STARGARDT DISEASE: A CASE REPORT ON A UNIQUE ADULT-ONSET PRESENTATION AND THE IMPERATIVE OF METICULOUS MONITORING

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ABSTRACT

Blurry vision in the pediatric population presents diagnostic hurdles, demanding meticulous history-taking to pinpoint causes, especially in children. Stargardt disease, the primary inherited retinal disorder in this demographic, causes progressive central vision loss, initially presenting as blurry or distorted vision and affecting near vision. Here, we present an atypical case of a patient diagnosed with Stargardt in adulthood and emphasize the importance of close follow-up with an ophthalmologist and primary care physician to ensure adequate management of this disease.

The patient is a 49-year-old female who presents for an 8-month follow up with her primary care physician. She was diagnosed with Stargardt disease about 10-15 years prior after having vision problems after refractive surgery. She initially complained about poor night vision, trouble with bright lights, reading, and overall trouble seeing. Her uncorrected visual acuity showed 20/60-1 OD and 20/70-2 OS. She showed full extraocular movements bilaterally and full perception of visual fields. Fundus exam showed bilateral blunt parafoveal depigmentation. SD-OCT showed abnormal central retinal architecture with foveal thinning. A genetic panel for the ABCA-4 gene confirmed the diagnosis. She has been maintained on Lutein 20mg daily. On her 8-month follow up with a new ophthalmologist, the patient continues to have difficulty with glare while driving at night, reading small fonts, and obscure/script/cursive handwriting. She admits to only a fair compliance with her medication, using it “off/on” complaining of a possible interference with her sleep. Because the existing research on Stargardt is limited, it is critical to maintain regular follow ups with a primary care physician and ophthalmologist to track the progression of this disease as closely as possible, with particular emphasis on medication adherence and possible adjustments when needed.

INTRODUCTION

Stargardt disease, also referred to as juvenile macular degeneration, remains a highly complex retinal disease that we are only just beginning to comprehend. Although extremely rare, it is the most common inherited single-gene retinal disease with a prevalence between 1 in 8,000 to 1 in 10,000. It is characterized by macular degeneration with an age of onset that varies, but typically manifests in childhood, adolescence or adulthood, eventually resulting in progressive loss of vision. Of note, there is no gender bias with this disease either.

Stargardt disease usually has an autosomal recessive inheritance caused by mutations in the ABCA4 gene. Rarely it has an autosomal dominant inheritance due to defects with ELOVL4 or PROM1 genes. These genes provide instructions for making proteins that are found in the light sensing photoreceptor cells in the retina. These proteins are responsible for clearing of a fatty substance called lipofuscin from the retina. When these proteins become non-functional, as in the case of Stargardt, the lipofuscin begins to accumulate, eventually leading to atrophy of a specific area of the retina known as the macula. How extensive the depositions are determine the staging. Stage 1 is defined as flecks of lipofuscin confined to the macula. Stage 2 is characterized by extension of flecks just beyond the macula. Stage 3 is characterized by resorbed flecks. The final stage, stage 4, is characterized by extensive atrophy of the retinal pigment epithelium and choroid and involves both the macula and peripheral retina. This may manifest with symptoms of gradual central vision loss. A common chief complaint may be blurry or wavy vision. Patients may complain of difficulty with reading, driving, or facial recognition. They may even experience scotomas and decreased color perception as the disease progresses. Usually, peripheral vision is spared due to most of the damage centering around the macula.

Diagnosis is mainly made through clinical exam including visual acuity and fundus exam. Imaging such as optical coherence tomography (OCT) and electroretinography (ERG) will show characteristic findings that may aid in diagnosis. Testing for ABCA4 gene is required to make the diagnosis. Management currently is only supportive, and no definitive treatment currently exists, however there are experimental therapies currently being studied.

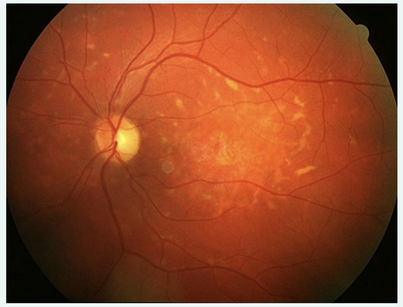
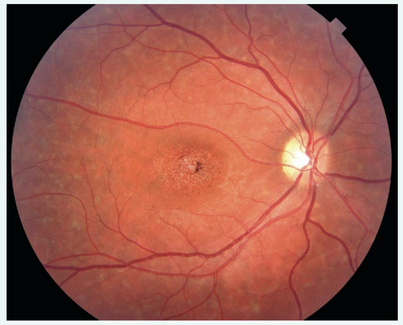


Figure 1. Stargardt Disease on fundoscopic exam
On fundoscopic exam the fovea will become atrophic, which will be visualized as surrounding white-yellow pisciform(fish-shaped) flecks. These flecks manifest at the retinal pigmented epithelium level and resemble drusen visually, but with a more angular shape compared to the typically round drusen. The atrophy appears initially followed by the flecks.

Figure 2. Stargardt Disease on fundoscopic exam
On examination of the fundus, an advanced stage of macular atrophy is evident, accompanied by notable yellow-white flecks and the ”beaten-bronze” appearance of the fovea characteristic of Stargardt. Notably, the peripapillary retina exhibits no visible signs of atrophy.



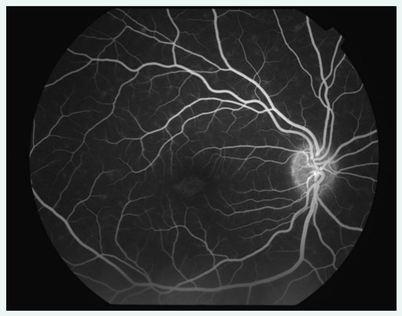


Figure 3. Stargardt disease on Fluorescein Angiography
An accumulation of abnormal lipofuscin/A2E in the retinal pigmental epithelium causes the RPE to block choroidal fluorescence, giving it the characteristic “dark choroid” appearance.

CASE DESCRIPTION

A 49-year-old female presented for her 8-month follow-up with her primary care physician. She had a history of Stargardt Disease diagnosed around 10-15 years ago following central vision issues post-laser-assisted in situ keratomileusis. Initially, she reported poor night vision, sensitivity to bright lights, difficulty reading small fonts and cursive, and no ocular pain or diplopia. Over time, her symptoms progressed to include facial recognition difficulties, worsened visual acuity in varying light conditions, trouble with overlapping colors but intact single-color perception, and no other ocular complaints.

Her medical history included breast cancer treated with Tamoxifen, human papillomavirus infection, and irritable bowel syndrome. Family history revealed glaucoma in her father but no other ocular conditions. During the examination, her uncorrected visual acuity was 20/50 +1 OD and 20/70 OS with no improvement using a pinhole. Intraocular pressures were normal, pupils were reactive, visual fields were intact, and extraocular movements were normal. Slit lamp examination showed no abnormalities, but fundus examination revealed parafoveal depigmentation and optical coherence tomography showed central retinal thinning bilaterally.

Further evaluation included an electroretinogram test, which remained normal, and a genetic analysis confirmed a biallelic mutation of the ABCA4 gene, characteristic of Stargardt Disease. The patient's ongoing management includes regular follow-ups for monitoring and potential intervention as needed to manage disease progression and optimize visual function. She is currently prescribed Lutein 20 mg orally once daily for management.

On follow up with her ophthalmologist, the patient admits to continual progression of presenting symptoms. She admits to infrequently adhering to medication usage due to possible interference with sleep. Follow-up every two years is recommended to monitor progression.

DISCUSSION

- Existing research on Stargardt is significantly limited.
- Regular follow-ups with the primary care physician are crucial for monitoring progression.
- Emphasize medication adherence as a key priority.
- Adjust medication as necessary, particularly when patients report adverse effects that impact their quality of life, such as sleep disturbances.

REFERENCES

American Academy of Ophthalmology OKAPs Review

EyeWiki

Macular Society