

# BEST VITELLIFORM MACULAR DYSTROPHY AND ADULT-ONSET FOVEOVITELLIFORM MACULAR DYSTROPHY



Macular  
Disease  
Foundation  
AUSTRALIA

Vitelliform macular dystrophy is an inherited eye disorder that can cause progressive vision loss. It causes build-up of fatty yellow pigment (lipofuscin) in the macular area which can damage the light sensitive cells responsible for clear central vision. Sight loss can be variable and can affect central vision in one or both eyes.

There are two main forms of vitelliform macular dystrophy. They are different clinical entities but have overlapping features and genetic changes. Best disease onsets in childhood and the severity of vision loss can vary widely. Adult-onset vitelliform macular dystrophy presents in your 30s, 40s and 50s, and generally causes mild vision loss that worsens slowly with time.

## How the eye works

Your eye works very much like an old-style film camera. The front of your eye, made up of the cornea, iris, pupil and lens, focuses the image onto the retina, which lines the back of your eye. The retina is a light sensitive tissue that acts like the film in a camera, capturing images and then sending them via the optic nerve to the brain, where the images are interpreted.

## What is the macula?

The macula is the name given to the area at the centre of the retina. It is approximately 5.5mm in diameter and is responsible for detailed central vision and most of your colour vision. You use your macula to read, recognise faces, drive a car, see colours clearly, and perform any other activity that requires fine vision.



# Genetics of vitelliform macular dystrophy

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Vitelliform macular dystrophy is typically inherited in an autosomal dominant pattern, which means children of an affected parent have a 50 per cent chance of receiving the gene. Family members who inherit the same genetic mutation may not experience the same changes. Other inheritance patterns are also possible.

Vitelliform macular dystrophy has been linked to a number of genes, including BEST1 and PRPH2. Researchers continue to investigate how these gene mutations cause vitelliform macular dystrophy. Further information and genetic counselling is available from specialist clinics and your GP will be able to advise you about local genetic services. People with vitelliform macular dystrophy may also wish to be entered onto the Australian Inherited Retinal Disease Register and DNA Bank, located at Sir Charles Gairdner Hospital in Perth.

## Progression of vitelliform macular dystrophy

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Vitelliform macular dystrophy causes a fatty yellow pigment (lipofuscin) to build up in the macula area, forming a lesion that has the appearance of an egg yolk. Over time, the accumulation of lipofuscin can damage the light sensitive cells that are needed for clear central vision. As a result, people with this disorder often lose their central vision, and their sight may become blurry or distorted. Vitelliform macular dystrophy typically does not affect side (peripheral) vision or the ability to see at night.



## Diagnosis

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Vitelliform macular dystrophy has characteristic changes in the macula that can be detected during an eye examination. Your eye health professional may order additional scans including optical coherence tomography (OCT) to investigate the layers of the retina and electrooculography (EOG), which assesses the electrical response of the retina when stimulated by light. Your ophthalmologist may also recommend genetic testing to confirm diagnosis, inform prognosis, and your eligibility for therapies and trials. This may have implications for your family members so you may want to discuss this with a genetics counsellor.

## Treatment

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There is currently no effective treatment for vitelliform macular dystrophy, but scientific research, both traditional and genetic, may provide useful treatments for the future.

Sometimes, new blood vessels develop under the retina. This is known as neovascularisation and is similar to what happens with the 'wet' form of age-related macular degeneration. This can lead to a rapid loss of central vision. In most cases, it can be treated very effectively with a series of injections of an anti-VEGF drug into the eye.



## Lifestyle

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There is no specific evidence regarding the effect of diet or vitamin supplements on the progression of vitelliform macular dystrophy. However, an eye-healthy diet, as recommended for age-related macular degeneration, may be beneficial. This includes eating fish two to three times a week, plenty of leafy greens and other fruit and vegetables, a handful of nuts a week, and where possible, eating low glycaemic index (GI) carbohydrates instead of high GI.

Other lifestyle recommendations include wearing sunglasses with good UV-blocking ability and not smoking.

## Managing vision loss

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If you experience vision loss as vitelliform macular dystrophy progresses, a key priority is maintaining quality of life and independence. A low vision assessment is an essential way to regain control of your situation and get started to ensure you can live well with vision loss. These assessments include tests to determine how much of your vision remains. The result of these assessments will help you gain a better understanding of your vision impairment and how to make the most of your remaining sight. It will also include valuable advice and support for your individual circumstances.

You can undergo an assessment at a low vision clinic, and sometimes in a major hospital or university. Some eye health professionals also provide low vision assessments. You may be able to have a low vision assessment in your own home. We recommend you contact MDFA so we can direct you to the appropriate low vision services for your needs.

## Using an Amsler grid

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Anti-VEGF treatment is most effective when you receive it soon after new blood vessels form. Therefore, you should monitor your vision regularly at home.

You can do this with an Amsler grid, available free from Macular Disease Foundation Australia. Check one eye at a time, covering the other eye. Any sudden changes in the appearance of the Amsler grid – such as distortion and blurry or blank patches – should be checked by your ophthalmologist promptly. It could be a sign of neovascularisation.



## Need more information?

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Learn more about macular disease at [www.mdfoundation.com.au](http://www.mdfoundation.com.au).

How's your macula? Take the quiz at [www.CheckMyMacula.com.au](http://www.CheckMyMacula.com.au).

You can also access our free, personalised support services and order information kits and Amsler grids by calling our National Helpline on **1800 111 709**.

M DFA has a free newsletter and you can sign up to receive invitations to education sessions and events in your area.

Macular Disease Foundation Australia is committed to reducing the incidence and impact of macular disease, by providing up-to-date information, advice and support.



**National Helpline**

☎ **1800 111 709**

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**W** [www.mdfoundation.com.au](http://www.mdfoundation.com.au)

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**May 2021**