Juvenile Macular Degeneration

What is juvenile macular degeneration?

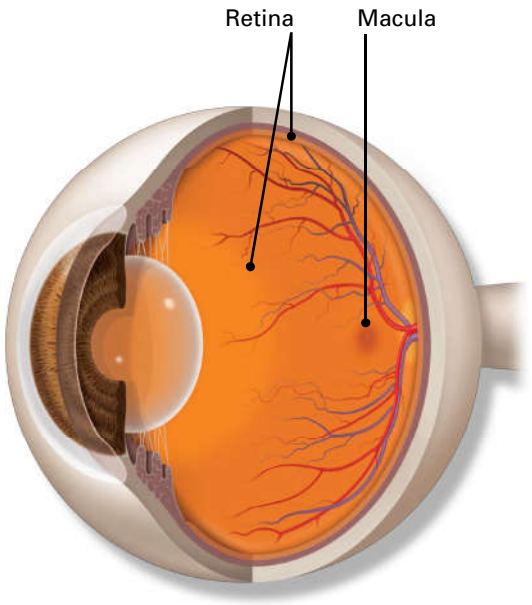
Juvenile macular degeneration is a group of inherited eye disorders that affects children and young adults. Juvenile macular degeneration is different from age-related macular degeneration (AMD). AMD occurs as part of the body's natural aging process. Juvenile macular degeneration is sometimes called macular dystrophy.

Macular degeneration is a deterioration or breakdown of the eye's macula. The macula is a small area in the retina — the light-sensitive tissue lining the back of the eye. The macula gives you your central vision and allows you to see fine details clearly. The macula makes up only a small part of the retina, yet it is much more sensitive to detail than the rest of the retina (called the peripheral retina). The macula is what allows you to thread a needle, read small print, and read street signs. The peripheral retina gives you side (or peripheral) vision. If someone is standing off to one side of you, your peripheral retina helps you know that. It does so by allowing you to see their general shape.

The most common form of juvenile macular degeneration is Stargardt disease. Other types of juvenile macular degeneration include:

- Best's disease (also called Best's vitelliform retinal dystrophy), and
- juvenile retinoschisis

All these diseases are rare and cause central vision loss. Unfortunately, there is no treatment available to prevent vision loss.



What are the symptoms of juvenile macular degeneration?

All forms of juvenile macular degeneration share similar characteristics. Juvenile macular degeneration causes problems with your central vision. Your central vision may be blurry, distorted or have dark areas. Side vision is usually not affected, but color perception may be affected in the later stages. Symptoms first appear in childhood or adolescence. These symptoms do not always affect each eye equally.

Some people with juvenile macular degeneration keep useful vision into adulthood. For others, the disease progresses more rapidly. People with Best's disease often have vision that is nearly normal for many decades. Many people may not be aware that they even have it. By contrast, Stargardt disease often results in vision of 20/200. This is the definition of legal blindness.

Juvenile retinoschisis also results in vision loss, ranging from 20/60 to 20/120. About half of people with the disease lose side vision. By age 60 or older, vision loss may reach 20/200.

Children with juvenile retinoschisis may also show signs of:

- strabismus (misaligned eyes) and
- nystagmus (involuntary eye movement)

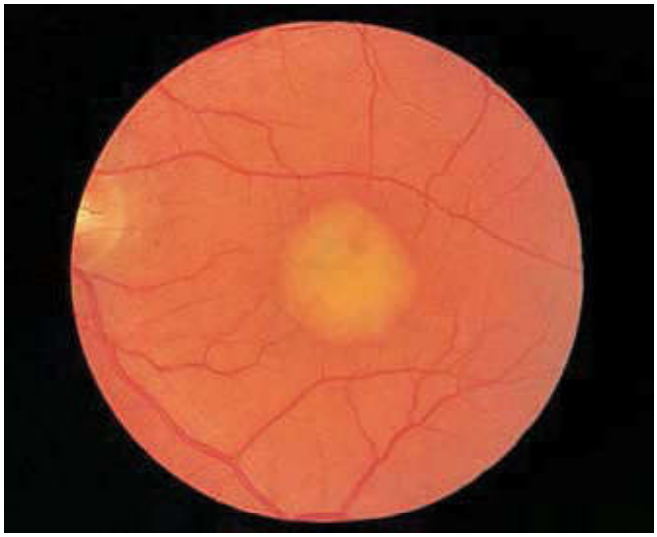
Who is at risk for juvenile macular degeneration?

Juvenile macular degeneration is an inherited genetic disorder. This means the disease passes from parent to child. Different types of juvenile macular degeneration have different inheritance patterns.

For example, Stargardt disease is recessive. This means a child must inherit it from both parents for the disease to develop.

Best's disease is a dominant gene. This means a child only has to inherit the gene from one parent to develop the disease. When an affected person has children with an unaffected partner, there is a 50% (5 out of 10) chance a child will get the disease.

Juvenile retinoschisis is an X-linked disorder and overwhelmingly affects males. The genetic mutation that causes the disease is found on the X chromosome. Males inherit this chromosome from their mothers. (Fathers contribute the Y chromosome.)



An image of a macula with Best disease (a form of hereditary juvenile macular degeneration) that has an "egg yolk" appearance. Vision is usually good early on but scarring may occur years later that may harm vision.

How is juvenile macular degeneration diagnosed?

Your ophthalmologist will conduct a dilated eye examination to look at the retina. People with juvenile macular degeneration have signs specific to their disorder.

People with Stargardt disease may have yellowish flecks in and under the macula. These flecks sometimes extend outward in a ring-like fashion. The flecks are deposits of lipofuscin, a fatty byproduct of normal cell activity. Liposfuscin builds up abnormally in patients with Stargardt disease.

Patients with Best's disease have a yellow cyst that forms under the macula. The cyst eventually ruptures, spreading fluid and yellow deposits. These may harm the macula.

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With juvenile retinoschisis, the retina splits into two layers, which affects the macula. The spaces between these layers can be filled with blisters. Blood vessels can leak into the vitreous, which is the fluid filling the eye. Juvenile retinoschisis can lead to retinal detachments.

Your ophthalmologist may perform a fluorescein angiography to confirm the diagnosis. In this test, a dye is injected into your arm. The dye is photographed as it circulates through the blood vessels in your retina. Your ophthalmologist may also order an ERG (electroretinography) test. This test measures the electrical activity of the retina.

How is juvenile macular degeneration treated?

There is no cure for juvenile macular degeneration. There is no treatment to slow its progression.

Wearing sunglasses to protect the eyes from UV light and bright light is helpful. Low vision aids and mobility training can help people adjust to their vision loss.

Summary

Juvenile macular degeneration is a group of inherited eye disorders. It affects children and young adults. It passes from parent to child. The most common form of juvenile macular degeneration is Stargardt disease. Other types include Best's disease and juvenile retinoschisis. Juvenile macular degeneration causes problems with your central vision.

Your ophthalmologist will conduct a dilated eye examination to diagnose juvenile macular degeneration. Though there is no cure, sunglasses, low vision aids, and mobility training can help patients adjust to their vision loss.

If you have any questions about your vision, speak with your ophthalmologist. He or she is committed to protecting your sight.

Get more information about juvenile macular degeneration from EyeSmart—provided by the American Academy of Ophthalmology—at **aao.org/juvenile-macular-degeneration-link**.

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