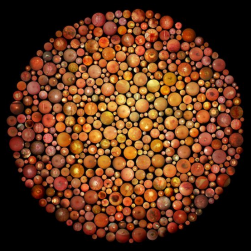
Macular Telangiectasia Type 2 (MacTel)

Patient Information Leaflet

MEDICAL RETINA



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KEY POINTS

- Macular telangiectasia type 2 (MacTel) is a rare, slowly progressive retinal disease that affects central vision in both eyes.
- It typically becomes apparent in the fifth or sixth decade of life and progresses gradually over years to decades.
- The diagnosis is often delayed or missed because the early changes are subtle and the visual letter chart may look reasonably good.
- MacTel cannot currently be reversed, but research is active and the first specific treatments are now emerging.
- A small minority of patients develop new abnormal blood vessels (neovascularisation) which is treatable with anti-VEGF injections.
- You are not alone - the international MacTel Project supports patients and runs research worldwide, including in the UK.

What Is MacTel?

The macula is the small but critical area at the centre of the retina that handles your central, detailed vision - reading, recognising faces, seeing fine detail. Within the macula sits an even smaller area called the fovea, the highest-resolution part of the eye.

In MacTel, the cells around the fovea gradually deteriorate. The earliest and most important change is the loss of specialised support cells called Muller cells, followed by the loss of light-sensing photoreceptors and changes in the small blood vessels of the retina (the "telangiectasia" of the name). Although MacTel was originally classified as a blood vessel disease, current understanding is that the vascular changes are secondary to the loss of these support cells - which is why the condition is now better thought of as a neurodegenerative retinal disease rather than a vascular one. The full name "macular telangiectasia type 2" and the shorter "MacTel" are used interchangeably; for the rest of this leaflet I will use MacTel.

MacTel affects both eyes, although often asymmetrically, and the pattern of damage is confined to a well-defined oval region centred on the fovea (the "MacTel zone"). The disease is rare, affecting around 1 in 1,000 people, and the average age of diagnosis is around 57.

Symptoms

Symptoms develop slowly and are sometimes only noticed many years after the first retinal changes. Common features include:

- Difficulty reading, particularly small print or text on screens
- Letters or words appearing to drop out, blur, or be missing
- Distortion of straight lines (metamorphopsia)
- A small grey or fading patch close to fixation
- Difficulty recognising faces
- A sense that vision is "not quite right" without being able to put a finger on why

One of the characteristic features of MacTel is that visual acuity measured on a letter chart often remains surprisingly good for years, even when reading and detail vision feel meaningfully worse. This is because the small area affected is highly important for everyday vision but does not always show up on standard testing.

How Is MacTel Diagnosed?

MacTel can be subtle, and misdiagnosis is unfortunately common - early changes can be mistaken for other macular conditions, including age-related macular degeneration. Modern retinal imaging is very helpful in making the diagnosis:

- **OCT scan (optical coherence tomography)** - shows characteristic small cavities or hyporeflective spaces in the inner and outer retina at the fovea.
- **OCT-angiography (OCT-A)** - a non-invasive scan that visualises the abnormal pattern of capillaries within the MacTel zone, often showing changes earlier and more clearly than older dye-based tests.
- **Fundus autofluorescence** - shows loss of macular pigment in a typical pattern.
- **Fluorescein angiography (FFA)** - shows late staining and leakage in the temporal parafoveal area; less commonly needed now that OCT-A is available.

I will arrange the imaging that best fits your situation. The combination of clinical examination and these scans usually makes the diagnosis straightforward.

Causes and Associations

The exact cause of MacTel is not fully understood, but research over the past two decades has made meaningful progress:

- **Genetics** - MacTel runs in some families, and specific gene variants have been identified that increase the risk of developing the disease. Most people with MacTel, however, have no clear family history.
- **Metabolism** - patients with MacTel tend to have lower levels of an amino acid called serine in the blood, and higher levels of certain lipid molecules (deoxysphingolipids) that may be toxic to retinal cells. This is currently the most active area of research.
- **Associations** - some studies suggest a higher prevalence in people with type 2 diabetes, hypertension, or higher body weight, though the relationships are not fully understood.

MacTel is not caused by lifestyle factors in the way that many eye conditions are, and there is nothing you have done that has caused you to develop it.

Treatment

There is currently no widely available treatment in the UK that can reverse the changes of MacTel or restore lost vision. However, this is an actively changing area:

Slowing progression

In March 2025, the US FDA approved the first specific treatment for MacTel - an implantable device (revakinagene taroretcel, brand name Encelto) that releases ciliary neurotrophic factor inside the eye to slow loss of photoreceptors. In two phase 3 trials it significantly slowed photoreceptor loss over two years. At the time of writing it is not yet licensed or available in the UK; whether and when it will become available here will depend on MHRA and NICE review. I will keep you informed if your treatment options change.

Treating neovascularisation

A small proportion of patients with MacTel go on to develop abnormal new blood vessels beneath or within the retina (neovascular MacTel). These vessels can leak or bleed and cause more rapid vision loss. When this happens, treatment with anti-VEGF injections - the same medications used for wet AMD - is effective at controlling the new vessels. Anti-VEGF injections are not used for routine MacTel without neovascularisation, as evidence shows they do not slow the underlying disease.

Supplements and diet

Although low serine and abnormal lipid metabolism are recognised features of MacTel, there is currently no evidence that taking serine, fenofibrate, or other supplements prevents, slows, or reverses the disease. Trials are ongoing and I would not recommend starting these on your own at present.

Research and the MacTel Project

MacTel is an unusually well-studied rare disease thanks to the international MacTel Project, run by the Lowy Medical Research Institute. The Project maintains a global registry of patients, supports clinical trials, and funds laboratory research into the causes of the disease. Patients in the UK can participate in the registry through several specialist UK sites.

If you would like to learn more, the Lowy Medical Research Institute website (lmri.net) has detailed information about MacTel, current research, and patient stories. It is the single best resource for further reading.

Living With MacTel

MacTel typically progresses slowly over 10 to 20 years, and most patients retain useful vision throughout their life - peripheral vision is not affected. Practical strategies that help include:

- Good lighting for reading and close work, and the use of magnifiers where helpful
- Larger print, larger screens, and adjusted font sizes on phones and tablets
- Using accessibility features built into modern devices (text-to-speech, contrast adjustment)
- Low vision rehabilitation services, which can offer formal assessment and aids
- Continuing to drive, work, and live independently for many years - the diagnosis itself does not require any of these to stop

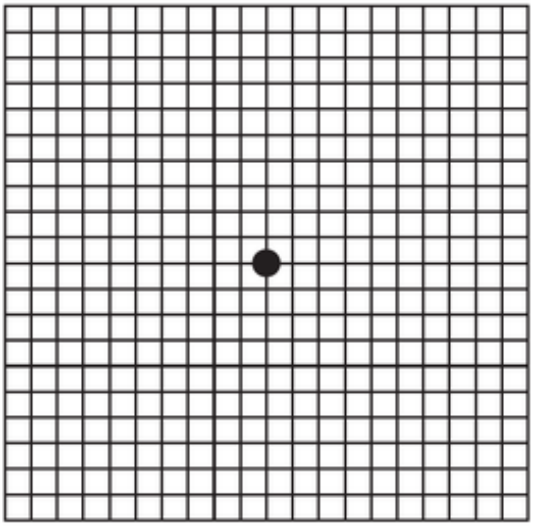
Monitoring and Follow-up

I will arrange regular review with repeat OCT scans to monitor the condition and to detect any conversion to neovascular MacTel early, when treatment is most effective. The interval depends on your stage of disease and any changes you are noticing.

If your vision worsens noticeably between appointments - particularly with new distortion, a new dark patch, or any sudden change - please contact my secretary so that you can be seen sooner.

Self-monitoring with the Amsler grid

An Amsler grid is a simple tool for detecting changes in your central vision between appointments. Wearing your reading glasses, hold the grid about 30cm from your eye and cover one eye at a time. Look directly at the central black dot. The lines should appear straight, evenly spaced, and complete with no gaps or missing areas. Repeat for the other eye. If lines appear wavy, distorted, or blurred, or if part of the grid looks missing or different from before - contact me promptly.



Standard Amsler grid (20x20 squares). Each square subtends 1 degree of visual angle at 30cm.

Support

The Macular Society in the UK provides information, support groups, counselling, and a telephone befriending service for people living with all types of macular disease, including MacTel. Their Advice and Information Service can be reached on **0300 3030 111** (Monday to Friday, 9am to 5pm) or at macularsociety.org.