

## Stargardt Disease

**This most prevalent inherited macular degeneration affects approximately 1 in 10,000 people. Disease onset is in childhood or the second decade of life. In the majority of patients it leads to legal blindness.**



Picture 1: The arrow indicates typical macular changes in the retina of a patient with Stargardt disease<sup>1</sup>



Picture 3: Impaired vision of a patient with Stargardt disease

Stargardt disease is the leading inherited cause of vision impairment and vision loss. It is currently an incurable condition.

Stargardt patients most commonly suffer from bilateral central visual loss, photophobia, color vision abnormalities, central scotomas and slow dark adaptation. The vision impairment is rapidly progressive and occurs most often between childhood and adolescence.

Stargardt disease was first described by Karl Stargardt, a German ophthalmologist, during his time at Strasbourg's university eye hospital in 1909. He observed a loss of central vision in a series of patients who all had similar lesions in the macula, a yellowish area of the retina near its center which hosts the largest density of photoreceptors and is the region of keenest vision. All patients had progressive vision loss over several years; his youngest patient was only 12 years old. While Stargardt could not clearly confirm the hereditary nature of the disorder, he described a clear family pattern.

Stargardt disease is associated with mutation in several genes. The most prevalent form follows a recessive pattern, i.e. one out of four children of parents who both carry one copy of the defective gene will develop clinical symptoms. There is also a rarer dominant form of the disease. The genetic defect affects a transport mechanism in the cell membrane. Subsequently, cells accumulate toxic retinoid by-products and eventually die.

Researchers and clinicians at the Institute of Molecular and Clinical Ophthalmology Basel have chosen Stargardt disease as a priority focus area and aim to develop a gene therapy for this condition.

<sup>1</sup> Picture sourced from: Leng T. (2015): Stem Cell Treatments: What's Happening in 2015. Review of Ophthalmology (link: <https://www.reviewofophthalmology.com/article/stem-cell-treatments-whats-happening-in-2015>)