Stargardt disease is a hereditary retinal disease leading to progressive degeneration of the Retinal Pigment Epithelium (RPE) and choriocapillaris, finally leading to loss of vision and progressive visual field defects.[1]

Stargardt disease causes progressive vision loss typically during childhood or adolescence. It is rare for people with Stargardt disease to become completely blind.[1]

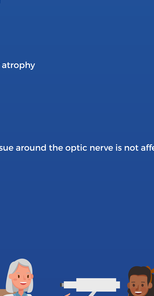
Whilst normal vision is 20/20, for most people with Stargardt disease, vision loss progresses slowly over time to 20/200 or worse.[1,2]

ONE IN 6,500

It is estimated that 1 in 6,500 people worldwide have Stargardt disease. However, this figure could be higher due to misdiagnosis or late diagnosis.[3]

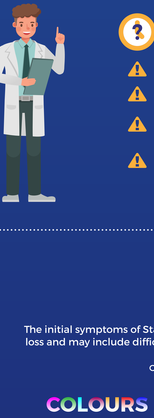
WHAT CAUSES STARGARDT DISEASE?[1,4,5,6]

- Stargardt disease is an inherited condition and is inherited in an autosomal recessive manner
- In healthy individuals the ABCA4 gene is responsible for the clearance of Vitamin A by-products of the visual cycle (lipofuscin)
- Lipofuscin is produced by retinal cells as part of the visual processing cycle
- In people with Stargardt disease the ABCA4 gene is mutated and lipofuscin is not cleared
- Excessive accumulation of lipofuscin in and around the macula has a toxic effect on the retinal pigment and vision cells, leading to central vision decrease
- With further progression, paracentral regions and the mid-periphery may also become affected leading to visual field loss over time



DIAGNOSIS AND ONSET OF SYMPTOMS[6,7,8,9]

There are three diagnostic clinical findings which, when they occur together, indicate a diagnosis of Stargardt disease:



1. The accumulation of yellowish flecks in the macula in a ring-like pattern
2. Central macular atrophy
3. The proximal tissue around the optic nerve is not affected

The most commonly used diagnostic tests for Stargardt disease are:

1. Autofluorescence imaging
2. Optical coherence tomography angiography (OCTA)
3. Electroretinogram (ERG)
4. Genetic testing

Although typically vision loss occurs between childhood and adolescence, the age of onset can vary. Some patients may experience delayed vision loss between their 30s and 60s.

Early onset Stargardt disease tends to have a more aggressive disease progression than late onset Stargardt disease.

Misdiagnosis of Stargardt disease is relatively common. This can be due to:

- Many patients, particularly children, may not be aware of their visual impairment
- A lack of clinical signs in early stage
- The broad spectrum of age of diagnosis, as many clinicians focus on looking for Stargardt disease in children and young adults
- Late presenters have very similar symptoms and signs as in patients with Geographic Atrophy (dry age-related macular degeneration)

SYMPTOMS[1,9]

Patient impact

The initial symptoms of Stargardt disease typically begin with central or pericentral vision loss and may include difficulty with dark adaptation as the disease progresses in severity.

Other symptoms of Stargardt disease include:

COLOURS

Fading colours.

BLURRY

Unclear or blurry vision

Increased sensitivity to bright light

There is a major impact on daily life and quality of life as patients lose their ability to:

CARRY OUT EVERYDAY TASKS

COMPLETE SCHOOL/ UNIVERSITY WORK

DRIVE

LEAD AN ACTIVE SOCIAL LIFE

READ

RECOGNIZE FACES

TREATMENT[1,2,10,11]

There is a high unmet medical need as there are currently no treatments available for Stargardt disease

There is currently ongoing research and development into:

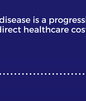
- The natural history of Stargardt disease to better understand the risk and rate of progression in different patients
- Modification of the visual cycle to reduce or eliminate lipofuscin
- Gene therapy which involves repairing or replacing the defective ABCA4 gene
- Stem cell therapy which could be used to repair RPE (Retinal Pigment Epithelium) cells

Current management strategies for symptomatic patients include disease monitoring, counselling, low vision rehabilitation, limiting the exposure to light.

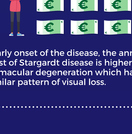
New treatments will aim to slow the progression of the disease and maintain acuity for longer.

The ultimate treatment goal is a curative treatment correcting the underlying genetic disorder.

COST[12]



As Stargardt disease is a progressive disease, there is a significant direct healthcare cost associated with it.



Due to the early onset of the disease, the annual per-patient cost of Stargardt disease is higher than age-related macular degeneration which has a similar pattern of visual loss.

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