CONDITION

Choroidal dystrophies

Learn how inherited choroidal dystrophies affect the retina and vision, including symptoms, genetic causes, testing, treatment, and follow-up.

WHAT IT IS
Inherited conditions causing gradual back-of-eye damage

BODY SYSTEM
Eyes and vision

MAIN SYMPTOMS
Night-vision difficulty, side-vision loss, reduced central vision, blind spots

WHAT CAUSES IT
Inherited gene changes

HOW IT IS DIAGNOSED
Retinal examination and imaging, genetic testing

MAIN TREATMENT
Long-term monitoring and supportive care

CAN IT BE CURED?
No, no single treatment for all

IS IT AN EMERGENCY?
Sometimes, sudden vision loss or retinal-detachment signs

LONG-TERM OUTLOOK
Vision may remain useful for decades but severe sight loss can occur

Overview

Choroidal dystrophies are a group of inherited eye conditions that cause gradual damage at the back of the eye. They often affect the choroid, a layer of blood vessels beneath the retina. The retina is the light-sensitive tissue that turns images into signals for the brain.

Many of these conditions also damage the retinal pigment epithelium, or RPE. This thin layer supports the retina's light-sensing cells. Because the choroid, RPE, and retina work closely together, inherited disease may affect more than one layer.

"Choroidal dystrophy" is an older, broad term rather than one diagnosis. Modern classification often uses the specific gene and retinal structures involved. Examples include choroideremia, gyrate atrophy of the choroid and retina, and some forms of central areolar choroidal dystrophy. Other inherited retinal diseases can produce similar areas of tissue loss.

The course differs by condition. Some mainly affect night and side vision for many years. Others begin near the center of the retina and reduce detailed vision used for reading and recognizing faces. Vision may remain useful for decades, but some forms can eventually cause severe sight loss.

An accurate diagnosis matters because inheritance patterns, expected progression, and treatment options differ. It can also help relatives understand whether genetic testing or an eye examination may be appropriate.

Symptoms

Symptoms depend on which part of the retina is affected and the person's age. They usually develop gradually rather than appearing over hours or days.

Possible symptoms include:

Choroideremia commonly begins with night-vision difficulty, often in childhood, followed by progressive loss of side vision. Central vision may remain relatively preserved until later in life.

Gyrate atrophy may also cause night blindness and shrinking side vision. Nearsightedness and cataracts can occur. Central areolar choroidal dystrophy more often affects the central macula, the retinal area responsible for sharp vision.

Symptoms can differ even among relatives with the same genetic change. Some people, including women who carry certain X-linked changes, may have visible retinal findings but few symptoms. Others may develop meaningful vision problems.

Sudden vision loss is not the usual pattern for an inherited choroidal dystrophy. Seek urgent eye care for new flashes of light, a sudden increase in floaters, a dark curtain over part of the vision, or a sudden drop in sight. These symptoms can indicate a retinal tear, retinal detachment, bleeding, or another problem that needs prompt assessment. Severe eye pain, marked redness, or sudden vision loss with weakness, facial drooping, or speech difficulty requires emergency care.

- Trouble seeing in dim light or taking longer to adjust after entering a dark room
- Loss of side vision, sometimes described as tunnel vision
- Reduced central or detailed vision
- Difficulty reading, driving, or recognizing faces
- Blind spots in the visual field
- Increased sensitivity to bright light or glare

- Reduced color discrimination
- Frequent changes in glasses without full improvement in vision

Causes and risk factors

Most choroidal dystrophies are caused by a disease-related change in a gene needed for retinal health. The condition may be inherited in different ways.

Choroideremia is usually caused by a change in the CHM gene on the X chromosome. Males with the gene change are typically affected. Females with one changed copy are often called carriers, but some develop patchy retinal changes or vision symptoms because cells use their X chromosomes differently.

Gyrate atrophy is caused by changes in both copies of the OAT gene. This is called autosomal recessive inheritance. The gene changes interfere with an enzyme that processes ornithine, an amino acid. Ornithine then builds up in the blood and contributes to retinal and choroidal damage.

Some central areolar choroidal dystrophies are associated with changes in genes such as PRPH2 . These may follow an autosomal dominant pattern, meaning one changed gene copy can be enough to cause disease. However, central retinal atrophy has several possible genetic and non-genetic causes.

A person may be the first in a family to receive a diagnosis. This can happen when earlier relatives had mild or unrecognized disease, when a condition is recessive, or when a new genetic change occurs. The absence of a known family history does not rule out an inherited disorder.

Age does not cause these conditions, although symptoms may become more noticeable with time. They should not be assumed to be age-related macular degeneration based only on areas of retinal atrophy.

Diagnosis

Diagnosis usually begins with a detailed history and examination by an ophthalmologist. A retinal specialist or an ophthalmologist who focuses on inherited retinal disease may be involved.

The clinician may ask about night vision, side vision, central vision, and the age when symptoms began. A three-generation family history can help identify an inheritance pattern.

Information about relatives with unexplained poor vision, night blindness, or a diagnosis such as macular degeneration may be useful.

Testing may include:

If gyrate atrophy is suspected, blood testing can measure ornithine. This is important because gyrate atrophy has metabolic management options that do not apply to most other choroidal dystrophies.

The examination also helps rule out other causes of chorioretinal damage, including inflammation, infection, medication toxicity, high myopia, and age-related macular degeneration. A genetic result should be interpreted alongside the person's symptoms, examination, and retinal imaging.

- Dilated eye examination: Eye drops widen the pupils so the retina, RPE, and choroid can be examined.
- Optical coherence tomography: OCT uses light to create cross-sectional images of the retina. It can show thinning, loss of retinal layers, swelling, or remaining central tissue.
- Fundus autofluorescence: This imaging test maps stress and loss within the RPE and can help document progression.
- Visual-field testing: This measures central and side vision and identifies blind spots.
- Electroretinography: ERG records the retina's electrical response to light and can show whether rod cells, cone cells, or both are affected.
- Color or low-light testing: These tests provide more detail about how vision functions.
- Genetic testing: A blood or saliva sample may identify the responsible gene. Testing is most useful when paired with genetic counseling because results can be positive, negative, or uncertain.

Treatment and management

There is no single treatment for all choroidal dystrophies. Management depends on the specific diagnosis, gene, current vision, and any treatable complications.

Regular eye care can identify changes that may benefit from treatment. Cataracts can sometimes be removed when they meaningfully affect vision. Retinal swelling, abnormal blood-vessel growth, or retinal tears may require medication, laser treatment, or surgery, depending on the problem.

Gyrate atrophy may be managed by lowering ornithine. This can involve a carefully planned restriction of arginine, an amino acid found in protein, and testing for response to vitamin B6.

These measures require supervision by clinicians experienced in metabolic disease and a registered dietitian. Restricting protein or taking high-dose vitamins without guidance can cause harm.

No vitamin or supplement has been shown to treat every choroidal dystrophy. Vitamin A and other high-dose supplements should not be started specifically for retinal disease unless the treating clinician recommends them. Their usefulness and risks depend on the exact condition and the person's broader health.

Gene-based treatment is an active area of research. Voretigene neparvovec is an approved gene therapy for a specific retinal dystrophy caused by disease-related changes in both copies of the RPE65 gene. It is not a general treatment for choroideremia or other choroidal dystrophies. Gene therapy for choroideremia and several other inherited retinal diseases remains under study.

A clinical trial may be an option for some people with a confirmed genetic diagnosis. Participation is voluntary. The research team should explain the study's purpose, possible burdens, uncertain benefits, alternatives, and follow-up requirements.

Low-vision rehabilitation can be helpful before vision becomes severe. It may include magnifiers, electronic reading tools, screen-reader software, improved lighting, glare control, orientation and mobility training, and workplace or school accommodations. Driving safety should be discussed if side vision, night vision, or central vision has declined.

Tinted lenses may improve comfort in bright conditions, although they do not stop the genetic disease. General eye protection, not smoking, and management of conditions such as diabetes and high blood pressure support overall eye health.

Complications

Progressive retinal and choroidal loss can lead to narrowing of the visual field or reduced central vision. This may affect driving, reading, work, independent travel, and the ability to see in low light.

Some people develop additional eye problems, including:

These problems are not inevitable, and some are treatable. A new symptom should not automatically be attributed to the inherited dystrophy.

Changes in vision can also affect emotional health. Worry about future sight, employment, parenting, or passing a condition to children is understandable. Genetic counseling, low-vision services, mental health support, and patient organizations can help with planning and adjustment.

- Cataracts
- Swelling in the central retina
- Abnormal blood-vessel growth beneath the retina
- Retinal tears or detachment
- Severe nearsightedness in certain conditions

Follow up

Follow-up is usually long term. The schedule depends on the diagnosis, rate of change, symptoms, and whether complications are present. Routine visits may include a dilated examination, OCT, retinal imaging, and visual-field testing. Repeating the same tests over time helps the clinician judge whether the condition is stable or progressing.

Genetic results may need to be reviewed again as scientific knowledge changes. A previously uncertain result may later be reclassified. People interested in research may also ask whether a clinical-trial registry or inherited-retinal-disease center is appropriate.

Family members should not assume that they are affected or unaffected based only on symptoms. When a disease-causing gene change is confirmed, a genetic counselor can explain who may wish to consider targeted testing.

Contact the eye clinic promptly for a noticeable new change in vision. Seek urgent same-day assessment for flashes, many new floaters, a curtain-like shadow, or sudden visual distortion. Sudden major vision loss, severe eye pain, or vision changes accompanied by signs of a possible stroke require emergency care.

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