What You Should Know About Best Disease

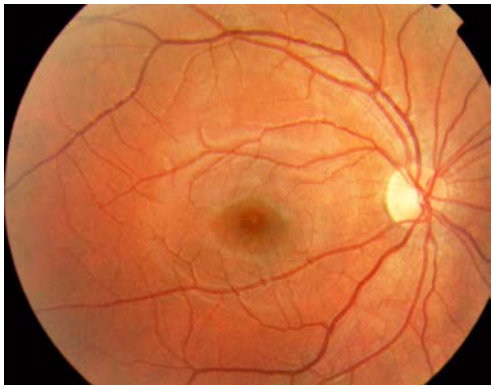
David J. Browning, MD, Ph.D.

Best disease is an inherited retinal condition in which metabolic waste material builds up under the center of the retina. Initially there is little effect on vision, but as the years pass, reading vision can diminish and in later life legal blindness may develop. The peripheral vision is not affected.

In most cases Best disease is inherited in an autosomal dominant pattern. This means that only one mutated gene must be inherited to develop the disease. The gene may be inherited from the father or the mother. Occasionally an autosomal recessive inheritance pattern is found. In these cases, two copies of the mutated gene must be inherited, one from each parent. In most cases the gene mutation involves the BEST1 gene which is located on chromosome 11 q12–q13.

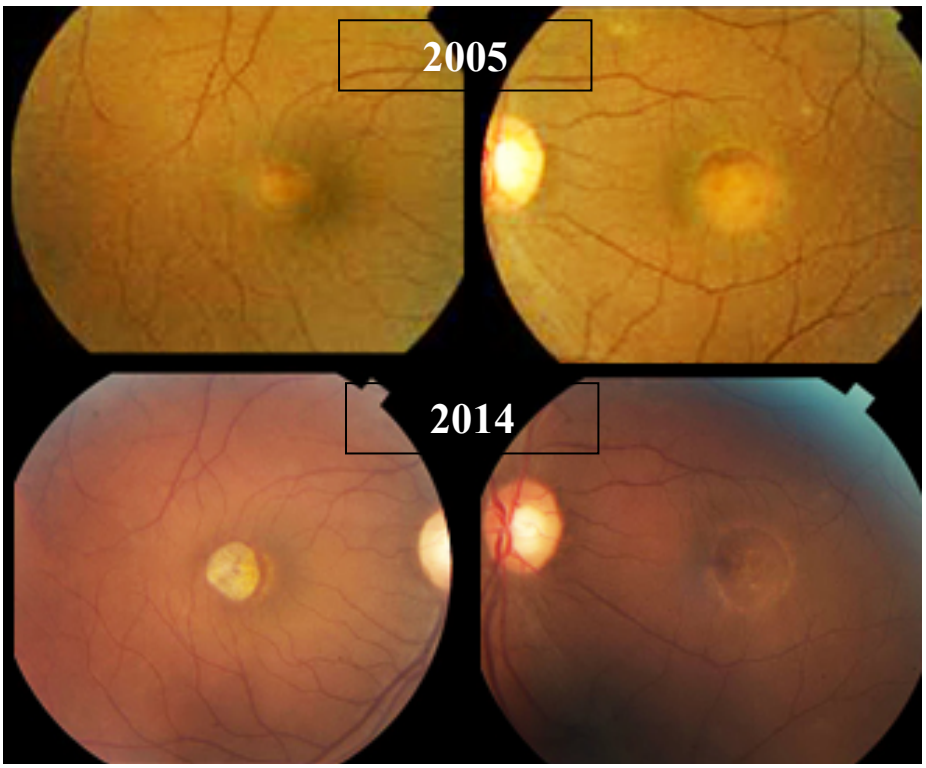
In the earliest stages the fundus may look normal (fig. 1). Later, a yellow, round lesion appears similar to an egg yolk. This is the basis for the synonym Vitelliform Macular Diystrophy. Vitelliform comes from the Latin vitellus, or egg yolk. The yellow material accumulates in the retinal pigment epithelium and under this layer. It can liquefy and settle into what is known is a pseudo-hypopyon.[1] Later there may be scarring under the retina and central macular atrophy. Some patients have secondary growth of blood vessels into the lesion from the choroid, a vascular layer underlying the retina.

Figure 1 Normal Fundus



Legend: Appearance of the fundus in a normal subject.

Figure 2 Best Disease



Legend: Fundus photographs of the right eye (left panels) and left eye (right panels) of a 38 year old woman with Best disease. The top row was the appearance in 2005. The lesions are at different stages. Visual acuity is 20/20 in both eyes.The right eye is undergoing resorption of the vitelliform material. The left eye shows a vitelliform lesion. The bottom row shows the appearance of the lesions in 2014. Over this time the lesions have changed. The right lesion has evolved into a flat scar. The left eye shows resorption of vitelliform material. Visual acuity is 20/30 right eye and 20/50 left eye.

Patients are examined using a number of tests to document the structure and assess the function of the retina. The most important is optical coherence tomography, which shows the layers of the retina and the deposit of the material under the center.[2] The electro-oculogram measures the voltage that is generated by a layer beneath the retina called the retinal pigment epithelium. The measurement is made with lights off and then again with lights on. Normally patients develop a larger voltage in the light than the dark, and the ratio usually exceeds 150%. Patients with Best

disease have a smaller ratio than this. Occasionally a fluorescein angiogram is done in which dye is injected into the vein and pictures are taken sequentially. This may show growth of abnormal blood vessels.

At this time there is no known treatment for the disease. Injections of drugs such as bevacizumab can help when secondary choroidal new blood vessels grow. Patients with the Best disease need baseline imaging studies to use for comparison in future visits and to provide a prognosis. It is beneficial to examinef other family members to determine the inheritance pattern of the condition. Blood can be drawn and analyzed for the known gene mutations associated with breast disease. If vision drops, low vision aids are useful.

If you have any questions after reading this brochure a good source of further information is Pubmed which is sponsored by the National Library of Medicine. You can also submit a question using the Contact button on the home page of this website www.retina reference.com.

References

1. Parodi MB, Iacono P, Campa C, de Turco C, Bandello F. Fundus autofluorescence patterns in best vitelliform macular dystrophy. Am J Ophthalmol 2014;158:1086-1092.

2. Qian CX, Charran D, Strong CR, Steffens TJ, Jayasundera T, Heckenlively JR. Optical coherence tomography examination of the retanl pigment epithelium in Best vitelliform macular dystrophy. Ophthalmology 2017;124:456-463.

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