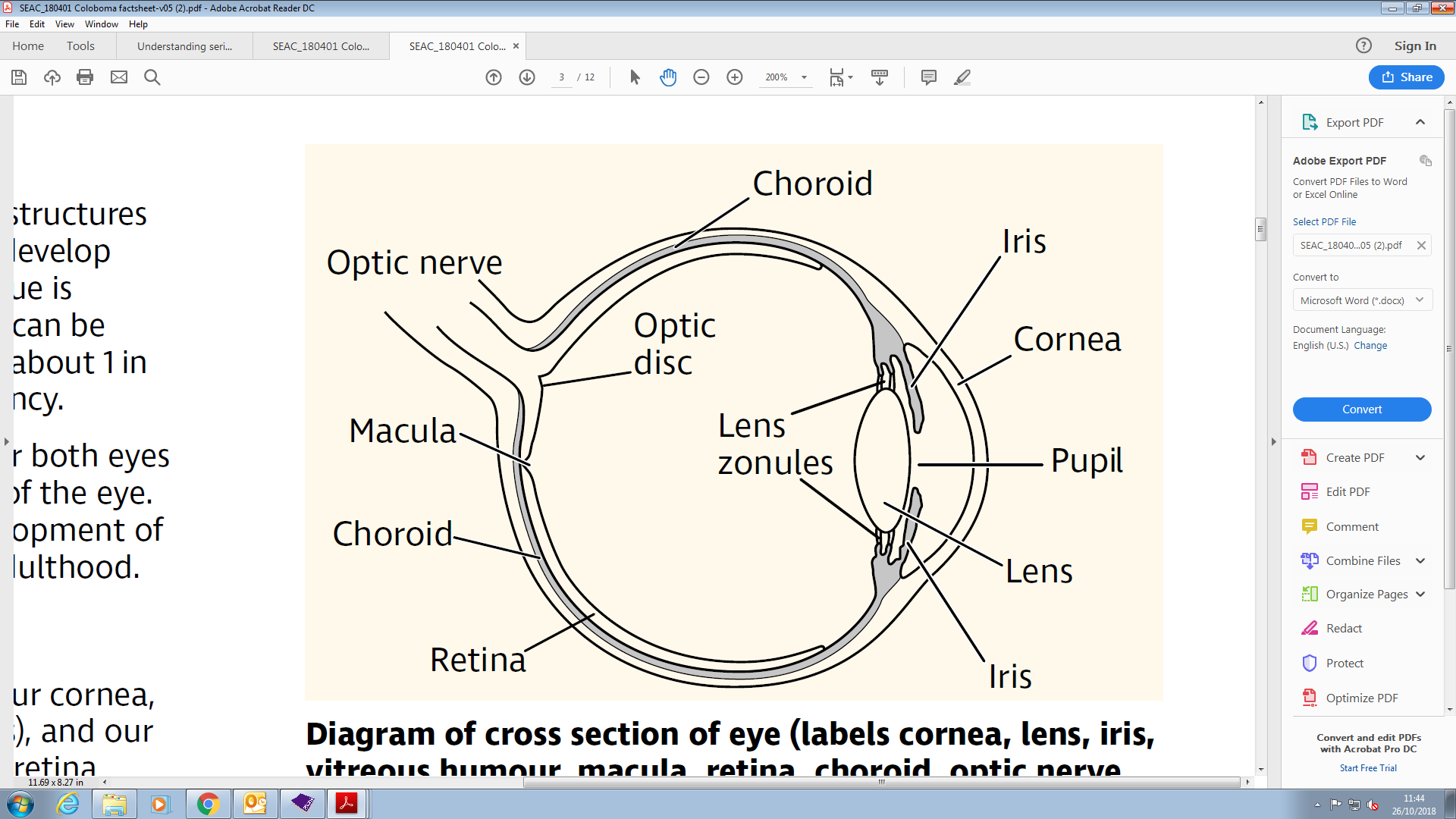
Coloboma means that part of one or more structures inside an unborn baby’s eye does not fully develop during pregnancy. This underdeveloped tissue is normally in the lower nasal (down and towards the nose) part of the eye and it can be small or large. A coloboma occurs in about one in 5,000 births and develops at around five to seven weeks of pregnancy.

Coloboma can affect one eye (unilateral) or both eyes (bilateral) and it can affect different parts of the eye. As coloboma forms during the initial development of the eye, it is present from birth and into adulthood.

## How do we see with our eyes?

Light enters our eyes by passing through our cornea, our pupil (the hole in the middle of the iris), and our lens so that it is sharply focused onto the retina lining the back of our eye. The retina is made up of light-sensitive cells that convert light into electrical impulses which then travel along the optic nerve to our brain. Our brain processes these signals so that we can “see” the world around us.

The following diagram shows the cross-section of the eye, and includes the labels cornea, pupil, lens, iris, lens zonules, macula, retina, choroid, optic nerve and optic disc.

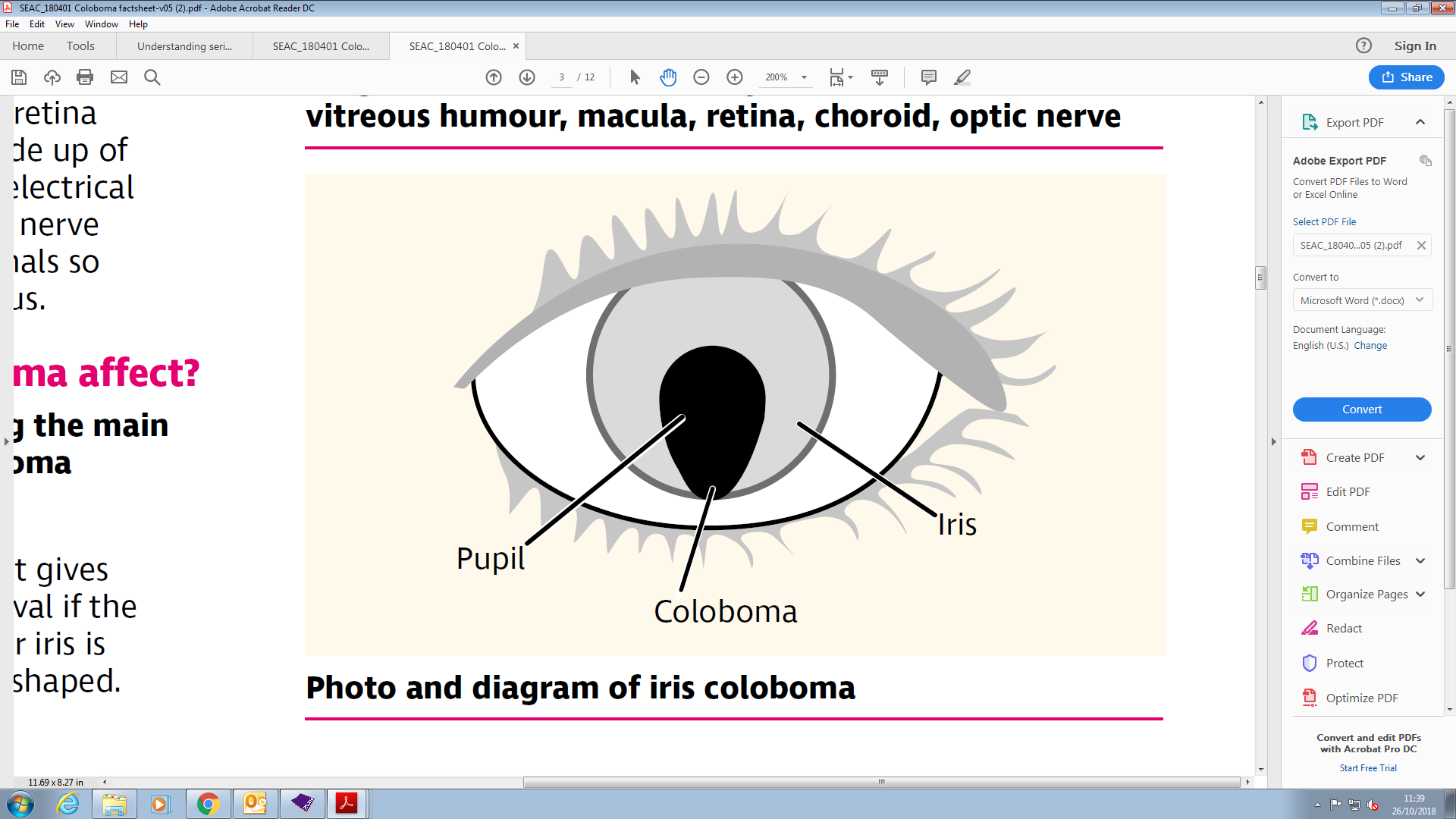


## Which parts of the eye can coloboma affect?

### Iris

Coloboma can affect the iris, the tissue that gives you your eye colour. Your pupil may look oval if the coloboma is partial but if more of the lower iris is missing, your pupil will look more keyhole shaped.

Below are pictures to illustrate an iris coloboma. The first picture is a diagram of a left eye with iris coloboma, including the labels pupil, coloboma and iris.



The second picture below is a photograph of a right eye with iris coloboma.



Although iris coloboma is the most obvious sign of coloboma in many cases, it’s possible that the condition can affect other parts of the eye without an iris coloboma being present.

### Lens zonules

Coloboma can also affect the supporting fibres of the lens, which are called the lens zonules. The zonules hold your lens in place, just behind your iris. If a section of zonules is defective or missing, the lens is not held as strongly in position all the way round. This alters its shape and as a result, there will be a notch or dent in the lens. This is often referred to as lens coloboma, even though none of the lens tissue itself is missing to form the notch.

### Retina and choroid (chorioretinal)

At the back of your eye, the retina and the choroid can be affected by coloboma. This is known as a chorioretinal coloboma. The choroid is the layer of tissue behind your retina, and it contains blood vessels that keep your retina healthy.

### Optic disc

Coloboma can also affect the optic nerve where it leaves the back of your eye, an area known as the optic disc.

### Eyelids

Eyelid coloboma is less common. Although gaps in the eyelids are still known as coloboma, they are not caused by the same development problem in pregnancy that leads to coloboma of structures inside the eye.

## What causes coloboma to form inside the eye?

Coloboma forms during pregnancy while a baby is developing. During the second month of pregnancy, the buds that become the eyes develop and fold round in a cup-like shape from which the structures of the eye will form. As the developing eye folds round, it should join together, from the back to the front of the bottom of the cup, along a seam-like structure called the embryonic fissure. If this seam doesn’t join completely, there will be tissue missing at the bottom of the structures being formed, leading to coloboma. The structure in the eye that is affected by coloboma relates to the part of the embryonic fissure that didn’t close properly. Coloboma may only affect the front of a baby’s eye if most of the fissure closed up.

However, coloboma may also affect structures further back along the length of the eye too, if less of the fissure closed properly. More than half (50%) of people with coloboma have more than one structure in the eye that is affected.

Most cases of coloboma appear without any previous family history, although in some families coloboma can be inherited, or could be caused by environmental factors such as certain drugs or infections during pregnancy. A fault in the gene PAX6 has also been found to cause coloboma. If a child is found to have a coloboma, the ophthalmologist (hospital eye doctor) may ask to examine both parents’ eyes too. This is because either one of them may have a coloboma as well but be unaware of it because they have no visual problems. If there is coloboma in your family, you may wish to talk with the hospital staff or your GP about genetic testing and speak to a genetic counsellor about the genetic inheritance of the condition.

Some families will be told about genetic changes that may be involved in their child’s coloboma, while others will not. This is because not all the genes relating to coloboma have been identified yet and not all coloboma is caused by a genetic change. Current research aims to find out more about the genes that relate to coloboma. There is more information about this research on the MACS (Microphthalmia, Anophthalmia and Coloboma Support) and Gene Vision websites listed in the further help and support section at the end of this information.

If coloboma is found in both eyes, an ophthalmologist is more likely to want to identify any underlying cause, whether genetic or not. Therefore, even if coloboma is only thought to be in one eye initially, they will still want to examine both eyes in detail.

## Does coloboma affect vision?

The effect that coloboma has on vision depends on which structures of the eye are involved and how much tissue is missing.

### Iris coloboma

Both children and adults with only iris coloboma will probably have good vision. However, iris coloboma can cause light sensitivity, (photophobia), in bright conditions. This is because the amount of light entering the eye is controlled by the muscles in the iris. If too much light enters the eye because some of the iris is missing and not working effectively, the glare that results can cause discomfort, and affect the quality of vision. Tinted glasses, sun hats or sun blinds in cars can help to relieve light sensitivity. Your optometrist (optician) will be able to offer further advice about this.

You can find out more about light sensitivity from our website **rnib.org.uk/eyehealth** or by calling our Helpline on **0303 123 9999**.

### Lens coloboma

If the lens of the eye is affected by coloboma, vision is likely to be blurred because of the change in lens shape caused by a notch or dent. There may also be some degree of cataract in the affected eye, which means the lens is cloudy instead of being clear. More information about the treatment for cataract can be found in the “Can coloboma lead to other eye health problems?” section later in this factsheet.

### Chorioretinal (retina and choroid) and optic disc coloboma

Some people with chorioretinal coloboma may not be aware of any visual symptoms. However, the more retinal tissue that is missing, the more their sight is likely to be affected, usually in the upper part of their field of vision.

If the chorioretinal coloboma is large or there is optic disc coloboma, central vision is likely to be poor. This will make activities such as reading, writing and watching television more difficult. If both eyes are affected by optic disc coloboma, nystagmus may also be present, which means that the eyes move constantly and uncontrollably. Vision that is affected by these types of coloboma cannot be corrected with glasses or contact lenses.

You can find out more about nystagmus from our website **rnib.org.uk/eyehealth** or by calling our Helpline on **0303 123 9999**.

It may be hard to tell what effect coloboma will have on a child’s sight until they are older when they will be able to communicate more accurately to the ophthalmologist or optometrist about how well they can see. Some children with coloboma may also have focusing problems that aren’t caused by the coloboma itself, such as being short-sighted, long-sighted or having astigmatism, so their optometrist may advise that they need to wear glasses. Although glasses and contact lenses are unable to improve any sight problems that are caused by coloboma, it’s important for children to have any focusing problems corrected to allow their sight to develop as fully as possible. Therefore, it’s important that children continue to have regular eye examinations as often as is advised by their optometrist.

## How is coloboma diagnosed?

Coloboma is normally diagnosed when a baby is born or in a very young child. Iris coloboma can usually be seen as a keyhole-shaped pupil when looking at their eyes. Some people with iris coloboma also have differently coloured eyes which is called iris heterochromia. This means that there is a different amount of pigment on the iris of each eye.

If you notice these features in your child, your GP or optometrist would normally refer them to an ophthalmologist. An ophthalmologist will examine their eyes in detail to check if there is coloboma in any other part of their eye. To do this, they normally use an instrument called an ophthalmoscope which they hold close to your child’s eye but without touching it. They may also carry out other tests as well, which may include:

* measuring what your child can see to the sides, above and below when looking straight ahead. This is a measure of their visual field, and it does require your child’s co-operation to give accurate results
* looking at the different structures of their eyes using a special microscope called a slit lamp, or similar device
* optical coherence tomography (OCT) which provides the ophthalmologist with an image of the different layers of the retina but without touching the eye
* CT (computerised tomography) or MRI (magnetic resonance imaging) scans so the ophthalmologist can examine the structures of the eye.

If a young baby is suspected of having coloboma, it may be necessary to give them a general anaesthetic which means they would be fully asleep so that the ophthalmologist can examine their eyes thoroughly without causing them distress.

If someone has a chorioretinal coloboma, it might appear that they have a white pupil instead of the red reflex normally seen in pictures taken using flash photography. The usual red reflex is caused when the camera flash lights up the retina with its rich blood supply at the back of the eye. If there is a gap in the retina due to coloboma, the red reflex will be replaced with a white one. A white pupil is known as leucocoria. If you notice that your child has the appearance of leucocoria at any time, it’s important that their eyes are examined by an optometrist or ophthalmologist urgently because there are other serious eye conditions that can give a similar appearance.

## What is the treatment for coloboma?

If a child has coloboma, they will receive specialist care at hospital during the early years to monitor the effect of the coloboma and to maximise their vision.The frequency of these checks will depend on their needs.

Many children and adults with iris coloboma will require no treatment. However, some have a special contact lens fitted that covers the keyhole-shaped pupil and makes it look round. This improves the cosmetic appearance of the eye as well as reducing light sensitivity. For some people, surgery is an option, where the gap in the iris is sewn together to correct the pupil shape and reduce light sensitivity. Your ophthalmologist will be able to discuss any appropriate treatment options with you.

For other types of coloboma inside the eye, there is no treatment at present. However, other eye health problems that are associated with coloboma, such as glaucoma, retinal detachment, choroidal neovascularisation and cataract can be treated. These conditions are described in the next section of this information. It’s important for people with coloboma to have regular eye examinations with their optometrist, who will be able to check for any changes to their eye health. An optometrist will advise how often an eye examination is necessary. However, anyone with coloboma who experiences any new symptoms or concerns should have their eyes examined by an eye health professional straight away.

## Can coloboma lead to other eye health problems?

Sometimes, coloboma can increase the risk of other eye conditions. An ophthalmologist or optometrist will be able to monitor the eye health of anyone with coloboma and offer further advice about these conditions. Some are listed here.

### Glaucoma

Glaucoma is an eye condition where your optic nerve is damaged by the pressure of the fluid inside your eye. There may be an increased risk of glaucoma in people with coloboma because there may be a malformation of the drainage meshwork inside the eye that helps to maintain the eye’s pressure. Treatment for glaucoma can be given to lower eye pressure to prevent further damage to the optic nerve and preserve sight.

You can find out more about glaucoma from our website **rnib.org.uk/eyehealth** or by calling our Helpline on **0303 123 9999**.

### Retinal detachment

There is an increased risk of a retinal detachment if there is coloboma at the back of the eye involving the retina, choroid or optic disc. This means that the retina near to the coloboma is more at risk of becoming separated (detached) from the back of the eye. A retinal detachment can be treated with surgery, but this does need to be done urgently to prevent further sight loss in that eye. The symptoms of retinal detachment include floaters and flashes of light in your vision and some people notice a curtain effect or shadow coming down, up or across their vision. It’s important if any of these symptoms develop that your eyes are checked within 24 hours by an optometrist or at your local A&E or eye casualty, so that an accurate diagnosis can be made.

You can find out more about retinal detachment from our website **rnib.org.uk/eyehealth** or by calling our Helpline on **0303 123 9999**.

### Choroidal neovascularisation (new blood vessel growth)

Some people with chorioretinal and optic disc coloboma can develop new blood vessels in the choroidal layer behind the retina at the back of their eye. These new vessels are weak, so they leak and bleed easily, which can damage your vision. Choroidal neovascularisation is a rare complication of coloboma at the back of the eye, but it can be treated with anti-VEGF injections.

Anti-VEGF treatments work by reducing the growth of these new, leaky blood vessels and the bleeding they cause, which in turn can reduce the risk of damage to the retina. This can help to avoid further deterioration in sight.

Anti-VEGF medicines are injected into the vitreous gel. This is called an intravitreal injection. The injection is given in a clean sterile room or an operating theatre to reduce the risk of infection. Before the injection, you’ll be given anaesthetic eye drops to make your eye numb, so the injection is not painful.

You can find more information about anti-VEGF treatment on our website **rnib.org.uk/eyehealth** or by calling our Helpline **0303 123 9999**.

### Cataract

Some people with coloboma will develop a cataract at an earlier age than people without coloboma. Having a cataract means the lens inside your eye has become cloudy. This cloudiness can cause symptoms such as blurred or misty vision, colours appearing dull, and glare in your vision, particularly with night driving and oncoming headlights. If vision cannot be improved properly with glasses, adults with cataracts can be treated using surgery to remove the cloudy lens, replacing it with a clear artificial lens implant. This procedure can be more complex for a child with cataracts and coloboma, so the ophthalmologist will carry out a detailed assessment before discussing ways to manage the condition with the child’s family.

You can find more information about cataracts and childhood (congenital) cataracts on our website **rnib.org.uk/eyehealth** or by calling our Helpline **0303 123 9999**.

## What other health problems can affect some children with coloboma?

Most children with coloboma do not have any other eye or general health problems.

However, some children with coloboma also have other eye health problems such as microphthalmia (where the eye is much smaller than it should be) and anophthalmia (where the eye has not developed at all and is absent). Some children with coloboma have other general health problems that need to be assessed and so, for this reason, the ophthalmologist may carry out additional tests to check for these. This is more common when there is coloboma in both eyes.

Coloboma can be part of a syndrome, where other parts of the body are also affected. CHARGE syndrome is a rare condition and is one example of a syndrome involving coloboma. The letters represent different features of the condition:

**C**oloboma

**H**eart defects

**A**tresia, or blockage, of the nasal passages (choanal atresia)

**R**estricted growth and development

**G**enital abnormalities

**E**ar abnormalities

The ophthalmologist caring for your child may refer them to a paediatrician (a doctor specialising in children and their conditions) to carry out the relevant tests that will detect whether they have any other general health problems associated with their coloboma, such as CHARGE syndrome.

**Sense** and **Sense Scotland** are other useful organisations that can provide information and support for those who have both sight loss and hearing loss or who have complex disabilities, including CHARGE syndrome. For more information in England, Wales and Northern Ireland, visit **sense.org.uk** or call **0300 330 9256** (Monday to Friday, 9am to 5pm)**.** For more information in Scotland, visit **sensescotland.org.uk** or call **0300 330 9292** (Monday to Friday, 9am to 5pm)**.**

## Coping

It’s completely natural to be concerned if you or your child has coloboma and normal to find yourself worrying about what it means now and in the future. You or your child may have normal vision or there may be some degree of sight loss. The extent of sight loss will relate to the type of coloboma is present, how large it is and whether it is in one or both eyes. For a child, it’s difficult to predict to what extent their vision will be affected by coloboma in the long term. Therefore, it’s important for them to attend their hospital eye clinic appointments as advised by their ophthalmologist, and to have regular eye examinations as advised by their optometrist, to ensure they have the greatest chance of developing the best vision they can. It’s also important for adults to have regular eye examinations too, with either an ophthalmologist (if still under the care of the hospital) or an optometrist at least every two years, or as recommended, to ensure that their eye health is monitored.

### Support from RNIB

It can sometimes be helpful to talk about how you’re feeling with someone outside your circle of friends or family. By calling our RNIB Helpline, you are no longer alone. We can support you at every step, putting you in touch with the advisors you need from any of our supportive teams. From support with education to advice on employment, from using assistive technology to understanding more about your eye condition, we are here to help. Our Counselling and Well-being team is also available to provide the emotional support you may need. Your GP or social worker may also find a counsellor for you if you feel this might help.

### The Eye Care Liaison Officer (ECLO)

You may think of further questions about coloboma on your way home from a hospital appointment or in the days and weeks that follow a diagnosis. There is someone to turn to with these questions. Your eye clinic may have a sight loss advisor working alongside the doctors and nursing staff. This advisor may be known as either the Eye Care Liaison Officer (ECLO), the Vision Support Officer or the Early Intervention Support Officer, and they are on hand within your hospital to provide you with further practical and emotional support about your eye health. To find out if your hospital eye clinic has an ECLO, you can search within the RNIB Sightline Directory: **sightlinedirectory.org.uk**

Alternatively, you can call our Helpline on **0303 123 9999** to speak to our advisors within our **Eye Health Information Service**, as they would be happy to discuss any questions you may have.

## Further help and support

### Making the most of your sight

When vision is reduced, some tasks, such as reading written text, may be made easier by making things bigger and bolder. Using contrasting colours can also help. When a person has coloboma, it can be more comfortable if the lighting around them and any devices being used are not causing any glare.

We have a series of leaflets with helpful information on living with sight loss, including how to make the most of your sight. You can find out more about our range of titles by calling our Helpline.

For children who have sight loss as a result of their coloboma, having the right support at an early age can make a big difference. It’s important for the local authority to be aware of any specialist educational needs a child has so that a plan can be put in place to provide for this. Local authorities should also be able to provide a specially trained qualified teacher of children and young people with vision impairment (QTVI) to work alongside children and their families. They can provide information on development, play, education and many other matters.

You can find out more about education professionals from our website **rnib.org.uk/education-professionals** or by calling our Helpline on **0303 123 9999**.

At an early stage, ask your local authority to put you in contact with a QTVI. They will support you and your child as soon as a visual impairment is suspected or diagnosed. If you have difficulty getting help or need the details of the specialist teacher in your area, our Children, Young People and Families support officers can help.

You can find out more about the practical help we can offer children, young people and families from our website **rnib.org.uk/children-young-people-education** or by calling our Helpline on **0303 123 9999**.

### The low vision assessment

When coloboma affects a person’s sight at any age, there are specialist support services which can help. If seeing detail with glasses or contact lenses is still difficult, an assessment in a low vision clinic can be beneficial, as a low vision specialist can prescribe magnifiers to use to make the most of the sight that remains. An ophthalmologist, optometrist, ECLO or GP can refer their patients to the local low vision clinic, which is often located in the hospital eye department. It is important for children to have regular low vision assessments because their visual needs change as they grow up.

### Assistive technology

There is also technology available that can help with low vision. Many smart phones and tablets are already equipped with in-built software that can enable people with low vision to access information. There are also specific apps and low vision devices that may help too, as well as computer software programmes that can be installed. If you would like to find out more about the assistive technology that is available and how it can help you, our Technology for Life team advisors would be happy to chat to you. You can get in touch with this team by calling our Helpline on **0303 123 9999**.

### Social services support

If required, your local social services should also be able to offer practical adaptations around the home and advice on getting out and about safely. If needed, they should also be able to offer you some practical mobility training to give you more confidence when you are out.

### Registration

Depending on how much of a person’s sight is affected by coloboma, they may be eligible to be registered as sight impaired (previously referred to as partially sighted) or severely sight impaired (previously referred to as blind). An ophthalmologist would be able to tell you whether registration is appropriate for you or your child. Registration can act as a passport to help and sometimes to financial concessions, but a lot of this support is still available to people who aren’t registered.

## Sources of support

### Helpline

If you have questions about anything you’ve read in this factsheet, or just want to speak further to someone about coloboma, please get in touch with us. It doesn’t matter if you or your child has just been diagnosed with coloboma or you’ve known about it for a while; we’re here to support you at every step.

Our Helpline is your direct line to the support, advice and services you need. We'll help you to find out what's available in your area and beyond, both from RNIB and other organisations.

Whether you want to know more about your eye condition, buy a product from our shop, join our library, find out about possible benefit entitlements, or be put in touch with a trained counsellor, or make a general enquiry, we’re only a call away.

Call our Helpline on **0303 123 9999**, we’re ready to answer your call Monday to Friday 8am – 8pm and Saturday 9am – 1pm. You can also email us at **helpline@rnib.org.uk**. You can also say, “**Alexa, call RNIB Helpline**” to an Alexa-enabled device.

You can also get in touch by post or by visiting our website:

**RNIB**

105 Judd Street

London WC1H 9NE

**rnib.org.uk**

### The Sight Advice FAQ

The Sight Advice FAQ answers questions about living with sight loss, eye health or being newly diagnosed with a sight condition. It is produced by RNIB in partnership with a number of other sight loss organisations. **sightadvicefaq.org.uk**

### Connect with others

You can meet or connect with others who are blind or partially sighted online, by phone or in your community to share interests, experiences and support for each other. From book clubs and social groups to sport and volunteering, our friendly, helpful and knowledgeable team can link you up with opportunities to suit you. Visit **rnib.org.uk/connect** or call **0303 123 9999**.

### Other useful organisations

**MACS (Microphthalmia, Anophthalmia and Coloboma Support)**

An organisation supporting children born without eyes or with underdeveloped eyes.

Helpline: **0800 169 8088**

Email: **enquiries@macs.org.uk**

Web: **macs.org.uk**

**Gene Vision**

A resource on rare genetic eye disorders for everyone **gene.vision**

**ROAM (Research Opportunities At Moorfields)**

Get involved in research

Web: **research.moorfields.nhs.uk**

**LOOK UK**

Supports young people (up to age 29) and families of children living with a visual impairment.

**Telephone:  07464 351958**

Message via form at look-uk.org/contact

**Write: Fred Bulmer Centre, Wall Street, Hereford, HR4 9HP**

**Web: look-uk.org**

**Guide Dogs**

**Supporting children and adults with sight loss with many services, both with and without dogs**

**Helpline: 0800 781 1444 (9am-5pm, Monday to Friday)**

Email:[information@guidedogs.org.uk](mailto:information@guidedogs.org.uk)

Web: **guidedogs.org.uk**

## We value your feedback

You can help us improve our information by letting us know what you think about it. Is this factsheet useful, easy to read and detailed enough – or could we improve it?

Send your comments to us by emailing us at [eyehealth@rnib.org.uk](mailto:eyehealth@rnib.org.uk) or by writing to the Eye Health Information Service, RNIB, 105 Judd Street, London, WC1H 9NE.

### Information sources

This factsheet has been written by the RNIB Eye Health Information service. Our factsheets have been produced with the assistance of patient and carer input and up-to-date reliable sources of evidence. The accuracy of medical information has been checked by medical specialists. If you would like a list of references for any of our factsheets, please contact us at [eyehealth@rnib.org.uk](mailto:eyehealth@rnib.org.uk).

All of our factsheets are available in a range of formats including print, audio and braille.

This factsheet has been produced jointly by RNIB and The Royal College of Ophthalmologists.

RNIB is a member of the Patient Information Forum (PIF) and have been certified under the PIF TICK quality mark scheme.

**Last updated: July 2022**

**Next review: July 2025**

