



## Vitelliform macular dystrophy

Vitelliform macular dystrophy is a genetic eye disorder that can cause progressive vision loss. This disorder affects the retina, the specialized light-sensitive tissue that lines the back of the eye. Specifically, vitelliform macular dystrophy disrupts cells in a small area near the center of the retina called the macula. The macula is responsible for sharp central vision, which is needed for detailed tasks such as reading, driving, and recognizing faces.

Vitelliform macular dystrophy causes a fatty yellow pigment (lipofuscin) to build up in cells underlying the macula. Over time, the abnormal accumulation of this substance can damage cells that are critical for clear central vision. As a result, people with this disorder often lose their central vision, and their eyesight may become blurry or distorted. Vitelliform macular dystrophy typically does not affect side (peripheral) vision or the ability to see at night.

Researchers have described two forms of vitelliform macular dystrophy with similar features. The early-onset form (known as Best disease) usually appears in childhood; the onset of symptoms and the severity of vision loss vary widely. The adult-onset form begins later, usually in mid-adulthood, and tends to cause vision loss that worsens slowly over time. The two forms of vitelliform macular dystrophy each have characteristic changes in the macula that can be detected during an eye examination.

### Frequency

Vitelliform macular dystrophy is a rare disorder; its incidence is unknown.

### Causes

Mutations in the *BEST1* and *PRPH2* genes cause vitelliform macular dystrophy. *BEST1* mutations are responsible for Best disease and for some cases of the adult-onset form of vitelliform macular dystrophy. Changes in the *PRPH2* gene can also cause the adult-onset form of vitelliform macular dystrophy; however, less than a quarter of all people with this form of the condition have mutations in the *BEST1* or *PRPH2* gene. In most cases, the cause of the adult-onset form is unknown.

The *BEST1* gene provides instructions for making a protein called bestrophin. This protein acts as a channel that controls the movement of charged chlorine atoms (chloride ions) into or out of cells in the retina. Mutations in the *BEST1* gene probably lead to the production of an abnormally shaped channel that cannot properly regulate the flow of chloride. Researchers have not determined how these malfunctioning channels are related to the buildup of lipofuscin in the macula and progressive vision loss.

The *PRPH2* gene provides instructions for making a protein called peripherin 2. This protein is essential for the normal function of light-sensing (photoreceptor) cells in the retina. Mutations in the *PRPH2* gene cause vision loss by disrupting structures in these cells that contain light-sensing pigments. It is unclear why *PRPH2* mutations affect only central vision in people with adult-onset vitelliform macular dystrophy.

## **Inheritance Pattern**

Best disease is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In most cases, an affected person has one parent with the condition.

The inheritance pattern of adult-onset vitelliform macular dystrophy is uncertain. Some studies have suggested that this disorder may be inherited in an autosomal dominant pattern. It is difficult to be sure, however, because many affected people have no history of the disorder in their family, and only a small number of affected families have been reported.

## **Other Names for This Condition**

- vitelliform dystrophy

## **Diagnosis & Management**

### Genetic Testing Information

- What is genetic testing?  
[/primer/testing/genetic-testing](#)
- Genetic Testing Registry: Macular dystrophy, vitelliform, adult-onset  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1842914/>
- Genetic Testing Registry: Vitelliform macular dystrophy  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/CN230163/>
- Genetic Testing Registry: Vitelliform macular dystrophy type 2  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0339510/>

### Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22vitelliform+macular+dystrophy%22>

### Other Diagnosis and Management Resources

- GeneReview: Best Vitelliform Macular Dystrophy  
<https://www.ncbi.nlm.nih.gov/books/NBK1167>
- MedlinePlus Encyclopedia: Macula (image)  
<https://medlineplus.gov/ency/imagepages/9608.htm>

## **Additional Information & Resources**

### Health Information from MedlinePlus

- Encyclopedia: Macula (image)  
<https://medlineplus.gov/ency/imagepages/9608.htm>
- Health Topic: Retinal Disorders  
<https://medlineplus.gov/retinaldisorders.html>

### Genetic and Rare Diseases Information Center

- Adult-onset vitelliform macular dystrophy  
<https://rarediseases.info.nih.gov/diseases/10909/adult-onset-vitelliform-macular-dystrophy>
- Best vitelliform macular dystrophy  
<https://rarediseases.info.nih.gov/diseases/182/best-vitelliform-macular-dystrophy>
- Macular dystrophy, atypical vitelliform  
<https://rarediseases.info.nih.gov/diseases/10120/macular-dystrophy-atypical-vitelliform>

### Additional NIH Resources

- National Eye Institute: Diagram of the Eye  
<https://nei.nih.gov/health/eyediagram/>

### Educational Resources

- MalaCards: best vitelliform macular dystrophy  
[https://www.malacards.org/card/best\\_vitelliform\\_macular\\_dystrophy](https://www.malacards.org/card/best_vitelliform_macular_dystrophy)
- Orphanet: Best vitelliform macular dystrophy  
[https://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=1243](https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1243)

### Patient Support and Advocacy Resources

- Foundation Fighting Blindness  
<https://www.fightingblindness.org/diseases/best-disease>
- Macular Degeneration Foundation  
<https://eyesight.org/>
- National Organization for Rare Disorders (NORD)  
<https://rarediseases.org/rare-diseases/best-vitelliform-macular-dystrophy/>
- Ophthalmic Edge  
<https://ophthalmicedge.org/patient/>
- Retina International  
<http://www.retina-international.org/for-patients/rare-conditions/what-is-best-disease/>

### Clinical Information from GeneReviews

- Best Vitelliform Macular Dystrophy  
<https://www.ncbi.nlm.nih.gov/books/NBK1167>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28vitelliform+macular+dystrophy%5BTIAB%5D%29+OR+%28vitelliform+dystrophy%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

### Catalog of Genes and Diseases from OMIM

- MACULAR DYSTROPHY, VITELLIFORM, 2  
<http://omim.org/entry/153700>
- MACULAR DYSTROPHY, VITELLIFORM, 3  
<http://omim.org/entry/608161>

### Medical Genetics Database from MedGen

- Juvenile Onset Vitelliform Macular Dystrophy  
<https://www.ncbi.nlm.nih.gov/medgen/411553>
- Vitelliform macular dystrophy  
<https://www.ncbi.nlm.nih.gov/medgen/833753>
- Vitelliform macular dystrophy type 2  
<https://www.ncbi.nlm.nih.gov/medgen/137920>

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