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Case Report

Unilateral Congenital Lamellar Cataract with Glucose-6-Phosphate Dehydrogenase Deficiency: A Case Report

Munish Sagar[1] and Anuradha Raj[2]*

[1]*Medical Officer, Department of Pediatrics, Jalandhar, Punjab, India*

[2]*Professor and HOD, Department of Ophthalmology, All India Institute of Medical Sciences, Bathinda, Punjab, India*

***Corresponding Author:** Anuradha Raj, Professor and HOD, Department of Ophthalmology, All India Institute of Medical Sciences, Bathinda, Punjab, India.

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Abstract

Glucose-6-phosphate dehydrogenase deficiency (G6PDD) is characterized by a genetic abnormality of the enzyme named Glucose-6-Phosphate dehydrogenase (G6PD) which leads to hemolysis, neonatal jaundice and anemia. Bilateral cataract can be the ocular manifestation of G6PDD. This case reports a patient with G6PDD who presented to us with a unilateral typical lamellar cataract.

Keywords: *Lamellar Cataract; Glucose-6-Phosphate Dehydrogenase Deficiency; Red Blood Cells*

Introduction

Glucose-6-Phosphate dehydrogenase deficiency (G6PDD) is a genetic metabolic abnormality due to deficiency of the Glucose-6-Phosphate dehydrogenase (G6PD) enzyme which is critical for the proper functioning of red blood cells (RBCs). This condition leads to hemolysis due to premature breakdown of RBCs. G6PD enzyme brings about its co-enzyme nicotinamide adenine dinucleotide phosphate hydrogen (NADPH) which protects cells from oxidative damage. G6PDD leads to lower levels of NADPH and depletion of an antioxidant glutathione which protects cells from oxidative damage. The oxidative stress leads to the denaturation of lens proteins and forms insoluble aggregates resulting in cataract [1]. In the literature, there is evidence of bilateral cataracts with neonatal sepsis associated with G6PDD [2].

We report a case of a young male child with unilateral lamellar cataract with G6PDD. To the best of our knowledge, we have not found any evidence of the same scenario in the literature.

Case Presentation

A 5-year-old male child was referred to us with a diminution of vision in the left eye since birth. As informed by his father the diminution of vision was insidious in onset and progressed gradually. There was a history of glare in the left eye and frequent blinking of both eyes since birth. These symptoms increased during the daytime and decreased during the night. The patient could not look at them or any light source properly and was habitual of turning his face to the opposite side of the light source. There was no history of ocular trauma or any ocular surgery There were no such complaints in the right eye. The systemic history was unremarkable without any drug intake or drug allergies.

The mother was suffering from pregnancy-induced hypertension from the 6th month of gestation and had anemia and decreased sleep during pregnancy. She had no history of any infection, fever, or rashes during pregnancy.

The patient was born prematurely at 32 weeks of gestation with a birth weight of 1.75 kg. There was the history of prolonged neonatal jaundice at the time of birth and was diagnosed with a case of G6PDD based on a fluorescent spot test which was done elsewhere. His Immunization was up to date. There was no history of birth trauma or ocular infections after birth.

On ophthalmological examination, the best corrected visual acuity was 20/20 and 20/80 in the right and left eye, respectively with -0.5.00 Dioptre (D) sphere and -1.00 D sphere. Keratometry for both eyes was 42.75 D/43.25 D @90° and 42.50 D/44.25 D @90°, respectively. The axial lengths were 22.0 mm and 22.2 mm for the right and left eye, respectively.

A slit-lamp examination showed a disc-shaped configuration of cataract in left eye. Arcuate opacities within the cortex straddle the equator of the lamellar cataract. The horseshoe-shaped opacities called riders were visible on retroillimination (Figure 1a and 1b). No evidence of phacodonesis and iridodonesis was observed.

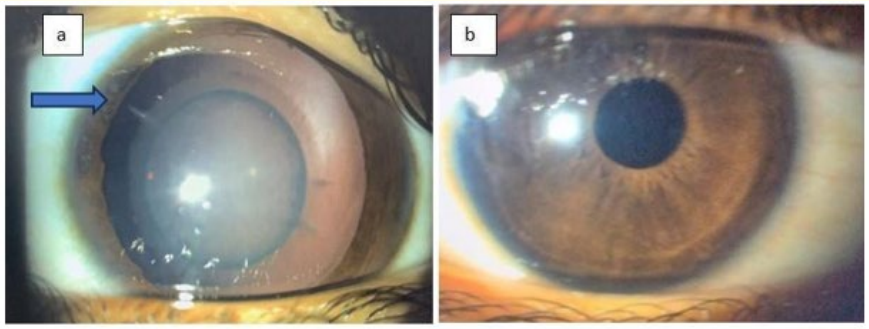

Figure 1: a. Shows rider on retro illumination b. Postoperative anterior segment

Figure

Extraocular movements were full, free and painless. No evidence of strabismus or nystagmus was seen.

Fundus examination was unremarkable for both eyes.

He was diagnosed as a case of unilateral lamellar congenital cataract with G6PDD. The patient underwent phaco aspiration with foldable intraocular lens implantation. Post-operative follow-up was uneventful and his best corrected visual acuity was 20/30 and N/8 with -0.5 D sphere and +3.0 D Sphere respectively.

Discussion

G6PD plays an important role in defense against cellular oxidative stress. The most common manifestation of G6PDD is jaundice and hemolysis due to oxidative stress. Almost all form of cataracts are associated with oxidative injury. Bilateral cataracts and color blindness are seen in G6PDD [2].

A lamellar or zonular cataract is a hereditary cataract that has a variable phenotype. It is inherited as an autosomal dominant trait and usually, it is bilateral and symmetrical [3]. This is a type of developmental cataract in which the fine crystalline opacities occupy a discrete zone located at the level of the primary fibers in the embryonic nucleus [4]. It is the most common type of congenital cataract presenting

with visual impairment. Environmental factors such as vitamin D deficiency, hypocalcemia, malnutrition and maternal rubella infection contracted between the 7th and 8th week of gestation can play an important role in the pathogenesis of the lamellar cataract

Clinically, the cataract is visible as an opacified layer that surrounds a clear center. The Arcuate opacities within the cortex straddle the equator and these horseshoe-shaped opacities are called riders. Typically occurs in the zone of the fetal nucleus surrounding the embryonic nucleus is involved with peripheral clear lamellae and linear opacities like the spokes of a wheel (called riders) may run outwards towards the equator and riders which are pathognomic [4].

Unilateral congenital cataracts are believed to result from localized ocular dysgenesis. Approximately 80– 90% of unilateral cataracts are idiopathic. Early diagnosis and prompt treatment are critical for visually significant unilateral congenital cataracts due to dominant normal eye competition and to avoid amblyopia [5]. In this current case, the typical lamellar cataract is seen as unilateral which is different from the existing literature [6].

Conclusion

This study provides clinical evidence that though bilateral congenital cataract can be seen in patients with G6PDD, unilateral lamellar cataract can also be seen in such cases. Early diagnosis and timely management are mandatory for better outcomes by avoiding amblyopia.

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