



Optic atrophy type 1

Optic atrophy type 1 is a condition that often causes slowly worsening vision, usually beginning in childhood. People with optic atrophy type 1 typically experience a narrowing of their field of vision (tunnel vision). Affected individuals gradually lose their sight as their field of vision becomes smaller. Both eyes are usually affected equally, but the severity of the vision loss varies widely, even among affected members of the same family, ranging from nearly normal vision to complete blindness.

In addition to vision loss, people with optic atrophy type 1 frequently have problems with color vision (color vision deficiency) that make it difficult or impossible to distinguish between shades of blue and green.

In the early stages of the condition, individuals with optic atrophy type 1 experience a progressive loss of certain cells within the retina, which is a specialized light-sensitive tissue that lines the back of the eye. The loss of these cells (known as retinal ganglion cells) is followed by the degeneration (atrophy) of the nerves that relay visual information from the eye to the brain (optic nerves), which results in further vision loss. Atrophy causes these nerves to have an abnormally pale appearance (pallor), which can be seen during an eye examination.

Frequency

Optic atrophy type 1 is estimated to affect 1 in 35,000 people worldwide. This condition is more common in Denmark, where it affects approximately 1 in 10,000 people.

Causes

Optic atrophy type 1 is caused by mutations in the *OPA1* gene. The protein produced from this gene is made in cells and tissues throughout the body. The *OPA1* protein is found within mitochondria, which are the energy-producing centers of cells. The protein plays a key role in the organization of the shape and structure of the mitochondria and in controlled cell death (apoptosis). The *OPA1* protein is also involved in a process called oxidative phosphorylation, from which cells derive much of their energy. Additionally, the protein plays a role in the maintenance of the DNA within mitochondria, called mitochondrial DNA (mtDNA).

Mutations in the *OPA1* gene lead to problems with mitochondrial function. The mitochondria become misshapen and disorganized and have reduced energy-producing capabilities. The maintenance of mtDNA may also be impaired, resulting in mtDNA mutations that further interfere with mitochondrial energy production. Cells that contain these poorly functioning mitochondria are more susceptible to apoptosis. In particular, cells that have high energy demands, such as retinal ganglion cells, die over time. Specialized extensions of retinal ganglion cells, called axons, form the optic

nerves, so when retinal ganglion cells die, the optic nerves atrophy and cannot transmit visual information to the brain.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

In most cases, an affected person inherits the mutation from one affected parent. Other cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

In rare cases, people who have an *OPA1* gene mutation do not develop optic atrophy type 1, a situation known as reduced penetrance.

Other Names for This Condition

- ADOA
- autosomal dominant optic atrophy
- autosomal dominant optic atrophy Kjer type
- DOA
- dominant optic atrophy
- Kjer type optic atrophy
- Kjer's optic atrophy
- optic atrophy, autosomal dominant
- optic atrophy, hereditary, autosomal dominant
- optic atrophy, juvenile
- optic atrophy, Kjer type

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
[/primer/testing/genetic-testing](#)
- Genetic Testing Registry: Dominant hereditary optic atrophy
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0338508/>

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22Optic+Atrophy%2C+Autosomal+Dominant%22+OR+%22optic+atrophy+type+1%22>

Other Diagnosis and Management Resources

- GeneReview: Optic Atrophy Type 1
<https://www.ncbi.nlm.nih.gov/books/NBK1248>
- MedlinePlus Encyclopedia: Optic Nerve Atrophy
<https://medlineplus.gov/ency/article/001622.htm>
- MedlinePlus Encyclopedia: Visual Acuity Test
<https://medlineplus.gov/ency/article/003396.htm>

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Optic Nerve Atrophy
<https://medlineplus.gov/ency/article/001622.htm>
- Encyclopedia: Visual Acuity Test
<https://medlineplus.gov/ency/article/003396.htm>
- Health Topic: Color Blindness
<https://medlineplus.gov/colorblindness.html>
- Health Topic: Optic Nerve Disorders
<https://medlineplus.gov/opticnervedisorders.html>

Genetic and Rare Diseases Information Center

- Optic atrophy 1
<https://rarediseases.info.nih.gov/diseases/9890/optic-atrophy-1>

Additional NIH Resources

- eyeGENE: The National Ophthalmic Disease Genotyping and Phenotyping Network
<https://eyegene.nih.gov/>
- National Eye Institute: Living with Low Vision
<https://nei.nih.gov/sites/default/files/health-pdfs/LivingWithLowVisionBooklet.pdf>

Educational Resources

- American Association for Pediatric Ophthalmology and Strabismus
<https://engage.aapos.org/browse/glossary/entry?GlossaryKey=9e3d1bd6-1817-4743-803f-a3b43ca647ba>
- Centers for Disease Control and Prevention: Vision Loss Fact Sheet
https://www.cdc.gov/ncbddd/actearly/pdf/parents_pdfs/VisionLossFactSheet.pdf
- KidsHealth from Nemours Foundation: Color Blindness Special Needs Factsheet
<https://kidshealth.org/en/parents/color-blind-factsheet.html>

- Merck Manual Consumer Version: Overview of Optic Nerve Disorders
<https://www.merckmanuals.com/home/eye-disorders/optic-nerve-disorders/overview-of-optic-nerve-disorders>
- Orphanet: Autosomal dominant optic atrophy
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=98672
- The University of Michigan Kellogg Eye Center: Optic Atrophy
<https://www.umkelloggeye.org/conditions-treatments/optic-atrophy>

Patient Support and Advocacy Resources

- American Foundation for the Blind
<https://www.afb.org/>
- Autosomal Dominant Optic Atrophy Association
<https://www.adoaa.org/>
- Foundation Fighting Blindness (Canada)
<https://www.fightingblindness.ca/>
- Prevent Blindness America
<https://www.preventblindness.org/>

Clinical Information from GeneReviews

- Optic Atrophy Type 1
<https://www.ncbi.nlm.nih.gov/books/NBK1248>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28optic+atrophy+type+1%5BTIAB%5D%29+OR+%28autosomal+dominant+optic+atrophy%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- OPTIC ATROPHY 1
<http://omim.org/entry/165500>

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