Coloboma Factsheet
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**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 part of the eye and it can be small or large in size. A coloboma occurs in about 1 in 10,000 births and by the eighth week 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 are able to “see” the world around us.

**Which parts of the eye can coloboma affect?**

**Opposite is a diagram of the eye showing the main structures that can be affected by coloboma**

**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.
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 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, despite the fact that 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 to form the shape and structures of the eye. As the developing eye folds round, it should join together at the bottom from the back to the front 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, it may also affect structures further back in the eye too, if less of the fissure closed properly.

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 find out about the genes involved in their child’s coloboma, while others do not. This is because not all the genes relating to coloboma have been identified yet. Research is going on to find out more about these genes. There is more information about this research on the MACS (Microphthalmia, Anophthalmia and Coloboma Support) website listed in the further help and support section at the end of this information.
**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 fairly good vision. However, iris coloboma is likely to cause light sensitivity, (also called photophobia), in bright conditions. This is because the iris is made of muscle that controls the amount of light entering the eye by changing the size of the pupil. 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 (also known as your 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](http://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 affected because of the change in lens shape caused by the 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. The treatment for cataract can be found in the “Can coloboma lead to other eye health problems?” section further on in this information.

**Chorioretinal coloboma**

Someone with chorioretinal coloboma, where the choroid and the retina are involved, 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 region of their field of vision.

If the chorioretinal coloboma is large or there is optic disc coloboma, they are likely to have poor central vision. This will make activities such as reading, writing and watching television more difficult. If both eyes are affected by optic disc coloboma, they may also have nystagmus, meaning that their 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](http://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 say more accurately 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 or long-sighted, 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.

Where coloboma affects a person’s sight, there are specialist support services which can help them. If they still have difficulty seeing detail with their glasses or contact lenses, they would benefit from having an assessment in a low vision clinic where a low vision specialist can prescribe magnifiers to use to make the most of the sight that they have. An ophthalmologist, optometrist or GP can refer them to their 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 vision needs change as they grow up.

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

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 issues.

You can find out more about education professionals from our website [rnib.org.uk/professionals](http://rnib.org.uk/professionals) or by calling our Helpline on **0303 123 9999**.

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**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 that determines eye colour 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;
- 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 so that the ophthalmologist can examine their eyes thoroughly without distressing them.

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 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 their eye health. 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 these options with you further.

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. 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. A person with coloboma who experiences any new symptoms or concerns should have their eyes examined 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 advise them further 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](http://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 of the retina, choroid or optic disc at the back of the eye. This means that the retina near to the coloboma is more at risk of becoming detached away 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 coming down, up or across their vision.

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 vessels)

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 injection

The body produces a chemical which encourages the growth of these abnormal blood vessels. By interfering with this chemical using drugs called anti-vascular endothelial growth factors (anti-VEGFs), new blood vessel growth and leakage can be reduced. Anti-VEGFs are given as an injection into the white part of the eye and offered as an out-patient procedure.

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. 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 one. This procedure can be more complex for a child with cataracts and coloboma so the ophthalmologist will carry out an in-depth assessment before discussing ways to manage the condition with the child’s family.

You can find more information about 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 health problems.

Some children with coloboma also have other eye problems such as microphthalmia (where the whole eye is small) and anophthalmia (where the eye has not developed at all). Some children with coloboma have other health problems that need to be assessed and so, for this reason, the ophthalmologist may carry out additional tests to check your child’s general health.

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:

- Coloboma
- Heart defects
- Atresia, or blockage, of the nasal passages (choanal atresia)
- Restricted growth and development
- Genital abnormalities
- Ear abnormalities

The ophthalmologist caring for your child will refer them to a paediatrician to carry out the relevant tests that will detect whether or not they have any other general health problems associated with their coloboma, such as CHARGE syndrome.

**Coping with sight problems relating to coloboma**

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 what type of coloboma is present, how large it is and whether it is found 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 hospital care) or an optometrist at least every two years, or as recommended, to ensure that their eye health is monitored.

It can sometimes be helpful to talk about how you are feeling with someone outside of your circle of friends or family. At RNIB, we can help with our telephone Helpline and our Sight Loss Counselling team. Your GP or social worker may also find a counsellor for you if you feel this might help.

Your eye clinic may also have an Eye Clinic Liaison Officer (ECLO), who can be on hand to provide you with further practical and emotional support about your child’s or your own eye condition.
Further help and support
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. Your local authority (LA) should have at least one qualified teacher of children and young people with vision impairment (QTVI) to work with you and your child both at home and at school. A QTVI is a qualified teacher who can provide support with development, play, learning and education. 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, contact RNIB Helpline on 0303 123 9999.

Local social services can help people of all ages who have sight loss to get out and about safely and can offer practical adaptations around the home.

Depending on how much of a person’s sight is affected by coloboma, they may be eligible to be registered as sight impaired (partially sighted) or severely sight impaired (blind). An ophthalmologist would be able to tell you whether you or your child is eligible. 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.

You can find more information about all the support available to children and adults with sight problems on our website rnib.org.uk or by calling our Helpline on 0303 123 9999.

Sources of support
If you have questions about anything you’ve read in this factsheet, or just want to speak to someone about this eye condition, 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. 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, we’re only a call away.

It’s also a way for you to join RNIB Connect, our community for anyone affected by sight loss. RNIB Connect is free to join and you’ll have the chance to meet other people with similar experiences in our helpful, welcoming and supportive community.
Give us a call today to find out how we can help you.

RNIB Helpline
0303 123 9999
helpline@rnib.org.uk

We're ready to answer your call Monday to Friday 8am to 8pm and Saturday 9am to 1pm.

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

RNIB
105 Judd Street
London WC1H 9NE
rnib.org.uk

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: www.macs.org.uk

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

Helpline: 01432 376314
Email: info@look-uk.org
Web: www.look-uk.org

Children and Young People's Services within Guide Dogs (formally Blind Children UK)

Helpline: 0800 781 1444.
Email: cypservices@guidedogs.org.uk
Web: www.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 or by writing to the Eye Health Information Service, RNIB, 105 Judd Street, London, WC1H 9NE.

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

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

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