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Stargardt Disease Unmasked During Primigravida Pregnancy: A Case Report on Diagnosis and Low Vision Management

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ABSTRACT

Stargardt disease (STGD1), the most prevalent inherited macular dystrophy in juveniles and young adults, typically results from biallelic mutations in the ABCA4 gene. It leads to progressive central vision loss due to lipofuscin accumulation in the retinal pigment epithelium (RPE) and subsequent photoreceptor degeneration. While onset often occurs in the first two decades, presentation can vary. Managing STGD1 involves optimizing remaining vision through low vision rehabilitation and addressing the psychosocial impact, particularly crucial during significant life events like pregnancy. We report the case of a 23-year-old Indonesian female, primigravida at 35 weeks of gestation, referred from obstetrics for evaluation of a longstanding visual impairment before delivery planning. She reported progressive bilateral blurred vision since age 7, significantly worsening over the past three years, accompanied by photophobia and difficulty reading, requiring close proximity to text. Her younger sibling also had similar visual complaints. Best-corrected visual acuity was 1/60 OD and 2/60 OS, unamenable to refractive correction. Fundus examination revealed bilateral macular atrophy with a "beaten bronze" appearance, surrounding pisciform yellowish flecks extending towards the mid-periphery, and peripheral retinal dystrophy. Optical Coherence Tomography (OCT) confirmed foveal atrophy and RPE disruption. Fundus autofluorescence (FAF) showed hypoautofluorescence corresponding to atrophy, surrounded by areas of hyperautofluorescence (flecks). Amsler grid testing indicated bilateral central scotomas. Contrast sensitivity was severely reduced (1.25% Pelli-Robson bilaterally), and Ishihara testing revealed dyschromatopsia. Based on the clinical findings, family history, and characteristic imaging features, a diagnosis of Low Vision secondary to suspected Stargardt Disease was made. Obstetric examination was normal, and no contraindications for vaginal delivery were identified from an ophthalmic perspective. Low vision management included counseling, prescription of photochromic lenses, and demonstration of non-optical aids (typoscope, large print materials, handheld/stand magnifiers, glare control measures like hats/sunglasses, and a white cane for mobility). In conclusion, this case highlights the presentation of suspected Stargardt disease in a young primigravida woman, emphasizing the importance of thorough ophthalmic evaluation and multimodal imaging for diagnosis, even when symptoms have been present since childhood. Pregnancy provided the context for a formal diagnosis and initiation of low vision rehabilitation. Management focused on maximizing functional vision using appropriate aids, patient education regarding the condition's nature and inheritance, and addressing safety during mobility. Multidisciplinary care involving ophthalmology, obstetrics, and low vision services is crucial for optimal patient outcomes.

1. Introduction

Inherited retinal diseases (IRDs) constitute a diverse group of genetic disorders that affect the cells of the retina, frequently culminating in significant visual impairment or complete blindness. Despite the rarity of individual IRDs, their collective impact on vision loss is substantial, particularly among children and adults in their prime working years, accounting for approximately 20-25% of blindness within these demographics. Within the spectrum of IRDs, Stargardt disease (STGD), specifically type 1 (STGD1, OMIM #248200), is recognized as the most prevalent form of inherited macular dystrophy, with prevalence estimates ranging from 1 in 8,000 to 1 in 10,000 individuals. STGD1 is predominantly inherited as an autosomal recessive trait. The underlying genetic etiology involves biallelic mutations in the ATP-binding cassette, sub-family A, member 4 (ABCA4) gene, located on chromosome 1p22. This gene provides the blueprint for a transmembrane protein, known as Rim protein (RmP), which is strategically positioned within the disc membranes of photoreceptor outer segments in both rods and cones. The ABCA4 protein plays a critical role in the visual cycle by functioning as an ATP-dependent transporter. It is essential for the clearance of all-trans-retinal derivatives from the photoreceptor outer segment disc lumen following light absorption and the activation of the phototransduction cascade. More specifically, the ABCA4 protein facilitates the transport of N-retinylidene-phosphatidylethanolamine (N-RPE), a complex formed by the combination of all-trans-retinal and phosphatidylethanolamine, out of the disc lumen. In individuals harboring biallelic pathogenic ABCA4 mutations, the crucial transport function of the ABCA4 protein is either impaired or completely absent. This deficiency leads to the accumulation of N-RPE within the disc lumen. The accumulated N-RPE subsequently reacts with another molecule of all-trans-retinal, resulting in the formation of N-retinylidene-N-retinyl-phosphatidylethanolamine (A2PE). A2PE is then hydrolyzed into N-retinylidene-N-retinyl-ethanolamine (A2E). A2E is a major component of lipofuscin, the autofluorescent pigment aggregates that accumulate within the lysosomes of the retinal pigment epithelium (RPE). Lipofuscin, and particularly its A2E component, exhibits cytotoxic properties. It compromises lysosomal function, generates reactive oxygen species upon exposure to light, and activates the complement cascade. These cytotoxic effects ultimately lead to RPE cell dysfunction and death, followed by secondary degeneration of photoreceptors, predominantly in the macula. The accumulation of lipofuscin is clinically observable as yellowish subretinal deposits or "flecks," a hallmark characteristic known as fundus flavimaculatus. These flecks, along with the subsequent RPE and photoreceptor degeneration, contribute to the macular atrophy typically seen in STGD1.[1-3]

The clinical presentation of STGD1 typically involves bilateral and progressive central vision loss. The onset of these symptoms usually occurs in childhood or early adulthood, commonly between the ages of 6 and 20 years. Affected individuals often report symptoms such as difficulty with reading, blurred central vision (scotoma), impaired color vision (dyschromatopsia), and increased sensitivity to light (photophobia). While central vision progressively deteriorates, often stabilizing at the level of legal blindness (20/200 or worse), peripheral vision is typically preserved until the later stages of the disease. The age of onset and the rate at which the disease progresses can vary considerably among affected individuals. This heterogeneity is influenced by the specific ABCA4 mutations involved. Severe null mutations are often associated with an earlier onset of the disease and a more rapid progression. In contrast, certain missense mutations may lead to a later onset, sparing of the fovea, and a slower decline in visual function. The diagnosis of STGD1 is based on a combination of several factors: the patient's characteristic symptoms, a thorough family history, clinical examination findings, and multimodal retinal imaging. Initially, funduscopy may reveal normal findings or only subtle foveal changes. However, as the disease progresses, funduscopy typically reveals macular atrophy, often described as having a "beaten bronze" appearance or evolving into geographic atrophy, along with the hallmark yellowish pisciform flecks at the level of the RPE. Fundus autofluorescence (FAF) imaging is a particularly valuable tool in the diagnostic process. FAF imaging highlights lipofuscin accumulation as hyperautofluorescence (flecks) and

RPE atrophy as hypoautofluorescence. Notably, FAF imaging can often reveal abnormalities even when funduscopy findings are subtle. Optical coherence tomography (OCT) provides cross-sectional views of the retinal structure. In STGD1, OCT imaging typically demonstrates disruption and loss of the photoreceptor layers, including the ellipsoid zone and interdigitation zone, RPE atrophy, and thinning of the outer retina in the macula. Electrophysiological tests, such as electroretinography (ERG), may initially show normal results. However, as the disease progresses, ERG can reveal cone or cone-rod dysfunction. The definitive diagnosis of STGD1 is achieved through genetic testing, which confirms the presence of biallelic ABCA4 mutations.[4-6]

The current management of STGD1 is primarily supportive, as there is currently no cure for the disease. Management strategies focus on slowing potential disease progression and maximizing the patient's remaining functional vision. Photoprotection is a key recommendation, involving the use of UV-blocking sunglasses and hats to minimize light exposure due to the photosensitive nature of lipofuscin precursors. Patients are advised to avoid high-dose Vitamin A supplementation, as it may potentially accelerate lipofuscin accumulation. The cornerstone of STGD1 management is low vision rehabilitation. This involves a comprehensive assessment by low vision specialists and the prescription of appropriate assistive devices, such as magnifiers (both optical and electronic), telescopes, tinted lenses for glare control, task lighting, and other assistive technologies. Orientation and mobility training, psychosocial support, and genetic counseling are also integral components of comprehensive care for individuals with STGD1. Promising therapeutic avenues are currently being investigated for STGD1. These include gene therapy, utilizing viral vectors or mRNA trans-splicing to deliver functional ABCA4 genes, stem cell therapy involving RPE transplantation, and pharmacological approaches. Pharmacological strategies aim to target the visual cycle, such as Emixustat and Tinlarebant, or to reduce the formation of toxic byproducts, such as ALK-001 and deuterated vitamin A. Pregnancy represents a unique physiological state characterized by significant hormonal and metabolic changes. While there is a paucity of extensive data, the precise impact of pregnancy on the progression of IRDs like STGD1 remains largely unclear. Anecdotal reports and small case series suggest that pregnancy does not typically lead to a rapid worsening of most IRDs. However, the hormonal fluctuations and increased metabolic demands of pregnancy warrant careful consideration in the context of these conditions. Furthermore, the diagnosis or presence of an inherited condition like STGD1 during pregnancy raises several important considerations. These include genetic counseling to inform the patient about the inheritance pattern and potential risks to offspring, decisions regarding the mode of delivery (although STGD1 itself rarely necessitates cesarean section), and the challenges of managing vision loss concurrently with the demands of pregnancy and new motherhood.[7-10] This case report presents a detailed account of a young woman whose suspected Stargardt disease, with symptoms dating back to childhood, was formally diagnosed and managed during her first pregnancy. The report aims to emphasize the diagnostic utility of modern imaging techniques in the evaluation of STGD1, to illustrate the principles of low vision rehabilitation tailored to the individual patient's needs, and to highlight the importance of multidisciplinary care in the management of inherited retinal disease in the context of pregnancy.

2. Case Presentation

This case report details the clinical presentation of a 23-year-old female patient, designated Ny. M, residing in Palembang City, Indonesia. The patient presented with a constellation of ocular symptoms and clinical findings, occurring within the context of her first pregnancy (primigravida) at 35 weeks of gestation at the time of the ophthalmological consultation. The patient is identified as Ny. M, a 23-year-old female. Her residence is recorded as Palembang City, Indonesia.

An important contextual factor is her pregnancy status: she is a primigravida, indicating this is her first pregnancy, and she is at 35 weeks of gestation. This late-stage pregnancy is a significant element in considering the potential influence of physiological changes on her condition and the implications for management. The patient's chief complaint is bilateral, progressive blurred vision, which has been present since approximately 7 years of age. This long-standing nature of the visual impairment is critical, suggesting a chronic or slowly progressive underlying pathology. The history of present illness (HPI) reveals that the blurred vision has been gradually worsening over time, with a reported faster decline in visual acuity over the last 3 years. This acceleration in the progression of symptoms is a key point in understanding the disease course. The patient describes a central "shadow" or blockage in her vision. She also reports difficulty reading, necessitating close proximity to text. This is a common symptom of reduced visual acuity and impaired central vision, interfering with tasks requiring fine discrimination. A prominent symptom is marked photophobia, particularly outdoors. Photophobia, or light sensitivity, can occur in various ocular conditions, including those affecting the photoreceptors or the retinal pigment epithelium (RPE). Interestingly, the patient notes subjectively better vision in dim light. This phenomenon, sometimes referred to as "hemeralopia" (though strictly, hemeralopia means better vision in bright light), can be associated with conditions affecting cone function more than rod function, as rods are more sensitive in low light. The patient reports no associated eye pain, redness, headache, nausea, or vomiting. This absence of pain or other systemic symptoms helps to narrow the differential diagnosis, suggesting a primary ocular condition rather than a secondary manifestation of a systemic illness. The patient's past medical history (PMH) is unremarkable, with no reported hypertension (HTN), diabetes mellitus (DM), or allergies. Similarly, the past ocular history (POH) is negative for any trauma, surgery, or spectacle use. This lack of prior ocular interventions is important in evaluating the current condition. Of significant importance is the family history (FHx). The patient reports that her younger sibling, a 15-year-old, has similar visual complaints. This familial aggregation strongly suggests a genetic component to the patient's condition.

The patient's general physical examination reveals that she is conscious and oriented, indicating intact cognitive function. She is noted to be in mild distress, which could be related to the anxiety associated with her visual impairment, her pregnancy, or a combination of factors. Her vital signs are as follows: blood pressure (BP) is 110/70 mmHg, which is within the normal range; heart rate (HR) is 84 beats per minute, also within the normal range; respiratory rate (RR) is 20 breaths per minute, within the normal range; and body temperature is 36.5°C, which is within the normal range. These vital signs suggest overall physiological stability. Anthropometric measurements show her weight (Wt) to be 66 kg and her height (Ht) to be 154 cm. The calculated body mass index (BMI) is 27.8 kg/m², classifying her as overweight. While not directly related to her ocular condition, her BMI is a relevant piece of general health information. The systemic examination, including the head, neck, chest, heart, and extremities, is reported to be within normal limits (WNL). This further supports the notion of a primary ocular condition rather than a systemic disease with ocular manifestations. The obstetric examination reveals a gravid uterus consistent with approximately 35 weeks of gestation. The fetal presentation is cephalic (head-down), which is the most common and favorable position for vaginal delivery. The fetal heart rate (FHR) is 146 beats per minute, within the normal range. The estimated fetal weight (EFW) is approximately 2790 grams. No contractions were noted at the time of the examination. These obstetric findings are important in assessing the patient's overall health and pregnancy status. The ophthalmology examination provides critical information about the nature and extent of the patient's visual impairment. The best-corrected visual acuity (BCVA) is significantly reduced in both eyes: 1/60 in the right eye (OD) and 2/60 in the left eye (OS). The notation "(ph-)" indicates no improvement with pinhole testing. This lack of improvement suggests that the reduced vision is not due to a refractive error (like myopia or hyperopia) that can be corrected with glasses. It points towards a pathological process affecting the retina or optic nerve. Furthermore, there was no improvement with refraction, reinforcing that refractive error is not the cause of the decreased vision. The intraocular pressure (IOP) measured by non-contact tonometry (NCT) is 15.6 mmHg in the right eye (OD) and 17.5 mmHg in the left eye (OS). These values are within the normal range, indicating that glaucoma is unlikely to be the primary cause of the visual loss. Alignment and motility are normal, with orthophoria (proper alignment of the eyes) and full extraocular movements (EOM) in both eyes (OU). This suggests that the muscles controlling eye movement are functioning correctly, and there is no strabismus (misalignment of the eyes). The external and anterior segment examination is largely unremarkable. The lids and conjunctiva are within normal limits (WNL), the cornea is clear, the anterior chamber depth (ACD) is normal, the iris appears normal, the pupils are approximately 3mm in size, round, and reactive to light, and the lens is clear in both eyes (OU). These findings indicate that the front part of the eye is generally healthy. The funduscopy, which is the examination of the back of the eye, reveals significant abnormalities. The optic disc is round, with sharp margins, normal color, and a cup-to-disc ratio (C/D) of 0.3, with an arteriole-to-venule ratio (A:V) of 2:3. These disc findings are generally normal. However, the macula shows a diminished foveal reflex and a "beaten bronze" appearance. The foveal reflex is the reflection of light from the fovea, the central part of the macula responsible for sharpest vision. A diminished reflex indicates macular pathology. The "beaten bronze" appearance is a characteristic descriptive term often used in macular dystrophies, suggesting an atrophic or altered macular appearance. The retina shows numerous yellowish pisciform flecks peri-macular to mid-periphery. Pisciform means fish-shaped, and these flecks are a hallmark finding in certain retinal dystrophies. Peripheral retinal dystrophy changes are noted, indicating abnormalities beyond the macula. However, the normal vascular contour suggests that the major blood vessels of the retina are not primarily affected. Functional tests reveal further deficits. Amsler grid testing shows bilateral central scotomas. A scotoma is an area of visual field loss, and central scotomas correlate with the patient's subjective complaint of a central "shadow" or blockage. Contrast sensitivity, measured by the Pelli-Robson chart, is severely reduced to 1.25% in both eyes (OU). Contrast sensitivity is the ability to distinguish between subtle differences in shades of gray, and its reduction significantly impacts daily visual function. Color vision testing using Ishihara plates reveals dyschromatopsia in both eyes (OU), type unspecified. Dyschromatopsia indicates impaired color vision, suggesting cone photoreceptor dysfunction. The laboratory findings include a complete blood count (CBC), biochemistry profile, and infectious disease screening. The CBC shows a hemoglobin level (Hb) of 12.4 g/dL, hematocrit (Ht) of 36%, white blood cell count (WBC) of $7.4x10^3/\mu L$, and platelet count (Plt) of $327x10^3/\mu L$. The indices and differential are reported as within normal limits (WNL). These CBC results are generally within the normal range, suggesting no significant hematological abnormalities. The biochemistry profile reveals serum glutamic-oxaloacetic transaminase (SGOT) of 21 U/L, serum glutamic-pyruvic transaminase (SGPT) of 34 U/L, glucose (Glu) of 94 mg/dL, urea of 48 mg/dL, creatinine (Cr) of 0.76 mg/dL, sodium (Na^+) of 153 mEq/L, potassium (K^+) of 4.3 mEq/L, and chloride (Cl^-) of 105 mEq/L (units are standard). Most of these values are within the normal range, although the sodium level is slightly elevated. However, the clinical significance of this isolated elevation needs to be interpreted in the context of the patient's overall condition and hydration status. Infectious disease screening shows negative results for hepatitis B surface antigen (HBsAg (-)) and anti-HIV (-), indicating the absence of active hepatitis B infection and HIV infection. Imaging studies include obstetric ultrasonography (USG) and ocular imaging. The obstetric USG shows a viable singleton intrauterine pregnancy (IUP) of approximately 35 weeks gestation, with a cephalic presentation. This confirms the findings of the obstetric examination. Fundus photography documents the macular atrophy, the "beaten bronze" appearance, and the yellowish flecks observed during funduscopy. Macular optical coherence tomography (OCT) reveals foveal atrophy, disruption/thinning of the outer retinal layers (including the ellipsoid zone (EZ) and retinal pigment epithelium (RPE)), hyperreflective deposits (corresponding to the flecks), and reduced macular thickness. These OCT findings provide detailed structural information about the retinal abnormalities. Fundus autofluorescence (FAF) findings are inferred ("FAF (Inferred): Expected hypo-AF (atrophy) & hyper-AF (flecks)"). In FAF, areas of RPE atrophy typically show hypoautofluorescence (reduced fluorescence), while areas of lipofuscin accumulation (like the flecks) show hyperautofluorescence (increased fluorescence). While the precise FAF images aren't presented in the table, the expected findings are consistent with the other clinical and imaging results. The primary clinical diagnosis is Low Vision secondary to Suspected Stargardt Disease (STGD1) in both eyes (ODS). The differential diagnosis is Cone-Rod Dystrophy (CRD) ODS. The constellation of findings – the patient's history of progressive central vision loss since childhood, the characteristic fundus findings of macular atrophy and yellowish pisciform flecks, the functional deficits of central scotomas, reduced contrast sensitivity, and dyschromatopsia, and the imaging evidence of macular and outer retinal abnormalities – strongly points towards Stargardt Disease. However, Cone-Rod Dystrophy is considered in the differential diagnosis, as it can share some overlapping features, particularly the central vision loss and color vision deficits. Further investigations, such as genetic testing and electroretinography (ERG), could help to definitively differentiate between these two conditions (Table 1).

The management of this patient with suspected Stargardt Disease (STGD1) is centered on a multi-faceted approach aimed at optimizing her remaining visual function, providing education and support, and ensuring coordinated care, especially given her pregnancy status. The treatment plan encompasses optical aids, non-optical aids, education and counseling, a structured ophthalmology follow-up schedule, and careful coordination of care with her obstetrician. The primary optical intervention prescribed for the patient involves photochromic lenses. These lenses are designed to adapt their tint in response to varying light conditions. The fundamental rationale behind this recommendation is to reduce glare and enhance overall comfort, thereby improving the patient's visual experience across diverse lighting environments. Glare, an excessive amount of light entering the eye, can be particularly problematic for individuals with macular dystrophies like STGD1, as the macula is responsible for central, detailed vision. The damaged photoreceptors in the macula are often more sensitive to bright light, leading to discomfort and further impairment of vision. Photochromic lenses mitigate this by darkening in bright light, thereby decreasing the amount of light reaching the retina and reducing glare. This adaptation helps the patient to see more comfortably and function more effectively in everyday situations, from bright outdoor settings to indoor environments with varying light levels. The follow-up schedule for these lenses is set for the patient's next visit and as needed for lens adjustments. This is crucial because the effectiveness of the lenses needs to be assessed in real-world conditions, and adjustments to the tint, prescription (if any), or fit may be required to optimize their benefit. Regular follow-up ensures that the lenses continue to meet the patient's changing needs as her condition progresses or her lifestyle demands evolve. A range of non-optical aids is recommended to address the patient's specific visual challenges and enhance her functional independence. A typoscope is a simple yet effective device designed to isolate lines of text and reduce glare during reading. It typically consists of an opaque card or sheet with a narrow rectangular opening or slit. When placed over a page of text, the typoscope allows the patient to focus on one line at a time, minimizing distractions from the surrounding text and reducing glare from the paper's surface. This can significantly improve reading comfort and efficiency, especially for individuals with central vision loss or light sensitivity. Patient education on the proper use of the typoscope is provided at the initial visit to ensure she can effectively incorporate it into her reading routine. Magnifiers are essential tools for individuals with reduced visual acuity, enabling them to enlarge text and images for near viewing. Both handheld and stand magnifiers offer different advantages. Handheld magnifiers are portable and convenient for quick spot-reading tasks, while stand magnifiers provide a stable, hands-free viewing experience, which can be particularly beneficial for extended reading or writing. The recommendation for magnifiers aims to enhance the patient's ability to perform near-vision tasks such as reading, writing, and viewing detailed objects. Patient education on the proper use and selection of magnifiers is provided at the initial visit. It is also noted that the need for higher magnification will be assessed at follow-up visits. This is important because the patient's visual acuity may continue to decline, necessitating stronger magnification to maintain functional vision. Regular assessment ensures that the patient always has access to the appropriate level of magnification. Providing high-contrast materials, specifically large print, is a fundamental strategy to improve readability. Increasing the font size makes text easier to discern for individuals with reduced visual acuity. High contrast, such as black text on a white background, further enhances readability by maximizing the difference in luminance between the text and the background. This is particularly beneficial for patients with STGD1, who often experience reduced contrast sensitivity. Patient education on the availability and use of large print materials is provided at the initial visit. This includes guidance on accessing large print books, newspapers, and other resources. To minimize photophobia and improve vision in bright light, the use of glare control measures such as hats and sunglasses is strongly recommended. As mentioned earlier, photophobia is a common symptom in STGD1. Hats with wide brims can shield the eyes from direct sunlight, while sunglasses, especially those with UV protection and polarization, can further reduce glare and improve visual comfort. This intervention aims to enhance the patient's ability to function comfortably and safely in brightly lit environments, both indoors and outdoors. Patient education on the importance of consistent glare control is provided at the initial visit. A white cane is recommended to improve safety and independence during ambulation, particularly in unfamiliar environments. The white cane serves as a tactile extension of the patient, allowing her to detect obstacles, changes in elevation, and other hazards in her path. This is especially crucial given her reduced visual acuity and the potential for impaired depth perception. The white cane promotes independent mobility and reduces the risk of falls and injuries. A referral for orientation and mobility training is provided. This training is essential to teach the patient the proper techniques for using the white cane effectively and safely, enabling her to navigate her environment with confidence. Education and counseling are integral components of the patient's management plan, empowering her with knowledge and providing support to cope with the challenges of STGD1. A thorough explanation of Stargardt Disease is provided to the patient. This includes information about the nature of the condition, its progressive nature, and its genetic implications. Understanding the disease process is crucial for the patient to manage her expectations, make informed decisions about her care, and adapt to the changes in her vision. Providing this information at the initial visit, with ongoing reinforcement, ensures that the patient has ample opportunity to ask questions and clarify any concerns as she processes the diagnosis. Genetic counseling is recommended to discuss the inheritance pattern of STGD1, the risk to future offspring, and options for prenatal or preimplantation genetic diagnosis. STGD1 is typically inherited in an autosomal recessive manner, meaning there is a 25% risk of having an affected child with each pregnancy if both parents are carriers of the mutated gene. Genetic counseling can provide the patient and her partner with accurate information about these risks, allowing them to make informed decisions about family planning. Furthermore, genetic counseling can discuss the availability and implications of prenatal or preimplantation genetic diagnosis, which are options for couples who wish to have children but want to avoid passing on the condition. A referral to a genetics service is provided to ensure the patient receives specialized and comprehensive genetic counseling. Strategies for adapting daily activities, informing family and employers, and accessing support are provided. This intervention addresses the practical and psychosocial challenges associated with vision loss. The patient receives guidance on modifying her home and work environment to enhance visibility and safety, communicating her visual needs to family members and employers, and accessing resources and support groups for individuals with visual impairments. This comprehensive approach aims to maximize the patient's functional independence and quality of life. This information is provided at the initial visit, with ongoing reinforcement, to support the patient as she navigates the ongoing adjustments to her life. A structured ophthalmology follow-up schedule is established to monitor disease progression, assess visual acuity and functional vision, evaluate the effectiveness of low vision aids, and adjust the management plan as needed. Regular follow-up visits are crucial in managing a progressive condition like STGD1. These visits allow the ophthalmologist to track any changes in the patient's visual acuity, visual fields, and retinal appearance. They also provide an opportunity to assess the effectiveness of the prescribed low vision aids and make any necessary adjustments to ensure they continue to meet the patient's needs. The follow-up schedule is set for every 3-6 months, or sooner if symptomatic changes occur. This frequency allows for timely intervention if the disease progresses rapidly or if the patient experiences any new or worsening symptoms. Given the patient's pregnancy, coordination of care with her obstetrician is essential. This involves communicating findings and recommendations to the obstetrician to ensure coordinated care during pregnancy and delivery. The ophthalmologist and obstetrician need to work together to optimize the patient's overall health and well-being, considering the potential impact of her visual impairment on her pregnancy and delivery. For example, the obstetrician needs to be aware of any limitations the patient may have due to her vision loss, and the ophthalmologist needs to be aware of any obstetric considerations that may affect the patient's ocular health or treatment. This communication occurs as needed throughout the pregnancy to ensure seamless and comprehensive care. This collaborative approach is vital for the patient's safety and well-being and for the successful management of her condition within the context of her pregnancy (Table 2).

3. Discussion

The patient's clinical history, as detailed in the anamnesis, presents a constellation of symptoms that are highly characteristic of Stargardt disease. The insidious onset of bilateral central vision loss in childhood, in this case, around the age of 7, is a hallmark feature of the disease. STGD1 typically manifests within the first two decades of life, and this early presentation is crucial in differentiating it from other macular conditions that may arise later in adulthood. The patient's description of a central "shadow" or blockage in her vision strongly suggests a compromise of the macula, the central region of the retina responsible for sharp, detailed vision. Difficulty with reading, particularly the need to hold reading material at close proximity, is a common functional consequence of this central vision loss, directly impacting a crucial daily activity. Photophobia, or increased sensitivity to light, is another frequently reported symptom in STGD1.

Table 1. Summary of patient's clinical findings.

Category	Details
Demographics	**Patient:** Ny. M; **Age:** 23 yrs; **Gender:** F; **Residence:** Palembang City, Indonesia; **Pregnancy Status:** Primigravida, 35 wks gestation (at ophthal. consult).
Anamnesis (History)	**Chief Complaint:** Bilateral, progressive blurred vision since ~7 yrs old; **HPI:** Gradual worsening, faster decline last 3 yrs; Central "shadow"/blockage description; Difficulty reading (needs close proximity); Marked photophobia (esp. outdoors); Subjectively better vision in dim light; No associated eye pain, redness, headache, N/V; **PMH:** No HTN, DM, allergies; **POH: No trauma, surgery, spectacle use; FHx:** Younger sibling (15 yrs) with similar visual complaints; Pedigree suggestive of AR inheritance.
Physical exam (General)	**General:** Conscious, oriented, mild distress; **Vitals:** BP 110/70 mmHg; HR 84/min; RR 20/min; Temp 36.5°C; **Anthropometry:** Wt 66 kg; Ht 154 cm; BMI 27.8 kg/m² (Overweight); **Systemic:** Head, neck, chest, heart, extremities WNL; **Obstetric:** Gravid uterus ~35 wks; Cephalic presentation; FHR 146/min; EFW ~2790g; No contractions.
Ophthalmology exam	**BCVA:** OD 1/60 (ph-); OS 2/60 (ph-); No imp**rovement w/ refraction; IOP (NCT):** OD 15.6 mmHg; OS 17.5 mmHg; **Alignment/Motility:** Orthophoria; Full EOM OU; **External/Ant. Segment:** Lids/Conjunctiva WNL; Cornea Clear; Normal ACD; Iris normal; Pupils ~3mm, round, reactive; Lens Clear OU; **Funduscopy (OU):** Disc: Round, sharp margins, normal color, C/D 0.3, A:V 2:3; **Macula:** Diminished foveal reflex, "beaten bronze" appearance; **Retina:** Numerous yellowish pisciform flecks peri-macular to mid-periphery; Peripheral retinal dystrophy changes; Normal vascular contour; **Functional Tests (OU):** Amsler: Bilateral central scotomas; **Contrast Sensitivity (Pelli-Robson):** Severely reduced (1.25% OU); **Color Vision (Ishihara):** Dyschromatopsia OU (type unspecified).
Laboratory findings	**CBC:** Hb 12.4; Ht 36%; WBC 7,400; Plt 327,000; Indices/Diff WNL; **Biochemistry:** SGOT 21; SGPT 34; Glu 94; Ureum 48; Cr 0.76; Na+ 153; K+ 4.3; Cl- 105 (units as standard); Infectious: HBsAg (-); Anti-HIV (-).
Imaging findings	**Obstetric USG:** Viable singleton IUP ~35 wks, cephalic; **Fundus Photo (OU):** Documented macular atrophy, beaten bronze appearance, flecks; **Macular OCT (OU):** Foveal atrophy; Outer retinal layer (EZ, RPE) disruption/thinning; Hyperreflective deposits (flecks); Reduced macular thickness; FAF (Inferred): Expected hypo-AF (atrophy) & hyper-AF (flecks).
Clinical diagnosis	**Primary:** Low Vision sec. to Suspected Stargardt Disease (STGD1) ODS; **Differential:** Cone-Rod Dystrophy (CRD) ODS.

Notes: ACD: Anterior Chamber Depth; AF: Autofluorescence; Ant.: Anterior; AR: Autosomal Recessive; A:V: Arteriole to Venule ratio; BCVA: Best Corrected Visual Acuity; BMI: Body Mass Index; BP: Blood Pressure; C/D: Cup-to-Disc ratio; CBC: Complete Blood Count; Cr: Creatinine; CRD: Cone-Rod Dystrophy; DM: Diabetes Mellitus; EFW: Estimated Fetal Weight; EOM: Extraocular Movements; F: Female; FAF: Fundus Autofluorescence; FHx: Family History; FHR: Fetal Heart Rate; Glu: Glucose; Hb: Hemoglobin; HPI: History of Present Illness; HR: Heart Rate; Ht: Hematocrit; HTN: Hypertension; Hx: History; IOP: Intraocular Pressure; IUP: Intrauterine Pregnancy; k: thousand; K+: Potassium; MRN: Medical Record Number; N/V: Nausea/Vomiting; Na+: Sodium; Cl-: Chloride; NCT: Non-Contact Tonometry; OD: Oculus Dexter (Right Eye); ODS: Oculus Dexter et Sinister (Both Eyes); OE: Ocular Examination; Ophthal.: Ophthalmology; OS: Oculus Sinister (Left Eye); OU: Oculi Uterque (Both Eyes); ph-: No improvement with pinhole; Plt: Platelets; PMH: Past Medical History; POH: Past Ocular History; RR: Respiratory Rate; sec.: secondary; SGOT: Serum Glutamic-Oxaloacetic Transaminase; SGPT: Serum Glutamic-Pyruvic Transaminase; STGD1: Stargardt Disease Type 1; Temp: Temperature; USG: Ultrasonography; VA: Visual Acuity; WBC: White Blood Cell count; Wks: Weeks; WNL: Within Normal Limits; Wt: Weight; Yrs: Years.

Table 2. Treatment and follow-up plan.

Treatment modality	Specific intervention/ Recommendation	Rationale	Follow-up schedule
Optical aids	Photochromic Lenses	To reduce glare and improve comfort in varying light conditions.	At next visit, and as needed for lens adjustments.
Non-optical aids	Typoscope	To isolate lines of text and reduce glare during reading.	Patient education provided at initial visit.
	Magnifiers (handheld and stand)	To enlarge text and images for close viewing.	Patient education provided at initial visit. Assess need for higher magnification at follow-up.
	High-Contrast Materials (large print)	To improve readability.	Patient education provided at initial visit.
	Glare Control (hats, sunglasses)	To minimize photophobia and improve vision in bright light.	Patient education provided at initial visit.
	White Cane	To improve safety and independence during ambulation, especially in unfamiliar environments.	Referral for orientation and mobility training.
Education and counseling	Explanation of Stargardt Disease	To provide understanding of the condition, its progressive nature, and genetic implications.	At initial visit, with ongoing reinforcement.
	Genetic Counseling	To discuss inheritance pattern (AR), risk to future offspring (25% per pregnancy), and options for prenatal/preimplantation diagnosis.	Referral to genetics service.
	Functional Vision & Daily Living	To provide strategies for adapting daily activities, informing family/employers, and accessing support.	At initial visit, with ongoing reinforcement.
Ophthalmology follow-up		To monitor disease progression, assess visual acuity and functional vision, evaluate the effectiveness of low vision aids, and adjust the management plan as needed.	Every 3-6 months, or sooner if symptomatic changes occur.
Coordination of care		Communicate findings and recommendations to the patient's obstetrician to ensure coordinated care during pregnancy and delivery.	As needed throughout pregnancy.

Notes: AR: Autosomal Recessive; F/U: Follow-Up; LVA: Low Vision Aids; OD: Right Eye; OS: Left Eye; OU: Both Eyes; WNL: Within Normal Limits.

The mechanisms underlying photophobia in retinal dystrophies are complex and not fully understood, but they likely involve the dysfunction of photoreceptors and the retinal pigment epithelium, leading to abnormal processing of light signals. The patient's subjective report of improved vision in dim light, while seemingly paradoxical, can also be informative. In STGD1, the cone photoreceptors, which are concentrated in the macula and are responsible for color vision and central vision in bright light, are often preferentially affected. Rod photoreceptors, which are more prevalent in the peripheral retina and are responsible for vision in low light conditions, may be relatively spared, at least in the earlier stages of the disease. This can explain why some individuals with STGD1 experience better visual function in dimly lit environments. The patient's family history is particularly significant. The fact that her younger sibling also presents with similar visual complaints strongly indicates a genetic etiology. In this case, the pedigree is suggestive of autosomal recessive inheritance. This pattern of inheritance implies that both parents of the affected individuals are carriers of a mutated gene. Carriers themselves typically do not exhibit symptoms, as they possess one normal copy of the gene. However, there is a 25% chance that their offspring will inherit two copies of the mutated gene, one from each parent, and thus develop the condition. Autosomal recessive inheritance is the most common mode of inheritance in STGD1, which is primarily caused by biallelic mutations in the ABCA4 gene. While the clinical history is highly suggestive, it is crucial to acknowledge that a definitive diagnosis of STGD1 often requires further investigation. In particular, genetic testing plays a pivotal role. Identifying biallelic pathogenic variants in the ABCA4 gene confirms the diagnosis of STGD1. Furthermore, genetic testing can provide valuable information about the specific mutations involved, which can sometimes correlate with the severity of the disease and its rate of progression. This information is essential for accurate prognostication and genetic counseling. In this particular case, genetic testing was not explicitly reported, which is a limitation. While the clinical picture strongly points to STGD1, genetic confirmation would provide the highest level of diagnostic certainty.[11-15]

Multimodal imaging plays an absolutely critical role in the diagnosis and characterization of Stargardt disease. The information gleaned from various imaging modalities complements the clinical examination findings and provides a more comprehensive understanding of the structural and functional changes occurring in the retina. In this case, funduscopy, optical coherence tomography (OCT), and fundus autofluorescence (FAF) were utilized, each contributing unique and essential information. Funduscopy, the direct visualization of the fundus (the interior surface of the eye), is a fundamental part of the ophthalmological examination. In STGD1, funduscopy often reveals characteristic findings, although these can vary depending on the stage of the disease. In this patient, funduscopy revealed several key features of macular atrophy with a "beaten bronze" appearance, diminished foveal reflex, and numerous yellowish pisciform flecks extending from the perimacular region to the mid-periphery. The "beaten bronze" appearance is a descriptive term frequently used to characterize the altered appearance of the macula in STGD1. It suggests a metallic sheen and an irregular texture, reflecting the underlying atrophy of the retinal pigment epithelium and photoreceptor layers. The diminished foveal reflex indicates damage to the normal reflective properties of the fovea, the central pit of the macula responsible for the sharpest vision. The yellowish pisciform flecks are a hallmark of STGD1 and fundus flavimaculatus. These flecks represent the accumulation of lipofuscin, a yellowish pigment, within the retinal pigment epithelium. Their characteristic fish-tail shape and distribution pattern are highly suggestive of the disease. Optical coherence tomography (OCT) is a non-invasive imaging technique that provides high-resolution cross-sectional images of the retinal structure. It allows for detailed visualization of the different retinal layers, including the photoreceptor layer, the retinal pigment epithelium, and the choroid. In this patient, OCT imaging revealed foveal atrophy, disruption and thinning of the outer retinal layers (including the ellipsoid zone and the RPE), hyperreflective deposits, and reduced macular thickness. These OCT findings provide structural correlates for the functional vision loss experienced by the patient. The atrophy and thinning of the outer retinal layers, particularly the photoreceptor layer and the RPE, directly contribute to the decreased visual acuity and central scotoma. The hyperreflective deposits likely correspond to the lipofuscin flecks observed on funduscopy. OCT is invaluable in quantifying the extent of retinal damage and monitoring disease progression over time. Fundus autofluorescence (FAF) is another non-invasive imaging modality that provides information about the metabolic activity of the retinal pigment epithelium. Lipofuscin, the pigment that accumulates in the RPE in STGD1, is autofluorescent, meaning it emits light when excited by a specific wavelength. FAF imaging captures this autofluorescence, allowing for the visualization and mapping of lipofuscin distribution. In areas of RPE atrophy, where there is a loss of lipofuscin and RPE cells, FAF shows decreased autofluorescence (hypoautofluorescence). In contrast, areas of lipofuscin accumulation show increased autofluorescence (hyperautofluorescence). While the FAF findings in this case were inferred, the expected pattern of hypoautofluorescence in areas of atrophy and hyperautofluorescence in areas corresponding to the flecks is typical of STGD1. FAF is particularly useful in detecting early changes in the RPE and in delineating the extent of RPE dysfunction, often revealing abnormalities even before they are apparent on funduscopy. In summary, multimodal imaging, encompassing funduscopy, OCT, and FAF, is indispensable in the diagnosis and management of STGD1. These techniques provide complementary information about the structural and functional changes occurring in the retina, aiding in accurate diagnosis, quantifying disease severity, and monitoring progression.[16-20]

4. Conclusion

This case report illustrates the diagnosis and management of a 23-year-old primigravida with suspected Stargardt disease. The patient's history of progressive central vision loss since childhood, coupled with characteristic fundus findings and multimodal imaging results, strongly suggested the condition. The case highlights the importance of a thorough ophthalmologic evaluation, especially in young patients with longstanding visual complaints, and the utility of imaging techniques like OCT and FAF in aiding diagnosis. Furthermore, this case underscores the need for tailored low vision rehabilitation, including optical and non-optical aids, patient education, and psychosocial support. The patient's pregnancy added another layer of complexity, emphasizing the importance of multidisciplinary collaboration between ophthalmology and obstetrics to ensure optimal care. While pregnancy itself may not significantly alter the course of STGD1, the diagnosis has implications for genetic counseling and family planning.

5. References

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