https://doi.org/10.21516/2072-0076-2026-19-2-177-181

Low vision assessment and management in Stargardt disease: diagnostic approach and therapeutic strategies. Case report

Rabia Faheem[1] ✉, Muhammad Saad[2], Saif Ullah[1, 2], Muqaddas Noor[1], Iffat Nasim[1]

[1] Pakistan Institute of Ophthalmology, Al-Shifa Trust Eye Hospital, Rawalpindi, 4600, Pakistan
[2] Lincoln University College, Wisma Lincoln, 4730, Petaling Jaya, Selangor Darul Ehsan, Malaysia
rabiafahim3@gmail.com

A 19-year-old female presented to the outpatient department with reduced visual acuity and recurrent headaches. Initially recorded as 15 due to an oversight, her age was clarified, and her best-corrected visual acuity was measured at 6/60P logMAR bilaterally, with a prescription of -0.5 D. Cyclorefraction revealed -1.5 D bilaterally. Fundus examination demonstrated vascular tortuosity, bull's-eye maculopathy, and peripheral pigmentary retinopathy, leading to a diagnosis of Stargardt disease, confirmed through advanced imaging. She was referred to the low vision department for a specialized assessment to address her progressive vision loss and was also evaluated by neurology for recurrent headaches. A comprehensive low vision evaluation helped determine her specific visual needs, with recommendations aimed at enhancing her daily activities and managing her visual prognosis. This case underscores the essential role of low vision assessments in juvenile macular dystrophies like Stargardt disease, where early intervention and personalized visual aids can significantly improve functional vision and quality of life.

Keywords: stargardt disease; case report; ABCA4; low vision; fundus autofluorescence; optical coherence tomography
Conflicts of interests: there is no conflict of interests.
Financial disclosure: no author has a financial or property interest in any material or method mentioned.
Acknowledgments. Authors extend their sincere thanks to the staff of hospital who supported and contributed to the completion of this report.
For citation: Faheem R., Saad M., Ullah S., Noor M., Nasim I. Low vision assessment and management in Stargardt disease: diagnostic approach and therapeutic strategies. Case report. Russian ophthalmological journal. 2026; 19 (2): 177-81. https://doi.org/10.21516/2072-0076-2026-19-2-177-181

Оценка и лечение слабовидения при болезни Штаргардта: диагностический подход и терапевтические стратегии. Клинический случай

Рабия Фахим[1] ✉, Мухаммад Саад[2], Саиф Улла[1, 2], Мукаддас Нур[1], Иффат Насим[1]

[1] Пакистанский институт офтальмологии, офтальмологическая больница Аль-Шифа, Равалпинди, 4600, Пакистан
[2] Университетский колледж Линкольна, Висма-Линкольн, 4730, Петалинг-Джая, Селангор Дарул Эхсан, Малайзия

Девушка 19 лет обратилась в клинику с жалобами на снижение остроты зрения и рецидивирующие головные боли. Острота зрения каждого глаза с коррекцией -0,5 D составила 6/60P logMAR, циклоплегическая рефракция -1,5 D. Осмотр глазного дна выявил извитость сосудов, макулопатию типа «бычий глаз» и периферическую пигментную ретинопатию, что позволило диагностировать болезнь Штаргардта, подтвержденную с помощью современных методов визуализации. Пациентка была направлена в отделение слабовидения для специализированного обследования с целью решения проблемы прогрессирующей потери зрения,

а также обследована неврологом по поводу рецидивирующих головных болей. Комплексное обследование слабовидения помогло определить ее специфические зрительные потребности, были даны рекомендации, направленные на улучшение ее повседневной активности и прогноза зрения. ***Заключение.*** *Данный случай подчеркивает важнейшую роль обследования людей с низким зрением при ювенильных макулярных дистрофиях, таких как болезнь Штаргардта, когда раннее вмешательство и индивидуально подобранные средства коррекции могут значительно улучшить функциональное зрение и качество жизни.*

Ключевые слова: болезнь Штаргардта; клинический случай; ABCA4; слабовидение; аутофлуоресценция глазного дна; оптическая когерентная томография
Конфликт интересов: отсутствует.
Прозрачность финансовой деятельности: никто из авторов не имеет финансовой заинтересованности в представленных материалах или методах.
Благодарности. Авторы выражают искреннюю благодарность сотрудникам больницы, которые оказали поддержку и внесли свой вклад в подготовку данного сообщения.
Для цитирования: Фахим Р., Саад М., Улла С., Нур М., Насим И. Оценка и лечение слабовидения при болезни Штаргардта: диагностический подход и терапевтические стратегии. Клинический случай. Российский офтальмологический журнал. 2026; 19 (2): 177-81. https://doi.org/10.21516/2072-0076-2026-19-2-177-181

By the World Health Organization (WHO) definition "a person with low vision is a one who has impairment of visual functioning even after treatment, surgery or standard refractive correction and has a visual acuity of less than 6/18 to light perception or a visual field of < 10° from the point of fixation, in the better eye but who is potentially able to use vision for the planning and execution of a task" [1]. Stargardt disease is typically inherited as an autosomal recessive condition caused by missense and null mutations in the adenosine triphosphate-binding cassette transporter alpha 4 (ABCA4) gene located on chromosome 1 [2]. While the exact incidence is unclear, the prevalence is estimated to range between 1 in 8,000 and 1 in 10,000 [3]. Funduscopically, the disease progresses through distinct stages as described by G.Fishman [4]. In Stage 1, yellowish "flecks" appear in the parafoveal area. Stage 2 is characterized by the presence of these flecks throughout the posterior pole. Stage 3 involves extensive flecking that undergoes resorption, and Stage 4 is marked by retinal pigment epithelium (RPE) atrophy [4]. Currently, there are no Food and Drug Administration (FDA)-approved treatments or cures for Stargardt's macular dystrophy (SMD). Research efforts are focused on nutritional supplementation, drug therapies [5], and gene therapy [6]. In the absence of approved treatments, management of Stargardt's disease primarily emphasizes low vision rehabilitation. This case report discusses the diagnostic process, low vision assessment, and management strategies for a young patient diagnosed with Stargardt disease, highlighting the importance of specialized low vision support and genetic counseling.

Case Report. A 19-year-old female presented to the outpatient department with complaints of reduced visual acuity, photophobia and headaches at her first visit. Her age was recorded as 15 due to an error by her uncle, who accompanied her instead of her parents. Best corrected visual acuity (BCVA) at presentation was 6/60p logMAR in both eyes, and she was using -0.5 D in her glasses. Cyclorefraction was performed, resulting in a prescription of -1.5 D bilaterally.

Informed consent was obtained from the patient at the time of assessment. Full fundus examination revealed a clear cornea, a formed and quiet anterior chamber, and a cup-to-disc ratio (CDR) of 0.2 bilaterally. Intraocular pressure (IOP) measured 12 mmHg bilaterally via Goldmann Applanation Tonometry (GAT). Additional findings included vascular tortuosity and bull's-eye maculopathy with peripheral pigmentary retinopathy in both eyes. Advanced diagnostic procedures, including fundus photography through Optos, and fundus autofluorescence (FAF), optical coherence tomography (OCT) of the macula, OCT of retinal nerve fiber layer (RNFL) were conducted (Fig. 1, A, B, Fig. 2, Fig. 3). Based on these reports, the attending doctor confirmed a diagnosis of Stargardt disease (fundus dystrophy).

The patient was subsequently referred to the low vision department and a neurologist for further assessment and prognosis. Two days after the initial checkup, a low vision assessment was conducted, where a thorough ocular and medical history was taken. She reported no history of ocular trauma or surgery and indicated no mobility issues indoors but experienced some difficulties with mobility outdoors and in unfamiliar settings. Her family history revealed that she was the only daughter of her parents, who had a consanguineous marriage. The patient reported difficulties with distance tasks, including recognizing faces, watching television, and reading bus or tempo numbers, which were exacerbated by a language barrier that necessitated her uncle's assistance during the visit.

During the low vision assessment, BCVA with her new glasses prescription (-1.5 D bilaterally) was re-evaluated and was 6/60 logMAR in both eyes as measured with the ETDRS chart at 4 meters. Subjective refraction yielded no improvement in visual acuity. A monocular clip-on telescope with 2.8x magnification was tried, slightly improving her visual acuity to 6/48 logMAR bilaterally, although stronger telescopes, such as a 3.5x, offered no additional improvement. Near vision was measured at 1.6 M using the Lighthouse Near Vision Assessment Chart. Contrast sensitivity was assessed with Mars Letter Contrast Sensitivity, showing 1.35 logMAR units bilaterally with corrective lenses. Color vision, tested with the Ishihara Color Vision Chart/Plates, was defective bilaterally.

Visual field testing using the Humphrey Field Analyzer was attempted but yielded unreliable results due to the patient's headaches, despite several attempts. Confrontation visual field testing was performed, where the patient and examiner sat face-to-face, with the patient covering one eye and the examiner covering the corresponding eye. The examiner then presented moving targets in different areas of the patient's peripheral vision, starting from the periphery and moving inward, while the patient indicated when they could see the target. This method showed the patient's visual field to be within normal limits.

The patient reported increased comfort in reduced lighting conditions and under fluorescent lamps, with significant photophobia, particularly in sunlight and car headlights. Testing with a yellow filter during daylight provided improved visual comfort and acceptance. She had no ocular preference between her eyes and denied any history of bumping into objects. The patient experienced challenges with mobility and orientation, particularly in low-light and high-glare conditions, impacting daily activities.

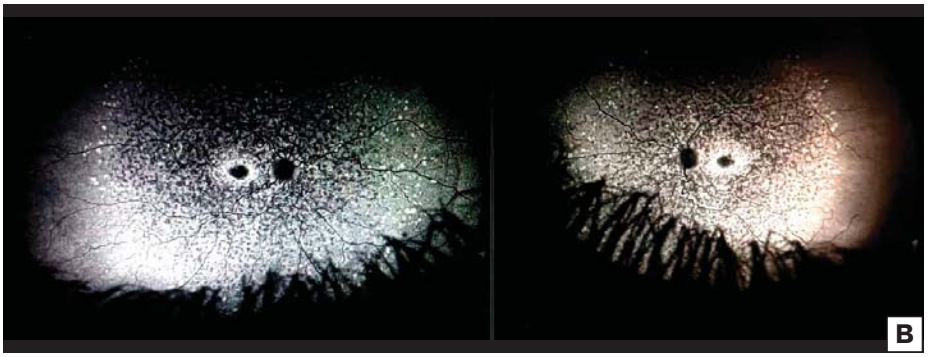

Fig. 1. A — optos wide-field color fundus photograph of both eyes, revealing characteristic yellowish flecks in the posterior pole, indicative of Stargardt disease. These deposits are a hallmark of this macular dystrophy. B — fundus autofluorescence (FAF) images of both eyes highlighting regions of hyperautofluorescence, representing areas of active lipofuscin accumulation, and hypoautofluorescence, indicating atrophic zones where the retinal pigment epithelium (RPE) has degenerated, consistent with disease progression

Рис. 1. А — широкопольная цветная фотография глазного дна обоих глаз, полученная с помощью Optos, демонстрирует характерные желтоватые пятна в заднем полюсе, указывающие на болезнь Штаргардта. Эти отложения являются характерным признаком данной макулярной дистрофии. В — изображения аутофлуоресценции глазного дна обоих глаз, выделяющие области гиперфлуоресценции, представляющие собой зоны активного накопления липофусцина, и гипофлуоресценции, указывающие на атрофические зоны, где произошла дегенерация пигментного эпителия сетчатки, что соответствует прогрессированию заболевания

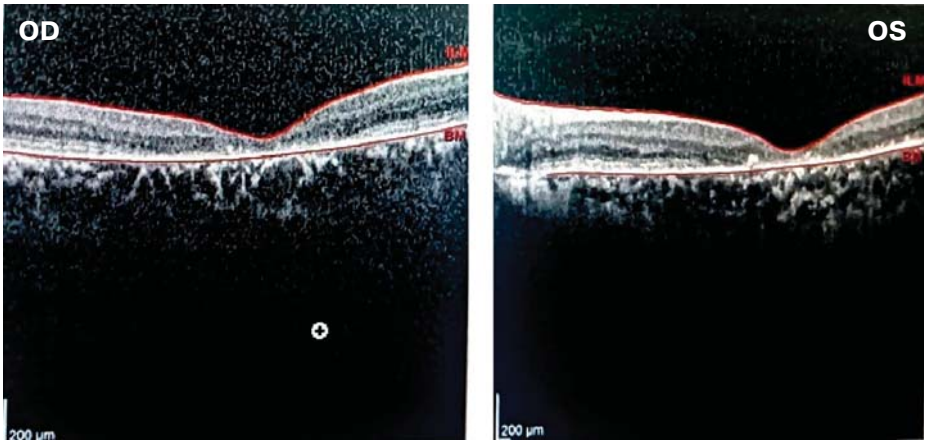

Fig. 2. Spectral-domain Optical Coherence Tomography (SD-OCT) Scans. OD — OCT scan showing retinal thinning, loss of the ellipsoid zone, and disruption of the retinal pigment epithelium (RPE) layers. OS — OCT scan demonstrating similar findings, including loss of outer retinal architecture and RPE irregularities

Рис. 2. SD-OCT сканирование. OD — истончение сетчатки, потеря эллипсоидной зоны и нарушение слоев пигментного эпителия сетчатки (ПЭС). OS — аналогичные изменения, включая потерю наружной архитектуры сетчатки и неровности ПЭС

To address this, training with a white cane was provided to enhance independence and safety.

For the time being, she was referred to a medical specialist for headache relief. The doctor recommended Tab Flexin 250 mg as a painkiller and referred her to a neurologist for further assessment.

She was advised to use low vision aids, specifically a 2.8x telescope, to assist with academic activities, while magnifiers were not necessary as her near vision was functional. Additional recommendations included maintaining a healthy diet, using sun protection (umbrella and avoiding direct sun exposure), and scheduling follow-up with a neurologist for her migraines. Vitamin A supplements were specifically avoided, as they could worsen the progression of the disease. Genetic counseling was also recommended to prevent future consanguineous marriages. A follow-up visit was planned in two months.

Patient Perspective. The patient expressed satisfaction with the personalized low vision aids, including the yellow filter and telescope, which significantly improved visual comfort and functionality.

DISCUSSION

Stargardt disease type 1 (STGD1) is caused by mutations in the ABCA4 gene, which encodes the ABCA4 protein, an ATP-binding cassette transporter located in the outer segment discs of rods and cones. This protein facilitates the transport of retinoids from photoreceptors to the retinal pigment epithelium (RPE) during the visual cycle. Dysfunctional ABCA4 leads to the accumulation of toxic retinoid byproducts, including A2E, a major component of lipofuscin. This buildup results in RPE damage, atrophy, and subsequent photoreceptor loss [7].

Other forms of Stargardt disease, such as STGD3 and STGD4, are associated with autosomal dominant mutations in the ELOVL4 and PROM1 genes, respectively. These mutations disrupt photoreceptor membrane integrity and function, leading to retinal degeneration. However, this article focuses on STGD1 [8].

M. Abalem et al. [9] studied 65 patients with ABCA4 Stargardt disease (STGD), demonstrating that greater disease extent on ultra-widefield fundus autofluorescence (UWF-FAF) correlated with more constricted peripheral visual fields, worse

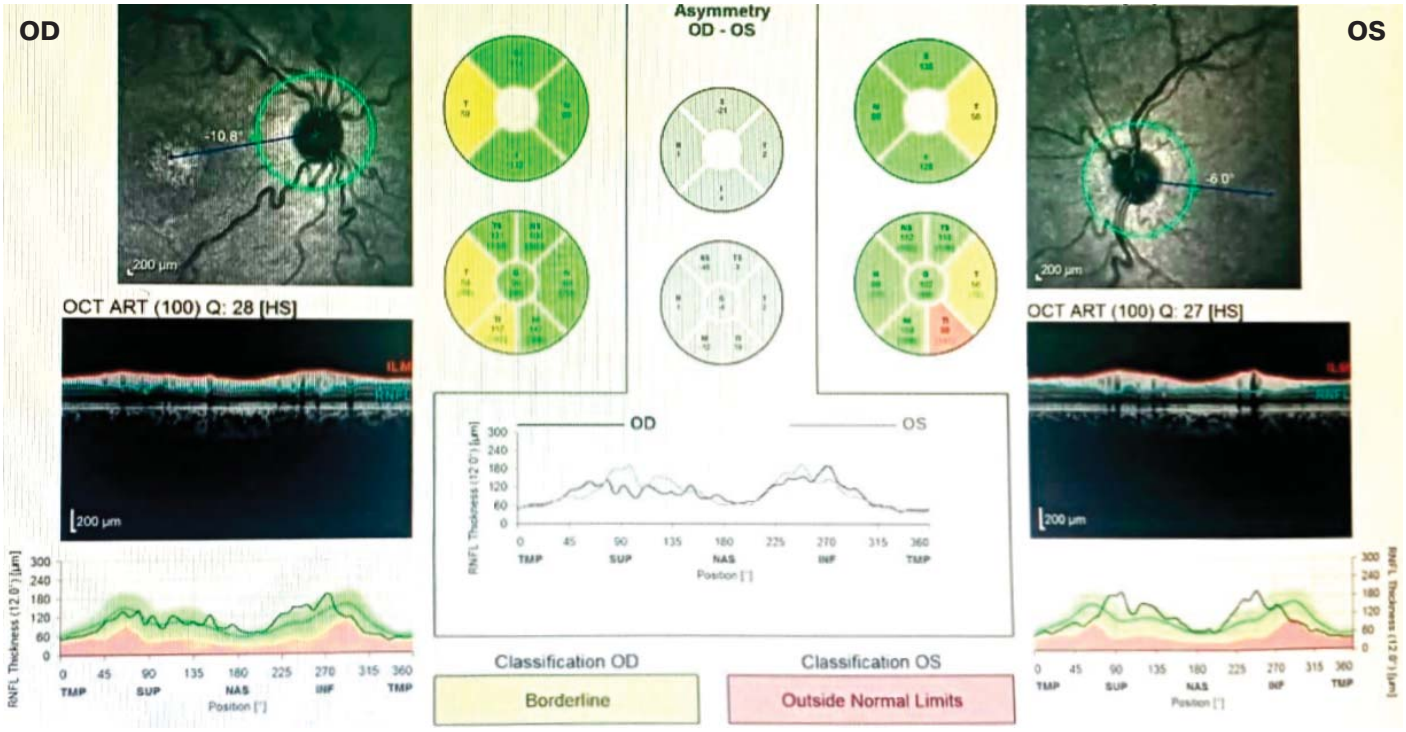

Fig. 3. OCT RNFL Analysis. OD — RNFL classified as borderline, with mild thinning in the inferior quadrant. Other quadrants are within normal limits. OS — RNFL classified as outside normal limits, with significant thinning in the inferior and superior quadrants

Рис. 3. Анализ слоя нервных волокон сетчатки (СНВ) с помощью ОКТ. OD — СНВ классифицируется как пограничное состояние, с умеренным истончением в нижнем квадранте. В других квадрантах показатели в пределах нормы. OS — СНВ классифицируется как выходящее за пределы нормы, со значительным истончением в нижнем и верхнем квадрантах

full-field electroretinogram (ffERG) patterns, and reduced visual acuity (VA), emphasizing UWF-FAF's role in assessing disease severity and visual function. This study highlights the importance of comprehensive diagnostic approaches, combining clinical examination with advanced imaging modalities such as OCT and FAF, to confirm hallmark features of STGD1. A key strength is the emphasis on low vision assessment, which addresses the patient's specific functional challenges, such as photophobia and difficulty with distance tasks. The integration of visual aids, including the successful use of a yellow filter and a telescope, demonstrates the potential of tailored interventions to enhance the patient's visual comfort, functional independence, and quality of life. Avoiding foods or supplements high in Vitamin A may help prevent exacerbation of lipofuscin accumulation in the retina [10].

Furthermore, the study underscores the necessity of regular follow-ups to assess residual visual acuity as the disease progresses. Follow-ups allow for the evaluation of current aids and provide opportunities to trial different telescopes or other devices, ensuring the most effective solutions are provided after clinical assessment and approval. Additionally, the case emphasizes the significance of genetic counseling and preventive measures for consanguineous families, addressing a critical public health concern in regions with a high prevalence of consanguinity. The focus on environmental modifications and lifestyle adjustments reflects a holistic approach to managing progressive vision loss. This study is limited by its focus on a single case, which may not capture the full spectrum of Stargardt disease presentations or responses to interventions. The long-term efficacy of the low vision aids, such as the yellow filter and telescope, was not assessed due to the lack of extended follow-up data. Furthermore, while genetic counseling is recommended, the outcomes or impact of this intervention were not evaluated. Additionally, the study does not explore or include patient access to emerging FDA-approved treatments, which could influence management strategies in the future.

Two treatment approaches have entered clinical trials. Gene therapy aims to replace the mutated gene with a healthy one in retinal cells, potentially halting or slowing lipofuscin accumulation and vision loss. Another approach involves transplanting RPE cells derived from stem cells to help clear lipofuscin buildup, with early trials already underway [11].

References/Литература

1. Dandona L, Dandona R. Revision of visual impairment definitions in the International Statistical Classification of Diseases. *BMC Med.* 2006 Mar 16; 4: 7. doi: 10.1186/1741-7015-4-7
2. Westerfeld C, Mukai S. Stargardt's disease and the ABCR gene. *Semin Ophthalmol.* 2008 Jan-Feb; 23 (1): 59–65. doi: 10.1080/08820530701745249
3. Al-Khuzaei S, Broadgate S, Foster CR, et al. An overview of the genetics of ABCA4 retinopathies, an evolving story. *Genes (Basel).* 2021 Aug 13; 12 (8): 1241. doi: 10.3390/genes12081241
4. Fishman GA. Fundus flavimaculatus. A clinical classification. *Arch Ophthalmol.* 1976 Dec; 94 (12): 2061–7. doi: 10.1001/archopht.1976.03910040721003
5. Huang D, Heath Jeffery RC, Aung-Htut MT, et al. Stargardt disease and progress in therapeutic strategies. *Ophthalmic Genet.* 2022 Feb; 43 (1): 1–26. doi: 10.1080/13816810.2021.1966053
6. Maguire AM, Bennett J, Aleman EM, Leroy BP, Aleman TS. Clinical perspective: Treating RPE65-associated retinal dystrophy. *Mol Ther.* 2021 Feb 3; 29 (2): 442–63. doi: 10.1016/j.ymthe.2020.11.029
7. Fujinami K, Waheed N, Laich Y, et al. Stargardt macular dystrophy and therapeutic approaches. *Br J Ophthalmol.* 2024 Mar 20; 108 (4): 495–505. doi: 10.1136/bjo-2022-323071
8. Ghenciu LA, Hațegan OA, Stoicescu ER, Iacob R, Șișu AM. Emerging therapeutic approaches and genetic insights in Stargardt disease: A comprehensive review. *Int J Mol Sci.* 2024 Aug 14; 25 (16): 8859. doi: 10.3390/ijms25168859
9. Abalem MF, Otte B, Andrews C, et al. Peripheral visual fields in ABCA4 Stargardt disease and correlation with disease extent on ultra-widefield fundus autofluorescence. *Am J Ophthalmol.* 2017 Dec; 184: 181–8. doi: 10.1016/j.ajo.2017.10.006

10. Sofi F, Sodi A, Franco F, et al. Dietary profile of patients with Stargardt's disease and Retinitis Pigmentosa: is there a role for a nutritional approach? *BMC Ophthalmol.* 2016 Jan 22; 16: 13. doi: 10.1186/s12886-016-0187-3
11. Rahman N, Georgiou M, Khan KN, Michaelides M. Macular dystrophies: clinical and imaging features, molecular genetics and therapeutic options. *Br J Ophthalmol.* 2020 Apr; 104 (4): 451–60. doi: 10.1136/bjophthalmol-2019-315086

Author's contributions: R. Faheem — conception and design, data acquisition, interpretation and drafting; M. Saad — revising, critical review and interpretation; S. Ullah — revising and critical review, final approval of work; M. Noor — revising and critical review; I. Nasim — revising and critical review.
Вклад авторов в работу: Р. Фахим — разработка концепции и дизайна, сбор данных и их интерпретация, написание статьи; М. Саад — редактирование, критический анализ и интерпретация; С. Улла — редактирование и критический анализ, окончательное утверждение работы; М. Нур — редактирование и критический анализ; И. Насим — редактирование и критический анализ.

Originally received: 06.12.2024. Final revision: 03.01.2025. Accepted: 04.01.2025
Поступила: 06.12.2024. Переработана: 03.01.2025. Принята к печати: 04.01.2025

INFORMATION ABOUT THE AUTHORS/ИНФОРМАЦИЯ ОБ АВТОРАХ

[1] Pakistan Institute of Ophthalmology, Al-Shifa Trust Eye Hospital Rawalpindi, 4600, Pakistan
[2] Lincoln University College, Wisma Lincoln, 4730, Petaling Jaya, Selangor Darul Ehsan, Malaysia
Rabia Faheem — Optometrist[1], ORCID 0009-0007-3212-0188
Muhammad Saad — Dr. of Medicine, fellow, Ophthalmology Department[1], ORCID 0009-0002-7235-5241
Saif Ullah — PhD Scholar[1], assistant professor, department of Health Sciences[2], ORCID 0000-0003-3254-5585
Muqaddas Noor — B.Sc. Honor Optometry[1]
Iffat Nasim — B.Sc. Honor Optometry and Orthoptics, Optometrist[1]
For contacts: Rabia Faheem,
rabiafahim3@gmail.com

[1] Пакистанский институт офтальмологии, глазная больница «Аль-Шифа», Равалпинди, 4600, Пакистан
[2] Университетский колледж Линкольна, Висма Линкольн, 4730, Петалинг Джая, Селангор Дарул Эхсан, Малайзия
Рабия Фахим — оптометрист[1], ORCID 0009-0007-3212-0188
Мухаммад Саад — д-р мед. наук, научный сотрудник отделения офтальмологии[1], ORCID 0009-0002-7235-5241
Саиф Улла — д-р философии[1], доцент кафедры медицинских наук[2], ORCID 0000-0003-3254-5585
Мукаддас Нур — бакалавр наук по оптометрии[1]
Иффат Насим — бакалавр наук по оптометрии и ортоптике, оптометрист[1]
Для контактов: Рабия Фахим,
rabiafahim3@gmail.com