What Is Juvenile Macular Degeneration?

Juvenile macular degeneration is a series of inherited eye disorders that affects children and young adults. Juvenile macular degeneration is different from age-related macular degeneration, which 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 is the part of the retina that is responsible for your central vision, allowing 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 your vision, your peripheral retina helps you know that person is there 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), which is pictured above, and juvenile retinoschisis. All of these diseases are rare and cause central vision loss. Unfortunately, there is no treatment available to prevent vision loss.

Juvenile Macular Degeneration Symptoms

All forms of juvenile macular degeneration share similar characteristics. Because it affects the macula, which allows you to see fine details, juvenile macular degeneration causes problems with your central vision, which may be blurry, distorted or have dark areas. Side vision is usually not affected, but color perception may be affected in the later stages of the disease. Symptoms first appear in childhood or adolescence and may not always affect each eye equally.

Some people with juvenile macular degeneration are able to retain useful vision into adulthood, while for others the disease progresses more rapidly. Patients with Best's disease often have vision that is nearly normal for many decades. This disease is inherited and often many family members may not be aware that they even have it. By contrast, Stargardt's disease often results in vision of 20/200, which 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. In addition, 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, meaning the disease is passed from parent to child. Different types of juvenile macular degeneration have different inheritance patterns. For example, Stargardt disease is recessive, meaning it has to be inherited from both parents for the disease to develop. Best's disease is a dominant gene, meaning 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 percent chance that the affected parent will pass the disease-causing gene to each child.

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

Juvenile Macular Degeneration Diagnosis

Your ophthalmologist (Eye M.D.) 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 that sometimes extend outward in a ring-like fashion. The flecks are deposit 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, which may harm the macula.

In patients with juvenile retinoschisis, the retina splits into two layers affecting the macula. The spaces between the layers can be filled with blisters, and blood vessels can leak into the vitreous, the fluid filling the eye. Juvenile retinoschisis can lead to retinal detachments.

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

Juvenile Macular Degeneration Treatment

There is no cure for juvenile macular degeneration and 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 be useful for patients adjusting to their vision loss.