Autosomal Dominant Optic Atrophy

What is autosomal dominant optic atrophy?

Autosomal dominant optic atrophy is a genetic condition. Optic atrophy is when there is a loss of nerve fibres at the optic nerve. The optic nerve carries visual information from the eyes to the brain. When there are a reduced number of nerve fibres, visual information from the eye cannot be carried to the brain properly. This can affect how well you can see. It can affect your central vision and sometimes your peripheral or side vision as well. How clearly a person sees can range from slightly blurry to very poor vision. The two most common forms of inherited optic atrophy are autosomal dominant optic atrophy (ADOA) and Leber hereditary optic neuropathy (LHON).

The information in this factsheet focuses on autosomal dominant optic atrophy. For further information on Leber hereditary optic neuropathy please see **gene.vision/knowledge-base/leber-hereditary-optic-neuropathy-for-patients/** and **lhonsociety.org** (further details at the end of this factsheet).

What is the optic nerve?

Your optic nerves are vital for sight. They carry the electrical signals from your eye to the brain. The light sensitive photoreceptor cells in your retina at the back of your eye, convert light into electrical signals. These signals are sent along the optic nerve to your brain, where they are interpreted to "see" the world around us.

The start of the optic nerve is known as the optic disc. This is where all the individual nerve fibres from each of the photoreceptor cells in your retina come together. Your optic disc is visible as a small circular area of your retina. It can be seen by the ophthalmologist (hospital eye doctor) or optometrist (optician) when they look into your eye. The nerve fibres travel together in a bundle, from your optic disc, forming your optic nerve.

The optic nerve is made up of 1.2 million nerve fibres. The nerve fibres are made up of retinal ganglion cells and a long section called an 'axon'. The axon is the long section of the nerve cell that connects it the next nerve cell. Having autosomal dominant optic atrophy means that there is slow loss of the retinal ganglion cells (RGCs) and their axons that form the optic nerve.

Diagram of cross section of eye (labels: retina, blood vessels, optic nerve, optic disc)

What are the symptoms of autosomal dominant optic atrophy (ADOA)?

ADOA causes gradual loss of vision in both eyes which is painless. It starts in childhood or teenage years but may not be diagnosed until adulthood in some people. Your vision can slowly worsen over time. ADOA also causes difficulties in recognising colours (dyschromatopsia). The amount of vision loss can vary from person to person, even in the same family. Children with sight loss due to ADOA usually adapt well. With the right help and support they can lead a full and enjoyable life.

Autosomal Dominant Optic Atrophy Plus

Roughly 1 in 5 people with ADOA can have additional symptoms that affect other parts of their body. When this happens, it's called autosomal dominant optic atrophy plus (ADOA+), and it usually comes with more severe visual loss.

Permanent hearing loss is the most common added symptom and occurs in roughly 2 out of 3 people with ADOA+. Hearing loss usually starts after sight loss, often in a person's 20s or 30s.

Other conditions associated with ADOA+ can include:

Weakness in the eye muscles (progressive external ophthalmoplegia)
Muscle weakness (myopathy)
Stiffness in the arms and legs (spasticity)
Numbness or tingling in hands and feet (peripheral neuropathy)
Cataracts, which can make vision cloudy.

What causes autosomal dominant optic atrophy?
ADOA is a genetic condition that affects the optic nerve, which carries visual information from your eyes to your brain. It happens because of a change (mutation) in one of the genes that helps keep the optic nerve healthy.

Your genes give the cells in your body the instructions they need to work well and stay healthy. If a gene has a mutation, the cells using those instructions can't work as they should. In ADOA, the changed gene leads to a slow loss of nerve fibres that make up the optic nerve - the retinal ganglion cells (RGCs) and their axons. The RGCs and their axons gradually stop working over time. RGCs seem to be especially sensitive to problems caused by the gene change.

So far, researchers have identified about eight different genes that can cause ADOA and are still discovering more. The names given to genes are often long, so they are usually identified using letters and numbers. About 75 out of 100 cases of ADOA are linked to a gene called OPA1.

OPA1 is important because it helps make a protein that keeps mitochondria healthy. Mitochondria are tiny structures inside cells that produce energy, like the battery of the cell. They produce the energy necessary for cells to survive and work properly. RGCs need a lot of energy to do their job and rely heavily on mitochondria. When the OPA1 gene isn't working properly, the mitochondria can't make enough energy for the RGCs. This causes the cells to weaken and eventually stop working, resulting in gradual loss of vision.

How is autosomal dominant optic atrophy inherited?
Genes usually come in pairs. You inherit one copy of each gene from each of your parents to make a pair. When you have a child, you only pass on one of the two copies to them.

ADOA is mostly inherited (passed on in families) by autosomal dominant inheritance.

Autosomal dominant inheritance means that one parent has ADOA. Each time that parent has a child there is a 1 in 2 chance of the child having the condition as well.

In the following diagram:

One parent has an autosomal dominant ADOA. This means that they have one changed gene and one normal gene.
The other parent does not have the condition. They have two normal genes.
Every time these parents have a child, there are four possible ways in which their genes can combine. These four possible ways are represented by the four children in the diagram. (The diagram does not show a family with four children in it.)
Each time they have a child, there is a 2 in 4 (50:50) chance that their child will inherit the condition.

There is also a 50:50 chance that they won't inherit the condition.

Other genes identified in autosomal ADOA and ADOA+ are: ACO2, SPG7, AFG3L2, MFN2 and OPA3.

Less commonly, OPA1-related ADOA can also be inherited in an autosomal recessive pattern. Recessively inherited ADOA means that neither parent has the condition themselves, but both parents are 'carriers'. When both parents are carriers, every time they have a child, there is a 1 in 4 chance that the child will have the condition. Both parents need to be carriers of the condition for their child to be affected.

In the following diagram:

Both parents are carriers of an autosomal recessive ADOA. This means that each parent has one changed gene and one normal gene.
Every time these parents have a child, there are four possible ways in which their genes can combine. These four possible ways are represented by the four children in the diagram. (The diagram does not show a family with four children in it.)

Each time they have a child:
There is a 1 in 4 chance that their child will inherit the condition.
There is a 2 in 4 (50:50) chance that the child will be a carrier of the condition.
There is a 1 in 4 chance that the child will inherit two normal genes, meaning they do not have the condition, and they are not carriers either.

How can other family members be affected?
The severity of ADOA can vary widely, both between different families and within the same family. Some people may have only mild vision problems, while others may be severely sight impaired.

Genetic testing
Genetic testing uses a blood test to look at your genes to see if any changes are present. Genetic testing can be carried out to:

Identify the specific gene change that is causing your condition.
Indicate the inheritance pattern to show how likely it is for other members of your family to have the condition.
Indicate whether you could pass on the gene change to your children.
Indicate how your vision may change over time and with what speed.

There are several genetic centres around the country that carry out genetic tests. The service is provided free on the NHS if your
ophthalmologist or your GP refers you to one.

Genetic counselling
Genetic counselling is not talking therapy or an emotional support service. Instead, it aims to provide the information you need to understand your condition. It can also enable you to make informed decisions relating to your condition.

Genetic counsellors are specialists in genetics and counselling. They work in genetic centres to offer you support around your test results. Genetic counselling can be invaluable even before genetic testing is carried out. It may prepare you for your results and what they may potentially mean for you.

If you know of a condition within your family, a genetic counsellor can provide you and your relatives with further information. They can explain how the condition has been passed on within your family by considering your family tree in detail. They can also offer you guidance and support in making any future decisions that may relate to your condition. For example, knowing the chances of passing on a condition may help if you are thinking about starting a family. Genetic counselling is a free service within the NHS. To access this service, you can ask your GP or ophthalmologist to refer you.

How is ADOA diagnosed?
ADOA is most often diagnosed during childhood or adolescence following an unexplained reduction in vision in both eyes. ADOA is diagnosed by an ophthalmologist. They may carry out several tests including:

Examination of the health of your eye
The ophthalmologist will look at the health of your optic nerve at the back of your eye using a bright light to check for any changes. They may also take a photograph of the optic disc at the back of your eye. The ophthalmologist may be able to diagnose ADOA based on the appearance of the optic disc.

Colour vision
ADOA often affects colour vision so a colour vision test will be done. To test your colour vision, you'll be asked to pick out numbers or patterns that you can see on a background of coloured dots. This test takes less than five minutes to do and shows what colours you're able to see.

Visual fields test
A visual field test checks how well you can see above, below and to the sides of what you're looking at. It helps find any missing areas of vision or blind spots.

ADOA can cause blind spots in the centre of your vision. It can also affect your side (peripheral) vision. A visual field test shows how much of your vision has been affected and which areas of vision are affected.

Optical coherence tomography (OCT)
An OCT is a scanner that provides a cross-section image of the inside of the eye using infrared light. It can give a quick but very detailed image of the structures at the optic nerve. This can show up any damage to the cells of the optic nerve and is a helpful way of spotting subtle changes over time.

Electrodiagnostic tests
Electro-diagnostic tests can tell your ophthalmologist how well your retina and optic nerve are working, by measuring responses to light and patterns.

You might have tests including the electroretinogram (ERG) which checks your whole retina, or the pattern electroretinogram (PERG) which uses a checkerboard pattern to check how your macula, at the centre of your retina, is working. The macula provides our central detailed vision and colour vision.

If the RCGs or their axons aren't working properly, the test results will show this.

Is there any treatment for ADOA?
Currently there is no treatment to stop or reverse vision loss from ADOA.

There is ongoing research to improve early diagnosis and detection of hereditary optic neuropathies. Having a diagnosis early on can allow for timely care and support for those affected. It can also help support future trials into the condition. There is also early phase research into potential treatments.

You can learn more about current research for ADOA at: **adoaa.org/treatment-research**

There is much that can be done to help make the most of the sight you have. This includes visual aids and adaptations around the home, adaptations and specialised software for computers at home and at work, and training from social services. Our Sight Loss Advice service can help provide information about all the different practical support available, contact our Helpline to speak with them.

Supporting you with sight loss
When ADOA affects a person's sight at any age, there are specialist support services which can help. There are lots of things that you can do to make the most of the vision you have. This may mean making things bigger, using brighter lighting or using colour to make things easier to see.

The low vision assessment
If seeing detail with glasses or contact lenses is 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, Eye Care Liaison Officer (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 Digital skills advisors would be happy to chat to you.

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 ADOA, 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.

Educational support

For children who have sight loss from ADOA, 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.

You can find out more about the practical help we can offer children, young people and families from our website **Parenting a child with a vision impairment | RNIB | RNIB** or by calling our Helpline on **03031239999**.

Emotional support and counselling services

On hearing that you or your child have issues with sight, you may feel like your world has been turned upside down. Different people respond in different ways but it's natural to experience many emotions which may include shock, fear, grief, sadness, or despair. All these feelings are quite common and part of the process you may go through during the early weeks and months.

You may have left the consulting room in a daze and may not have heard all the words that were said to you during the consultation. You may be asking yourself what happens next and what the future holds.

You probably have a million different questions. You'll want to know which organisations can help you, what resources are available or simply want to know "what do I do next?" You may also want to meet other parents who have been through a similar experience.

It's a good idea to write down any questions that come to your mind on some paper and take it along when attending eye clinic appointments. This will ensure that you don't forget to ask the medical team about them during your visit.

Eye Care Liaison Officer

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 condition or sight loss. To find out if your hospital eye clinic has an ECLO, you can call our Helpline.

Further help and support

If you have questions about anything you've read in this information, please get in touch with us.

RNIB Helpline

If you need someone who understands sight loss, call our Helpline on **0303 123 9999**, say **"Alexa, call RNIB Helpline"** to an Alexa enabled device, or email **helpline@rnib.org.uk**. Our opening hours are weekdays from 9am – 6pm.

You can also get in touch by post or by visiting our website: **rnib.org.uk**

RNIB
The Grimaldi Building
154A Pentonville Road
London N1 9JE

Other useful contacts

Specific to ADOA:

CURE ADOA FOUNDATION
Brings patients together and is committed to research

Helpline: **+31-(0)657-276427**
Email: **info@adoa.eu**
Web: **https://adoa.eu/en**

ADOA Association - The Autosomal Dominant Optic Atrophy Association (ADOAA) was created to raise awareness about ADOA and to help fund the medical research to find a cure.

Email: **lindsey.allen@adoaa.org**
Telephone: +**01 570-419-8799**
Web**: adoaa.org**

Mitochondrial disease related:

Leber's Hereditary Optic Neuropathy (LHON) Society
provides support and information to those impacted by LHON; patients, their family, friends and healthcare providers.

Web: **Patient-led support group for LHON - LHON Society**
Contact via online form: **Contact us - LHON Society**

Lily Foundation provides information and support to patients and families affected by mitochondrial disease.

Telephone: **0300 400 1234**
Email: **liz@thelilyfoundation.org.uk**

NHS Rare Mitochondrial Serviceprovides a comprehensive high quality service for this rare group of disorders, with centres in London, Newcastle and Oxford.

Web: **Home - Rare Mitochondrial Disorders Service**

General:

Contact supports families with disabled children, providing guidance and information as well as bringing families together to support each other.

Helpline: **0808 808 3555**
Email**: info@contact.org.uk**
Web: **contact.org.uk**

Genetic Alliance UK
The Genetic Alliance is a national alliance of patient organisations with a membership of over 130

charities which support children, families and individuals affected by genetic disorders.

Tel: **020 7831 0883**
Email: **contactus@geneticalliance.org.uk**
Web: **geneticalliance.org.uk**

Gene Vision
A resource on rare genetic eye disorders for everyone.

Web: **gene.vision**

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

Tel: **07464 351958**
Message via form at **look-uk.org/contact**
Write to Fred Bulmer Centre, Wall Street, Hereford, HR4 9HP
Web: **look-uk.org**

Guide Dogs - Children and Young People's service (formerly Blind Children UK) supports children and adults with sight loss with many services, both with and without dogs.

Tel: **0800 781 1444**
Web: **guidedogs.org.uk**

Royal Society for Blind Children provide a range of services in London and across England and Wales for blind and partially sighted children and young people, their families, and the professionals who work alongside them.

Tel: **020 3198 0225** (9am-5pm, Monday to Friday)
Email: **connections@rsbc.org.uk**
Web: **rsbc.org.uk**

Visually Impaired Children Taking Action (Victa)
Tel: **01908 240831**
Email: **admin@victa.org.uk**
Web: **victa.org.uk**

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Is this factsheet useful, easy to read and understand?
Is there anything missing?
How clear, relevant and helpful did you find the images and diagrams?
How 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, Grimaldi Building 154a Pentonville Road, London N1 9JE.

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**.

Our factsheets are available in print, audio and braille. To request these formats, contact our Helpline **0303 123 9999**.

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