CASE REPORT

A Case Series of Hereditary Congenital Cataract

Nurul Faaiqah Jainuddin[1], Azlindarita Aisyah Mohd Abdullah[1,2], Visvaraja Subrayan[1], Norlina Ramli[1], Nurliza Khaliddin[1]

[1] Ophthalmology Department, Faculty of Medicine, University Malaya, Malaysia
[2] Ophthalmology Department, Faculty of Medicine, Universiti Teknologi MARA, Sg Buloh, Selangor, Malaysia

ABSTRACT

Congenital cataract is a major cause of preventable blindness in children. It can be either hereditary or non-hereditary. In this case series, we present three cases of congenital cataract with a strong family history. The grandparents and parents of these patients had cataract diagnosed in late childhood whilst the patients in this case series were diagnosed with congenital cataract earlier. There was no history of consanguineous marriage in any of the families. These cases show that, in the presence of a strong family history, a child will present with congenital cataract earlier in their life. Hence, babies from such families should be screened at birth and at regular intervals within the first year of life to enable early diagnosis and subsequent surgical intervention to reduce the incidence and burden of amblyopia.

Keywords: Congenital cataract, Paediatric visual impairment, Hereditary cataract

Corresponding Author:
Nurliza Khaliddin, MOphthal
Email: nurliza@ummc.edu.my
Tel: +6013-3690415

INTRODUCTION

Congenital cataract is one of the leading causes of preventable blindness worldwide (1,2). The prevalence is 1 to 15 in 10,000 live births and it is higher in developing countries (1). Congenital cataract usually develops intrauterine but often present later in life due to lack of awareness. Time of presentation, laterality of cataract and the cause of cataract, will determine the timing, type of management and the mode of visual rehabilitation. We present 3 cases of congenital cataract with strong family history presenting at different ages.

CASE PRESENTATION

Case 1

An 8-month-old term female infant presented in May 2017 with leukocoria which was noticed by her parents since the age of 5 months. Both her father and her elder brother were diagnosed with bilateral congenital cataract at the age of 3 years and underwent bilateral cataract surgery in the same year. The patient was not syndromic and infectious screening including serum toxoplasmosis, cytomegalovirus, herpes and rubella were negative. The family was not keen to proceed with genetic studies. On examination, the child had bilateral leukocoria and an alternating exotropia. She underwent an uneventful bilateral lens aspiration, primary posterior capsulotomy, anterior vitrectomy and posterior chamber intraocular lens implantation at the age of 9 months. After 5 months, the left eye developed posterior capsule opacification which covered the visual axis nasally. A surgical posterior capsulectomy was carried out. Post-operatively both eyes remained orthophoric and good visual acuity was maintained since then. Cycloplegic refraction was +5.00DS for right eye and +6.50DS for left eye. Hence spectacles were prescribed for visual rehabilitation. This child is currently 2 years old and under 6 monthly follow up for cycloplegic refraction and visual rehabilitation. Parents are very supportive and compliant to the child's full-time spectacles wear and the follow up appointments (Table 1).

Case 2

A 1-month-old term male infant presented to our clinic in August 2018 with leukocoria. His mother and his sister were diagnosed with congenital cataract at 7 years old and 7 months old respectively. Both mother and elder sister underwent cataract surgery during their childhood. Clinically the patient has no dysmorphic features, no squint and nystagmus. Parents agreed to proceed with genetic test. Both mother and child have Nance Horan Syndrome gene detected. He underwent bilateral lens aspiration, primary posterior capsulotomy with anterior vitrectomy within 2 weeks of presentation. No intraocular lens was inserted and the patient remained aphakic. Two months after surgery, his right eye developed posterior synechiae with capsular phimosis involving the visual axis. Right eye anterior vitrectomy was performed and release of the synechiae was done. Post-operative cyclorefraction was +21.00DS

Table 1: Summary of our cases of congenital cataract

Case	Clinical presentation	Cause of congenital cataract	Operation	3 months post-operative refraction	1 year post-operative refraction
Case 1	Leucocoria	Not proceed with genetic test	Both eyes lens aspiration/ PCIOL	RE +8.00 LE +9.00/-1.50 x10	RE +5.00 LE +6.50 Patient not cooperative for VA
Case 2	Leucocoria	Nance Horan Syndrome (x-linked)	Both eyes lens aspiration/ aphakia	RE +21.00 LE +20.00	Follow up at private centre
Case 3	Roving eye movement	CMV infection	Both eyes lens aspiration/ aphakia	RE +18.00/-0.50 x90 LE +17.00/-0.50 x 90	RE +14.50 LE+16.00/-0.50 x90

for right eye and +20.00DS for left eye. Patient is currently 11-month-old and is visually rehabilitated with contact lens and aphakic glasses. He continued follow up at private centre.

Case 3

A 3-month-old full term male infant was noticed by his mother to have 'roving' movement of both eyes since birth. He made no eye contact and was unable to focus on objects in front of him. His father had a history of bilateral congenital cataract which was operated at 8 years old. His paternal uncle, paternal aunt and paternal grandmother were diagnosed with congenital cataract at birth, 3 years old and 22 years old respectively (Figure 1). All of them underwent cataract surgery at the time of diagnosis. Infectious screening was done for this patient and was positive for Cytomegalovirus infection IgM and IgG. Case was consulted with infectious disease doctor and no treatment was initiated since the child is well and healthy but need for regular monitoring. On examination, both eyes showed downbeat nystagmus and was unable to fixate. Leukocoria was present bilaterally. The patient underwent right eye lens aspiration, primary posterior capsulotomy and anterior vitrectomy followed by a similar operation on the left eye 2 weeks later. Both eyes were left aphakic. Postoperative cycloplegic refraction was +18.00/-0.50 x90 for right eye and +17.00/-0.50 x 90 for left eye. He was able to follow objects and nystagmus improved with contact lens and aphakic glasses.

DISCUSSION

Congenital cataract is the leading cause of preventable blindness in children worldwide and also in Malaysia (1,2). In Vision 2020, prevention of blindness due to congenital cataract has been put as an international priority, in accordance to its motto- The Right to Sight (5). The causes of congenital cataract can be either hereditary or sporadic. Inheritance patterns can be either autosomal dominant, autosomal recessive or X-linked. Most inherited cataracts will present bilaterally. Meanwhile sporadic cases usually present as unilateral cataract (3). There are more than 100 genes that has been found to be associated with congenital cataract in which most of them are autosomal dominant (3). The genes involved include PAX6 and PITX3(3). Other common causes of congenital cataract are infections (Toxoplasmosis, rubella, herpes group such as CMV, HSV1 and 2, VZV, syphilis, measles), inborn error of metabolic disorders and endocrine diseases.

A child with familial congenital cataract will present with leukocoria, nystagmus and strabismus earlier in life due to defect in developmental genes affecting lens development. This was evidenced by our cases and as shown in other studies (3). The cataract will develop earlier in life due to genes defect and usually they will present earlier because of the awareness among family members. Examining the parents and other family members may explain the aetiology of a child's cataract.

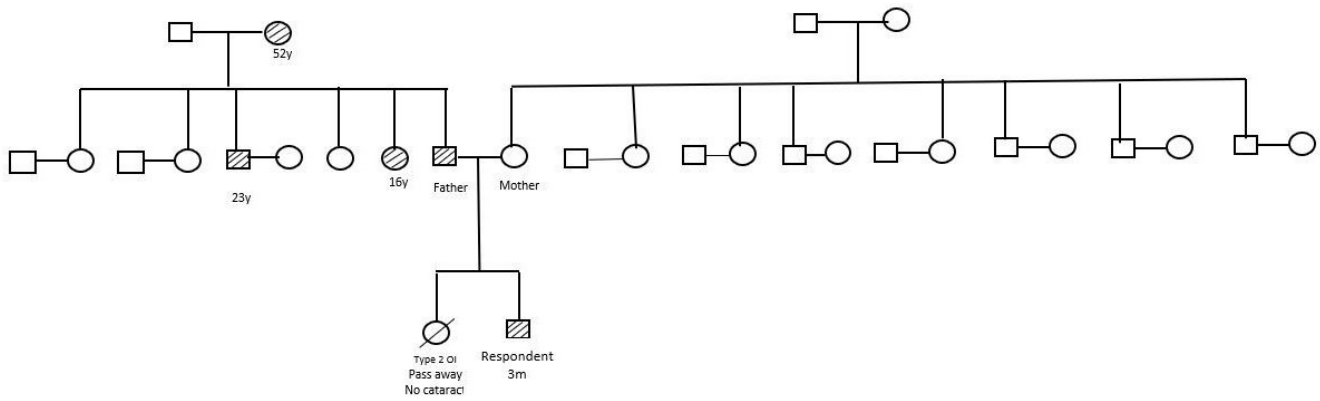

Figure 1: Family tree of respondent in case 3 who has strong family history of congenital cataract

Full antenatal history, systemic examination, and events during pregnancy, delivery and neonatal period are important.

It is essential for early diagnosis and detection of congenital cataract particularly in the presence of a dense cataract, for appropriate and timely intervention which in return will result in good visual function. Once diagnosis of visually significant cataract has been made, cataract surgery must be performed as early as possible to prevent irreversible amblyopia. The risks of the complications such as severe inflammation and secondary glaucoma need to be balanced with the benefit of early surgery and all factors must be discussed thoroughly with the parents to ensure adherence to the long term follow ups. For unilateral cataract, excellent results have been reported in children who undergo surgery before 6 weeks of age, and for bilateral cases, before 10 weeks of age (4). Worldwide, implantation of an artificial intraocular lens after removal of the cataract has become an established practice in children over 2 years of age (4). In our case, 2 patients were left aphakic and 1 patient had hydrophobic acrylic intraocular lens implantation. The decision of intraocular lens implantation is dependent on patients age at presentation, time of surgery, patients' condition, surgeon preference and parent's cooperation for visual rehabilitation (4). Those with easy access to health care and patients less than 2 years old can opt for contact lens or spectacles as the power can change according to the physiological age. If the patient is unable to come for frequent follow up or the family finds it difficult to adhere to visual rehabilitation, they can opt for intraocular lens implantation either as primary or secondary procedure. Specifically, in our first case, intraocular lens was implanted for early visual rehabilitation. Parents of the child were keen for intraocular lens implantation as they foresee difficulty in handling contact lens post operatively. But it is important to emphasize to the parents the risk of amblyopia later in life when the refractive correction is no longer suitable for the age. Hence, long term adherence to follow up is essential to monitor child progression.

The importance of post-operative rehabilitation must be emphasized to the parents. Compliance to medication and adherence to follow up is important to ensure the child retains good vision throughout their life. Despite early surgery and aggressive optical rehabilitation, the incidence of deprivation amblyopia, nystagmus, strabismus and glaucoma remains high.

CONCLUSION

Our case series demonstrated earlier presentation of congenital cataract in successive generations. Hence, awareness of congenital cataract and early neonatal screening is crucial to enable early surgical intervention which gives a better visual outcome. A strong family history of congenital cataract warrants genetic counselling for earlier detection of the disease. Management of congenital cataract remains a challenge to paediatric ophthalmologists worldwide. A multidisciplinary approach with good family support and education are essential in achieving optimal patient care.

ACKNOWLEDGEMENT

The authors are grateful to the parents and their family for the information and description.

REFERENCES

1. Apple DJ, Ram J, Foster A, Peng Q. Elimination of cataract blindness: a global perspective entering the new millenium. Surv Ophthalmol. 2000 Nov;45:S1–196.
2. Reddy, Sagili C, Benito CT. Causes of childhood blindness in Malaysia: result from a national study of blind school students. International Ophthalmology. 2001;24: 53-9.
3. Wirth MG, Russell-Eggitt IM, Craig JE, Elder JE, Mackey DA. Aetiology of congenital and paediatric cataract in an Australian population. British Journal of Ophthalmology. 2002; 86(7):782-6.
4. Solebo AL, Russell-Eggitt I, Nischal KK. Cataract surgery and primary intraocular lens implantation in children ≤2 years old in the UK and Ireland: finding of national surveys. British Journal of Ophthalmology. 2009; 93:1495-8.
5. Gilbert C, Foster A. Childhood blindness in the context of VISION 2020--the right to sight. Bull World Health Organ. 2003;79(3):227-32.