RETINA australia

Best disease

Best disease, also known as vitelliform macular dystrophy, was discovered in 1905 by Friedrich Best, a German ophthalmologist. Best Disease is an inherited form of retinal degeneration affecting the portion of the retina known as the macula. The macula is the central part of the retina containing the photoreceptor cells known as cone cells, which are responsible for fine visual detail and colour perception.

Best disease is usually diagnosed during the teenage years, but vision does not generally deteriorate until later in life. The progression of visual loss varies between individuals, but side or peripheral vision usually remains unaffected. Therefore, people with Best disease do not generally have issues with independent mobility.

What are the symptoms of Best disease?

The first symptoms of Best disease can vary from person to person but always involves the central vision. In the initial stages, a fatty yellow pigment builds up in the cells underneath the macula. Over time, the abnormal accumulation of this substance can damage the cone cells located in the macula, leading to a blurring or distortion of central vision. Best disease generally doesn't affect peripheral or side vision. It does not always affect both eyes equally and clearer vision is sometimes retained in one eye.

What is the cause of Best disease and how is it inherited?

Best disease is a genetic disease and is caused by mutations in the BEST1 gene which produces the protein Bestrophin-1. Best disease is passed down through families by an autosomal dominant pattern of inheritance.

In this pattern of inheritance, an affected person has a mutated BEST1 gene paired with a normal copy of the BEST1 gene. When the affected person has children with an unaffected partner, there is a 50 per cent chance that the affected parent will pass the mutated BEST1 gene to each child. The unaffected partner will only pass normal copies of BEST1 genes. A child who does not have a mutated BEST1 gene will not have the disease and cannot then pass the disease to his or her children.

Owing to the variable expression of the BEST1 mutation some people who have the mutation may have very mild, or in rare cases no, symptoms.

What treatments are available?

Currently, there is no treatment for Best disease. Genetic research identified the BEST1 gene as causative for Best disease in 1998 and researchers are now working on understanding the function of this gene in the retina.

Recently, there have been encouraging gene therapy studies in preclinical models of Best disease. These findings mark the first clear steps to developing a therapy that could prevent vision loss. Using this approach, which has proven safe in other retinal conditions, researchers engineer small, safe viruses to deliver the correct version of the BEST1 gene to the retina.

An observational study has been underway at the Mayo Clinic, U.S.A. (U.S.A.) since 2014. Information about clinical trials that are currently being conducted worldwide can be found on www.clinicaltrials.gov and can be searched by condition and trial location.

Research into Best disease is also being undertaken in Australia. Retina Australia has awarded a 2019 grant to Dr Michael O'Connor from Western Sydney University for a project called "the mechanism by which C.1 rescues some functions in Best Disease Retinal Pigment Epithelium". We look forward to hearing Dr O'Connor speak about this work at a future national conference.