FOUNDATION FIGHTING BLINDNESS

BEST DISEASE

What is Best Disease?

Best disease, also known as vitelliform macular dystrophy, is an inherited form of macular degeneration. Best disease affects the macula, the central part of the retina, and is characterized by a loss of central vision, as well as the ability to perceive colors and details.

What are the symptoms?

Although the age of onset for those with Best disease can vary, it is usually diagnosed during childhood or adolescence. In the initial stages, a bright yellow cyst (fluid-filled sac) forms under the retinal pigment epithelium (RPE) beneath the macula. Upon examination, the cyst looks like a sunny-side-up egg. Despite the presence of the cyst, visual acuity may remain normal or near normal (between 20/30 and 20/50) for many years. Peripheral (side) vision usually remains unaffected.

In many individuals with Best disease, the cyst eventually ruptures. Fluid and yellow deposits from the ruptured cyst spread throughout the macula. At this point the macula has a scrambled egg appearance. Once the cyst ruptures, the macula and the underlying RPE begin to atrophy (degenerate) causing further vision loss.

Retinas affected by Best disease also have the accumulation of yellow flecks called lipofuscin, which can also cause vision loss.

In many people with Best disease, central vision deteriorates to about 20/100 later in life. However, the condition does not always affect both eyes equally. Many individuals retain useful central vision in one eye with a visual acuity of about 20/40 in the lesser-affected eye.

In some cases, Best disease does not progress far enough to cause significant central vision loss. However, retinal specialists can still detect the disease using sophisticated diagnostic tests that measure the function of the RPE and the retina. Individuals with Best

disease are also often farsighted. Farsightedness can be corrected with glasses.

Is it inherited?

Best disease is genetically passed through families by the autosomal dominant pattern of inheritance. In this pattern of inheritance, an affected person has one Best disease gene paired with one normal gene. When the 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. The unaffected partner will only pass normal genes. A child who does not have the Best disease gene will not have the disease and cannot then pass the disease to his or her children.

What treatment is available?

Foundation-funded research is directed at understanding the cause of Best disease and developing treatments to save and restore vision. Researchers have identified the genetic mutations that cause Best disease, and developed a large animal model of the condition for evaluating potential treatments. Investigators at the University of Pennsylvania are currently developing a gene therapy to correct the underlying genetic defect and halt the disease process.

Individuals with Best disease may benefit from the use of low-vision aids and, possibly, orientation and mobility training. Orientation and mobility training, adaptive training skills, job placement, and income assistance are available through community resources.

Are there any related diseases?

Stargardt disease is another inherited form of macular degeneration characterized by numerous yellow flecks (lipofuscin) in the retina similar to those in Best disease. In older adults, it may also be difficult to distinguish Best disease from age-related macular degeneration. A thorough ophthalmologic examination including diagnostic tests measuring retinal function and an accurate documentation of family history can help to distinguish between these related conditions.