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Understanding early-onset cataracts: A rare case of hereditary influence without metabolic disorders

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Abstract
Background: Early-onset cataract is a rare clinical presentation, particularly in the absence of systemic comorbidities. This case emphasizes the interaction between familial predisposition and environmental factors in a 57-year-old woman who has no history of diabetes mellitus, hypertension, or significant systemic illness.
Case Presentation: A South Asian school teacher reported occasional blurriness and watery eyes because of protracted digital screen exposure. An early-stage bilateral cataract was identified during the clinical evaluation, with normal metabolic parameters. Family history was notable for cataracts in her father, who also had diabetes mellitus type 2. Grade 1 cataract was confirmed by a slit-lamp exam, and retinopathy was excluded by fundoscopy.
Intervention and Outcome: To alleviate symptoms and avert progression, the patient was prescribed multivitamins, tinted spectacles, and hydrating eye drops. The patient was informed of the indications of progression and scheduled for routine follow-up appointments. There has been no evidence of metabolic abnormalities or symptoms progression to date.
Conclusion: This case emphasizes the significance of early detection, tailored interventions, and patient education in the management of early-onset cataracts, particularly in individuals with a positive family history.

Keywords: Early-onset cataract, familial predisposition, digital screen exposure, slit-lamp examination, patient education

Introduction

Cataract is defined as any opacity of the crystalline lens, results when the refractive index of the lens varies significantly over distances approximating the wavelength of the transmitted light. This variation in the refractive index can result from changes in lens cell structure, changes in lens protein constituents, or both. Defined by age at onset, a congenital or infantile cataract is visible within the first year of life; a juvenile cataract occurs within the first decade of life; a presenile cataract occurs before age 45 years; and senile or age-related cataract, after 60 years of age [1]. Increase susceptibility to environmental insults, such as light, hyperglycaemia, or oxidative damage contribute to age-related cataract [2]. Majority of age-related cataracts generally occur after 60 years of age but symptoms may start to notice under the age of 60 if associated with underlying conditions such as diabetes [3].

According to 2017 Global Burden of Disease, cataract is the second largest burden of eye disease counting 8 million affected people worldwide [4]. Mostly age onset cataract is secondary to metabolic disorders, infection, or trauma but a positive family history of cataract can be the primary reason for early onset cataract.

Certain genetic mutations are associated with the occurrence of congenital cataracts, which are present at birth. In the context of age-related cataracts, it has been observed that genetic mutations may increase the susceptibility of the lens to damage caused by environmental risk factors [3]. The genes under consideration are involved in antioxidant metabolism (GSTM1, GSTT1), lactose metabolism (LCT), drug metabolism (NAT2), folate metabolism (MTHFR), DNA repair (XPD), lipid and cholesterol transport (APOE), kinesin and microtubule motor transport (KLC1), actin-cytoskeleton regulation (EZR), chaperone activity (HSP70), and ephrin signalling (EFNA5) [2].

This rare case is a presentation of early onset cataract in 57-year-old with no history of ocular issues, diabetes mellitus (DM), or hypertension but has a positive family history of cataract in her father.

Case Presentation

A 57-year-old South Asian woman employed as a school teacher reported to the clinic on 21 October 2023 with primary complaints of lacrimation and intermittent visual blurriness, particularly following prolonged computer use. Her symptoms commenced suddenly, within the past month, and she has never encountered them previously. She has no previous history of ocular problems, diabetes mellitus, or hypertension. Her father has a medical history of cataracts for which he underwent surgery; he also has type 2 diabetes mellitus and asthma. The patient's normal health examination conducted last month revealed borderline blood sugar levels [HbA1c = 3.2%, fasting plasma glucose 95 mg/dl, oral glucose tolerance test 120 mg/dl].

She possesses an unremarkable medical history and does not consume any drugs. The patient reported experiencing periodic bladder infections treated with medications and a significant body odour, necessitating three showers daily. Her primary concern is the detrimental effect of her ocular symptoms on her teaching efficacy, given that her vocation necessitates extensive engagement with digital systems.

Clinical Findings

During the ocular examination, the patient exhibited a slight reduction in visual acuity while utilizing spherical and cylindrical prescription eyeglasses. A slit-lamp examination revealed early indications of cataract formation in both eyes. No signs of conjunctival or corneal injury were present, and intraocular pressure was within normal limits. A fundoscopic examination revealed early beginnings of cataract, with no evidence of diabetic or hypertensive retinopathy. Her fasting glucose level today was 98, with a HbA1c of 3.4%, indicating normal glucose metabolism.

Table 1: Timeline of the events in this case

Timeline	Event
Past Medical History	No history of ocular issues, DM, or Hypertension Has history of occasional urinary infections and notable sweat odor
Family History	Father had cataracts (had cataract surgery), DM type 2, Asthma
Initial Visit	Primary complaint of watery eyes and occasional blurriness
General Physical Exam	Normal with no signs of systemic illness
Ocular examination	Mildly decreased visual acuity Snellen acuity: 20/40; use powered eyeglasses; Mild cloudy cornea and normal conjunctiva (Mild grade 1 cataract)
Slit-lamp exam	Early signs of cataract development
Fundoscopic Exam	Early stage of cataract noted
Blood Tests	Normal fasting blood glucose levels and HBA1C is 3.4%

Therapeutic Intervention

The patient received a diagnosis of mild Grade 1 cataract and was thus advised to undergo follow-up examinations every six months, along with routine metabolic blood tests, including blood sugar and lipid profile assessments. The patient was advised to utilize hydrating eye drops and multivitamins, including vitamin A. She was informed and instructed regarding the signs and symptoms of cataract advancement and was urged to report manifestations such as impaired vision, floaters, flashes, and diplopia. Tinted lenses were advised to diminish light exposure to the eye, preventing vision impairment caused by lens transparency. The patient was advised of the potential necessity for cataract surgery should the cataract progress. The patient is currently in good health and has not expressed any eye issues. Her metabolic data are under the normal range, and she is adhering to the recommended eye care regimen.

Discussion

This case highlights the significance of prompt diagnosis, comprehensive assessment, and individualized patient education in the management of early-onset cataracts, especially in individuals with a familial predisposition to the condition. Multidisciplinary care, incorporating metabolic and lifestyle factors, is essential for preventing disease progression and enhancing patient outcomes.

The diagnosis and management in this case integrated clinical evidence with a focus on patient-centred care. Genetic predisposition, in conjunction with early symptoms, required a tailored strategy focusing on prevention and education. Consistent follow-ups and metabolic assessments maintain control of the condition, postponing the need for surgical intervention.

Conclusion

While age-related cataracts frequently occur post-60 years, this case highlights the significance of genetic predisposition in early onset. Cataract is predominantly linked to type 2 diabetes mellitus. This case represents a distinctive instance of negative comorbidities that challenges the diagnosis and treatment protocol. The patient's laptop usage and positive family history of cataract were significant factors in her case. Numerous genes regulate cataract formation, and this aspect should not be overlooked. This case emphasises the necessity of regular follow-ups in individuals at elevated risk for type 2 diabetes mellitus and hypercholesterolemia. Such patients should be counselled on the importance of routine check-ups and physical activity.

Conflict of Interest

Not available

Financial Support

Not available

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