Published on *My46* (https://www.my46.org) 09/16/2021

My46 Trait Profile

Best vitelliform macular dystrophy

Other Names: Best disease, vitelliform macular dystrophy type 2

Best vitelliform macular dystrophy is a slowly progressing childhood eye disease resulting in central vision loss. It is caused by mutations in the BEST1 gene that makes a protein called bestrophin.

Characteristics of Best vitelliform macular dystrophy

Symptoms of best vitelliform macular dystrophy (BVMD) typically begin to appear in childhood or teenage years but can vary even within the same family. The symptoms generally progress slowly. At first, an egg yolk-like cyst develops over the retinal pigment epithelium (RPE) that is located at the back of the eye. On dilated eye examination, this has the appearance of a sunny-side up egg. Even with the presence of a cyst, individuals can maintain good visual function. Over time, this cyst may break open, leading to a "scrambled egg" appearance. Distorted vision (metamorphopsia) and a decrease in the clarity of vision (visual acuity) develop centrally. Although it is difficult to predict the severity of vision loss based on what is seen in the eye, vision is often better than expected. While vision loss can be substantial, it is not to the point of legal blindness. Legal blindness is defined as less than or equal to a visual acuity of 20/200. This means that the individual can see an object clearly at 20 feet that an individual without nearsightedness (myopia) could see from 200 feet. When vision loss occurs, it often affects one eye more than the other.

Diagnosis/Testing

The clinical diagnosis of BVMD can be made based on an examination of the back of the eye (fundus), electrooculography (EOG), and family history evaluation. The fundus examination by a medical doctor who is specializes in eye and vision care (ophthalmologist) can detect the typical yolk-like cyst found in this condition whereas an EOG can detect the characteristic difference in electrical charge between the front and the back of the eye (reduced Arden ratio) of BVMD.

Genetic testing can confirm the clinical diagnosis by detecting a change or mutation in a gene called BEST1. This gene makes the bestrophin protein that is involved in ion transport in the RPE. Mutations in this gene interfere with the function of bestrophin. This causes fluid build-up that develops into a cyst.

Management/Surveillance

Annual eye exams by an ophthalmologist are important. These visits improve quality of life as much as possible, allowing for vision assessments, updated refractions (to determine the need for corrective lenses), and low vision evaluations as needed. Individuals with BVMD often require low vision aids. Smoking should be avoided. Laser therapy and anti-vascular endothelial growth factor (anti-VEGF) injections may be used to treat abnormal blood vessel growth from the choroid (choroidal neovascularization) or bleeding, if either occurs.

Mode of inheritance

BVMD is typically inherited in an autosomal dominant pattern. This means inheriting one BEST1 mutation is enough for an individual to be affected and show signs of BVMD. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

There have been cases of BVMD inherited in an autosomal recessive pattern, although this is rare. In these families, an individual has to inherit two BEST1 mutations (i.e., one from each parent) to be affected with BVMD.

Risk to family members

In the autosomal dominant form of BVMD, the risk to family members depends on whether or not the individual with BVMD has a parent affected with BVMD. If a parent also has BVMD, the risk of having a child with BVMD is 50% with each pregnancy. If a parent does not have BVMD, the risk of other siblings being affected is very low.

In the autosomal recessive form of BVMD, if a sibling of a child with BVMD is unaffected, he/she has a 2 in 3 (66%) chance of being a carrier of BVMD.

Special considerations

None

Resources

Genetics Home Reference: Vitelliform Macular Dystrophy
http://ghr.nlm.nih.gov/condition/vitelliform-macular-dystrophy
Royal National Institute of Blind People (RNIB)
https://www.rnib.org.uk/eye-health-eye-conditions-z-eye-conditions/best-disease-vitelliform-macular-degeneration

References

MacDonald I, Lee T. (Updated 12 December 2013). Best Vitelliform Macular Dystrophy. In: GeneReviews at GeneTests Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1993-2014. Available at http://www.ncbi.nlm.nih.gov/books/NBK1167/. Accessed [05/07/2014].

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