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Visual Outcomes in Three Siblings with Bilateral Inherited Congenital Cataract: A Case Series

Nur Fitriana Corprianti Marchilia[1,2], Indri Wahyuni[1, *2]

[1]Department of Ophthalmology, Faculty of Medicine Universitas Airlangga, Surabaya

[2]Department of Ophthalmology, Dr Soetomo General Academic Hospital, Surabaya

***Correspondence Author:**

Email ID: indri-w@fk.unair.ac.id

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ABSTRACT

Congenital cataracts, often hereditary, contribute significantly to visual morbidity in children. In this case series, we explore the visual outcomes of three siblings with bilateral inherited congenital cataracts, all of whom underwent lens extraction. The study was conducted at Dr. Soetomo General Hospital in Surabaya, Indonesia, and followed these patients for two years post-surgery. The siblings exhibited varying ages of presentation and surgical intervention timelines, leading to different visual outcomes. The first patient, a 15-year-old girl, experienced significant opacities and poor visual acuity, which improved minimally with aphakic glasses and Nd:YAG laser capsulotomy. The second patient, an 11-year-old girl, had similar lens opacities, and despite lens extraction, her visual acuity improved to 5/30 after two years. The third patient, a 13-year-old boy, underwent early lens extraction but had suboptimal visual outcomes due to posterior capsule opacification (PCO). The inherited condition followed an autosomal dominant pattern, with the mother who had also suffered from the same condition since childhood. The timing of diagnosis and surgical intervention played a crucial role in the visual outcomes. This study emphasizes the importance of early detection, surgical intervention, and effective postoperative rehabilitation in managing congenital cataracts, highlighting that early intervention can significantly influence visual development and reduce the risks of amblyopia and strabismus.

Keywords: *Congenital cataract, hereditary, visual outcomes, pediatric cataract surgery, postoperative rehabilitation.*

1. INTRODUCTION

Hereditary factors are responsible for one-third of all congenital cataract cases. Autosomal dominance is the most common type, occurring in 44% of families. Inherited congenital cataract significantly contribute to visual morbidity in children. The severity depend on the type of underlying gene mutation. Parents of children with congenital cataracts need a comprehensive information about treatment, the prognosis for potential visual outcomes, and how the condition might impact their child's future growth and academic development. Antenatal screening enable prompt medical attention and much improves the prognosis [1].

Early presentation, timely surgical intervention, and effective postoperative visual rehabilitation all contribute to better visual outcomes following paediatric cataract surgery. However, available statistics indicate that a significant number of children who underwent cataract surgery did not experience substantial vision improvement due to suboptimal surgical result or inadequate visual rehabilitation. A study revealed that outcomes vary depending on the age at diagnosis and timeliness of surgical intervention [2]. This study highlight the visual outcomes of three siblings in a family with bilateral hereditary congenital cataracts. Each of whom had a different timelines for age of presentation and surgical intervention. This case series describes three siblings presenting with bilateral inherited congenital cataract who underwent lens extraction. This study was conducted at Dr. Soetomo General Hospital Surabaya, East Java, Indonesia. All patients were follow up for a period of two years after their initial presentation.

2. CASE ILLUSTRATION

There are seven children in a family. Three of whom had bilateral congenital cataracts The mother was affected by the same condition but had also neglected it. She brought her child to the outpatient clinic at different times.

Case 1: A 15-year-old girl, the third child, was referred to an ophthalmologist with a chief complaint of difficulty reading since the age of nine. She had previously consulted another ophthalmologist, who reported leukocoria and advised lens extraction. However, the mother refused due to financial and transportation issues. Currently, the patient experienced worsening vision and frequent involuntary eye movements. There was no history of abnormalities during pregnancy, birth, systemic disease, or trauma. On examination, her visual acuity in both eyes was limited to hand movements, and both lenses exhibited significant opacities (Figure 1). Ocular ultrasonography showed normal findings (Figure 2), and laboratory tests for Toxoplasma, Rubella, Cytomegalovirus (CMV), and Herpes (TORCH) infections were negative. Based on these findings, the patient was diagnosed with bilateral inherited congenital cataracts. Examination under general anesthesia revealed horizontal and vertical corneal diameters of 9 mm in both eyes. Lens extraction was performed without intraocular lens implantation. Aphakic glasses were planned to correct vision once postoperative inflammation resolved.Two years later, her visual acuity was 3 meters counting fingers, with difficulty achieving correction due to posterior capsule opacification. She had not returned to the clinic earlier and had not received aphakic glasses as planned. Neodymium-doped yttrium aluminium garnet (Nd:YAG) laser capsulotomy was performed, followed by the prescription of aphakic glasses, which improved her binocular visual acuity to 5/15.

Figure 1. (A) and (B) shows the lens opacity in anterior segment.

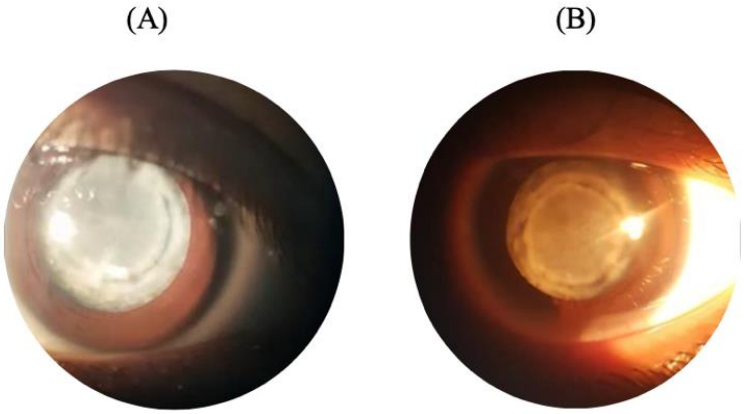
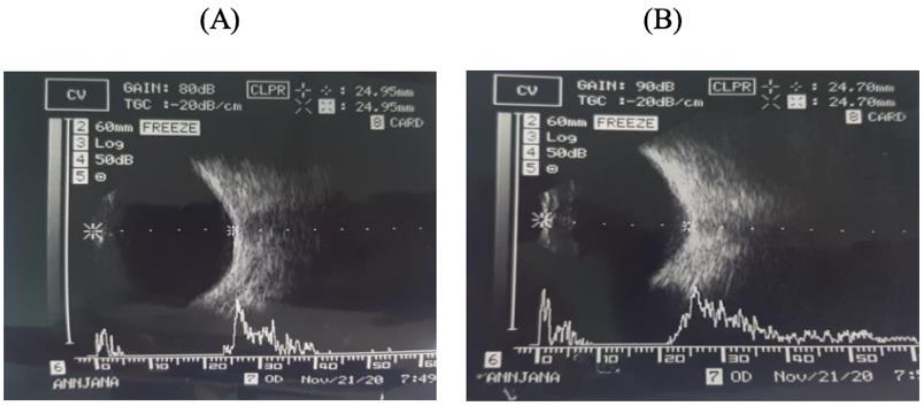


Figure 2. (A) Right eye ultrasonography result. (B) Left eye ultrasonography result. Both show vitreous echogenic free and retina on place.



Case 2: An 11-year-old girl, the sixth child among the siblings, was referred to an ophthalmologist with a chief complaint of whitish matter in both eyes, which had been noticeable since birth. She was brought to consultation shortly after her sister underwent surgery. Her visual acuity was 3 meters counting fingers in the right eye and 1 meter counting fingers in the left eye. Examination revealed significant lens opacities in both eyes, with normal findings on ocular ultrasonography (Figures 3 and 4). Laboratory tests for TORCH serology were negative.Preoperative examination under general anesthesia showed bilateral microcornea. Lens extraction without intraocular lens implantation was performed. Aphakic glasses were prescribed

for visual correction, but unfortunately, she returned to the clinic only two years later. At that time, her visual acuity had deteriorated to 1 meter counting fingers in both eyes, with difficulty achieving correction due to posterior capsule opacification. Nd:YAG laser capsulotomy was performed, and correction with aphakic glasses improved her visual acuity to 5/30 in both eyes.

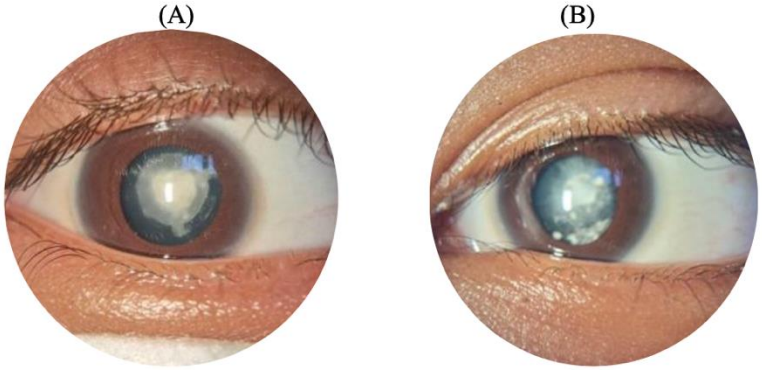


Figure 3. (A) and (B) are the anterior segment documentation of 2nd patient at first presentation. Both eyes showed opacity in her lens.

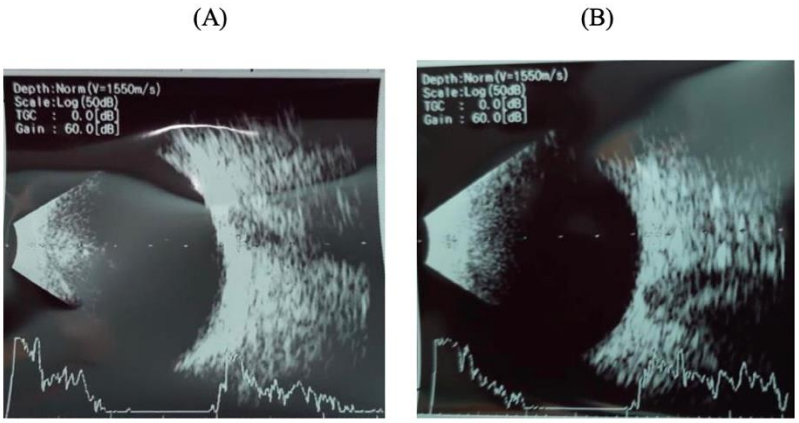


Figure 4. (A) Right eye ultrasonography result. (B) Left eye ultrasonography result. Both show echogenic free at vitreous and retina on place.

Case 3: A 13-year-old boy was referred at the same time as the second patient. He had a history of lens opacity in both eyes since birth. At nine months old, he underwent lens extraction at another institution. On examination, his visual acuity was 4/32 in the right eye and 1/40 in the left eye, both of which were difficult to correct. This condition was likely caused by posterior capsule opacification. Additionally, the pupils were irregular in shape and decentered, possibly as a postoperative consequence of the earlier procedure (Figure 5).

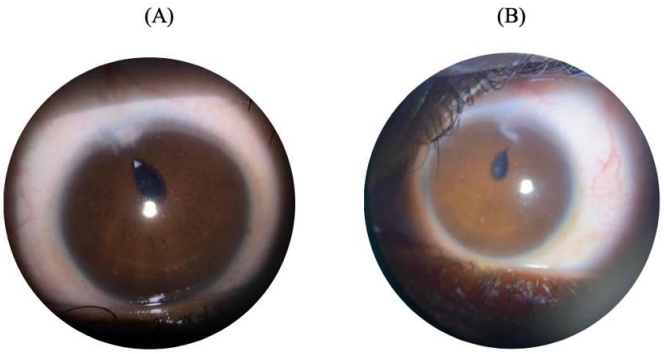


Figure 5. Anterior segment of the 3rd patients shows an aphakic eye, opacification in posterior capsul, and the pupil not round in shape.

Their ages at initial diagnosis and management varied among the patients. Each patient has a unique clinical presentation both before and after surgery. Their visual acuity also differed when they returned two years later. The family history and genetic pattern of congenital cataract in this family are depicted in Figure 6.

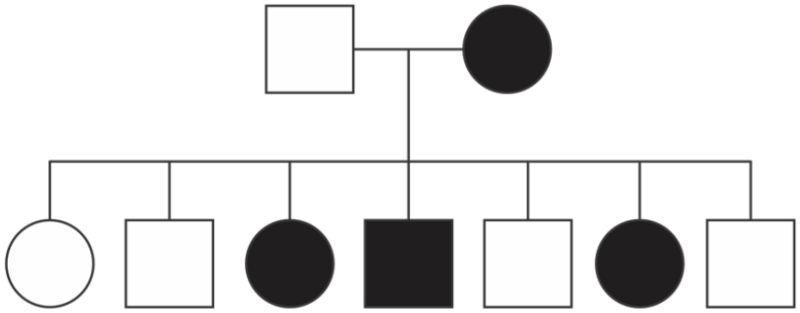


Figure 6. A family pedigree of this family. The black in colour were affected individual. The mother and three of seven child are affected.

3. DISCUSSION

A hereditary component is responsible for 8% to 30% of congenital cataracts. Inherited congenital cataracts are categorized into two types: syndromic and non-syndromic. Among these, the most common mode of inheritance is autosomal dominant. Both dominant and recessive patterns of autosomal inheritance can be observed in congenital cataracts without other systemic abnormalities. Lens opacities may occur in isolation or alongside other ocular abnormalities, particularly in non-syndromic cataracts, and without associated systemic conditions [1,4]. In this case, approximately 50% of the children in the family inherited the condition from their mother, who had also suffered from the same condition since childhood. The inheritance pattern was determined to be autosomal dominant, as the condition affected individuals regardless of gender, and a single mutated copy of the gene from one parent was sufficient to cause the disorder. A child born to a parent with an autosomal dominant condition has a 50% chance of inheriting the mutated allele.

Congenital cataracts can also result from various factors, including intrauterine infections, exposure to radiation during pregnancy, certain medications, or metabolic disorders [5]. Viral and parasitic infections during gestation, such as those caused by Toxoplasma, Rubella, Cytomegalovirus (CMV), and Herpes virus (collectively known as the TORCH group), are significant contributors to the development of congenital cataracts. The presence of TORCH infections can be confirmed by measuring serum immunoglobulin (Ig) M and Ig G titers [6]. In this case, serologic assays for the first and second patients yielded negative results. Additionally, there was no significant history of radiation exposure or drug use during pregnancy. Therefore, despite the absence of genomic DNA analysis, the congenital cataracts in this family are most likely attributed to a genetic component.

The timing of surgery is one of the primary factors influencing visual prognosis; therefore, early detection of congenital cataracts is essential, particularly in cases with a family history. This often requires collaboration among multidisciplinary teams [7]. Globally, congenital cataract screening programs have been established to enhance the care provided to affected children. These initiatives include early gestational genetic testing and prenatal ultrasound imaging for diagnosis. The first patient had a congenital cataract and was the third child in the family. The mother was also affected by the same condition. She reported experiencing cataracts and blurred vision as a toddler, but her condition was neglected. Despite having seven children, she did not undergo genetic testing after the birth of her children or screening examinations during her pregnancies.

The first and second patients presented with similar signs and symptoms, except for differences in visual acuity. The first patient's visual acuity was limited to hand movements in both eyes, whereas the second patient had a visual acuity of 3/60 in the right eye and 1/60 in the left eye. The first patient has poorer visual acuity, likely due to her denser and thicker lens compared to her sister's. Both siblings also suffer from nystagmus. Since the third patient's preoperative data was unavailable, a comparison with the others could not be made. All affected children in this family have non-syndromic congenital cataracts. A comprehensive, multidisciplinary evaluation, including assessments by the pediatric and ENT (Ear Nose Throat) departments, confirmed the absence of any systemic abnormalities.

Compared to adult cataracts, pediatric cataracts are more challenging to treat. These challenges include anatomical differences, timing of surgery, prevention and management of amblyopia, addressing visual axis opacification, and determining the optimal type and power of intraocular lenses. Despite surgical intervention, the outcomes are often suboptimal, with a high risk of amblyopia. For unilateral cases, surgery should ideally be performed before 6 weeks of age,

and for bilateral cases, before 8 weeks [8,9]. While not statistically significant, a study observed a linear trend indicating that visual acuity outcomes at ages 4 to 6 tend to worsen as the age at the time of surgery increases. Another study found that the mean visual acuity decreased by one line for every three weeks that surgery was delayed between 0 to 14 weeks. However, the final visual acuity was not significantly affected by the age at surgery for patients operated on between 14 to 31 weeks. On average, visual acuity was reported as 20/80 [10].

The current gold standard for treating pediatric cataracts is microincision cataract aspiration combined with anterior vitrectomy, posterior capsulotomy, and primary intraocular lens (IOL) implantation. Posterior capsulotomy and anterior vitrectomy are especially important for minimizing the risk of posterior capsule opacification (PCO). These procedures are typically recommended until children reach 4 to 8 years of age, which is considered the cooperative age for Nd:YAG laser treatment. Primary IOL implantation is generally reserved for children older than two years. However, despite growing evidence supporting the safety of the procedure, IOL implantation in infants under two years old remains a topic of debate. Implanting IOLs before 7 months of age is associated with a higher risk of severe postoperative complications, including glaucoma, PCO, and inflammation, particularly in bilateral cases. Nonetheless, a recent meta-analysis found that primary IOL implantation in children under two years significantly improved visual acuity compared to aphakia managed with contact lenses. However, it also increased the risk of PCO.When cataract is linked to other eye disorders such microphthalmia, it is best to postpone IOL implantation [3,11]. Upon examination, the corneas of both patients were found to have a horizontal diameter of less than 10 mm in each eye, classifying them as microcornea. Implanting an intraocular lens (IOL) in the capsule bag is not recommended in such cases, as microcornea is often associated with zonular dialysis and subluxated cataracts. One of the traditional techniques for IOL placement in these situations is sutured scleral-fixated IOL, but this method carries a significant risk of exacerbating pre-existing retinal conditions, such as choroidal coloboma. While scleral-fixated IOL placement is possible in eyes with coloboma, achieving optimal placement can be highly challenging, and the scleral suturing technique may pose additional risks in such cases. Moreover, anterior chamber IOLs are not advisable due to the increased likelihood of endothelial damage, which can result from a shallow anterior chamber and difficulties in appropriately sizing the anterior chamber IOL [12]. Considering all these factors, the surgeon opted to remove the lens without implanting an IOL, leaving the eyes of both the first and second patients aphakic. Similarly, the third patient also presented with aphakia, but microphthalmia and microcornea cannot be definitively ruled out based on the available data. Both of the third patient's eyes remained aphakic, likely following surgery performed at nine months of age. However, the patient never underwent secondary IOL implantation as he did not return to the ophthalmologist for post-operative follow-ups.

Congenital cataracts pose a significant challenge for postoperative vision rehabilitation. To reduce the prevalence of amblyopia, strabismus, and poor fusion, optical restoration should begin as early as possible. The visual rehabilitation process includes IOL implantation, spectacles, contact lenses, and epikeratophakia. Spectacles are the least effective form of visual correction. Children with bilateral aphakia are often treated with aphakic spectacles. While contact lenses are considered optically superior to spectacles, they cannot be worn for extended periods. This case demonstrated that, although the third patient underwent surgery earlier than the others, his visual acuity remained low. Postoperative complications, including bilateral eccentric pupils and the development of PCO, may have contributed to his suboptimal visual outcomes. There was little difference in visual acuity between the second and third patients, though the second patient had better visual acuity than the third. The first patient, despite being the most recently diagnosed, managed, and treated, also developed PCO following surgery. A study has shown that bilateral surgery and older age at the time of surgery are associated with improved visual acuity. Since cataracts typically form during the period of visual maturity, children who undergo surgery later in life often experience normal visual development, which aligns with previous research [13]. These factors may have influenced the visual outcomes of the three siblings who underwent cataract surgery. Although there was minimal restoration of visual acuity two years post-surgery, an improvement in the vision chart line was observed. Research indicates that cataract surgery not only improves visual function but also enhances brain function, including increased gyrus volume in both visual and cognitively related regions. As a result, patients may find it easier to learn in class due to the restoration of their visual function [14].

4. CONCLUSIONS

The timing of diagnosis and treatment are predictive factors that influence visual outcomes after pediatric cataract surgery. The timing of surgery plays a crucial role in the development of vision. Furthermore, optimal visual outcomes are more likely when complications are identified early, managed effectively, and followed by appropriate visual rehabilitation therapy. In the medical field, providing prenatal counseling and recommendations to parents with a history of cataracts can help prevent late diagnoses and treatment when they eventually have a child.

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