Best Vitelliform Dystrophy or Bests disease

What is it?

This is a term used to describe an eye condition that affects the retina. It was first described in 1905 and is an uncommon but very well know disease of the retina.

It is a genetic defect know as the Bestrophin Gene and thus does tend to run in families. There is now genetic testing for the gene.

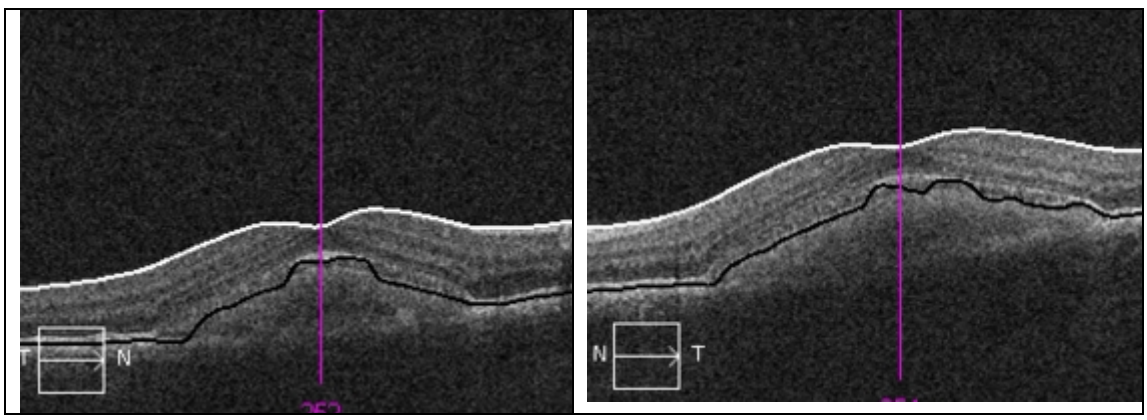
How does this affect me?

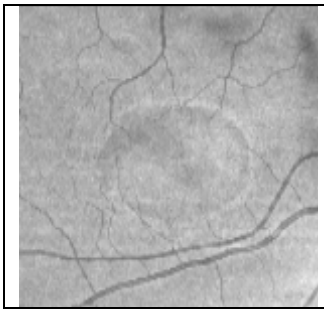
Initially up to 25 years old there is normally no affect on the vision. Later on teyhpical yellow spots develop.
Sight loss can be variable but, like other macular problems, Best's disease threatens central vision in one or both eyes.

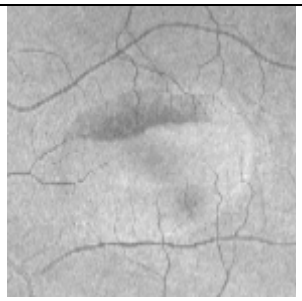
Electroretinogram tests (this is like an ECG on the eye) , which assess the electrical response of the retina when stimulated by light, can be used to identify people with the problem.

5 Stages
1 and 2 there may be little or no effect on sight.
2nd stage (usually between 10-25 years of age), typical yellow spots, sometimes accompanied by material leaking into a space by the retina, can be observed; an observation called "egg-yolk" lesion.
3rd Stage The lesion becomes absorbed. Even at this stage there may be little affect on vision.
4th stage, when the "egg-yolk" breaks up, in a process referred to as "scrambled-egg", sight will probably be affected.
5th stage is when the condition causes the most severe sight loss.

Below is an Optical Coherence Tomography (MRI like scan of the retina (back of the eye)).
Here the Best Material (Fatty yellow Lipofuscin material) can be seen in Stage 4 with vision moderately reduced.







Genetic Testing
Is available for Best 1 which makes the protein Bestrophin. This protein is found in the retinal pigment epithelial layer. More than 100 mutations of the BEST1 gene have been found causing Vitelliform Maculopathy or Best Disease. It is situated on Chromosone 13 Locus q(long arm) position 13.

Inheritance
Usually autosomal dominant, ie passed down through families. Thus if a parent has the gene then there is a 50% chance of their children having the disease. There are rare reports of an autosomal recessive gene as well.

Further information can be found on a dedicated site to Bests disease
http://www.bestdisease.net/

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