CLINICAL STUDY

Predictive value of family data for the management of infantile bilateral partial cataract

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Abstract

Purpose **To examine the data on outcome of surgery performed over a wide age range in members of a family affected by familial infantile bilateral partial cataract, with the purpose of assessing their predictive value concerning the timing of surgery.**

Methods **A retrospective clinical study was carried out of a family with dominant inheritance of familial infantile bilateral partial cataract. The family spanned four generations and consisted of 53 members, 31 of whom were examined in our department. Of these, 18 were affected. Cataract surgery was performed in 26 eyes of 15 patients, whose ages ranged from 6 to 58 years at the time of operation. As the surgical procedures spanned the years from 1978 to 1996, different techniques were used.**

Results **In 24 eyes (92%) the post-operative visual acuity was 6/9 or better. One eye achieved 6/12 and another 6/15.**

Conclusions **In this particular family there was no relationship between the post-operative visual acuity and the age at which surgery was performed. In deciding when to operate on family members with infantile bilateral partial cataract with similar morphology, in addition to the commonly used criteria the family data should also be taken into account. In infants and young children, delaying surgery may allow better development of visual acuity aided by accommodation, stabilisation of binocularity and more precise determination of the power of an intraocular lens. Success of very early surgery in such cases may not be attributable to the timing of the operation.**

Key words Bilateral, Cataract, Familial, Infantile

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Early surgery is accepted as the best way to prevent intractable amblyopia in patients with complete congenital cataract.[1–5] However, in cases of partial lens opacification in patients with congenital and early infantile cataracts, the optimal timing of surgery is still a subject of controversy.[1,6–8]

We describe a family with dominantly inherited infantile bilateral partial cataract. Affected family members underwent cataract surgery at different ages. The clinical histories and post-operative visual acuity results of different members of this family may provide relevant information concerning the appropriate time at which other affected family members should undergo cataract surgery in the future. Our findings may also be applicable to other similar cases.

Subjects and methods

A family spanning four generations consisted of 53 members (Fig. 1), of whom 31 were examined in our department. Of these, 18 (8 females and 10 males) were affected, and constitute the group upon which this study is based. All had bilateral partial cataract. Autosomal dominant inheritance is evident from the pedigree shown in Fig. 1. A consanguineous marriage of first cousins had occurred in the second generation (II-5; II-6).

Between May 1978 and July 1996, 31 members of the affected family were examined in the Eye Department of the Kaplan Medical Center. A review of their records revealed that apart from bilateral partial lens opacities in 18 individuals, there was no other eye pathology. A total of 26 cataract procedures were performed in 15 affected patients (Table 1). Our findings in the right eye of patient II-5 were not included in the study because this eye had been operated on elsewhere.

At the first examination all patients underwent a complete ocular evaluation. At the last pre-operative examination (Table 1) all patients were already mature enough to be tested by Snellen chart. The best corrected visual acuity on initial examination varied from 6/6 to 6/90. At the last pre-operative

Table 1. *Age at surgery, visual acuity and operative procedures in affected family members*

Patient[a]	Eye	Age at surgery (years)	Pre-operative VA	Post-operative VA	Post-operative follow-up (years)	First surgical procedure	Second surgical procedure
I-1	RE	–	6/90	–	–	Not operated	
	LE	–	6/90	–	–	Not operated	
II-1	RE	–	6/12	–	–	Not operated	
	LE	41	6/15	6/12	2	ECCE + PC IOL	Nd:YAG laser
II-5	RE	U	U	6/12	–	Operated on in another hospital	
	LE	40	U	6/9	10	ECCE + PC IOL	
II-6	RE	–	6/21	–	–	Not operated	
	LE	–	6/21	–	–	Not operated	
II-8	RE	–	U	–	–	Not operated	
	LE	58	6/60	6/9	1	ECCE + PC IOL	
II-9	RE	49	6/21	6/9	0.2	ECCE + PC IOL	
	LE	43	6/15	6/9	6	ECCE + PC IOL	
III-1	RE	19	6/15	6/9	3	PP lensectomy; ant. vit.; post. caps.	
	LE	18	6/21	6/9	4	PP lensectomy; ant. vit.; post. caps.	
III-13	RE	–	6/12	–	–	Not operated	
	LE	22	6/15	6/9	1	Ant. caps.; lens asp.; PC IOL	
III-15	RE	6	CF 3 m	6/7.5	13	Ant. caps.; lens asp. (Scheie)	Nd:YAG laser
	LE	15	CF 1 m	6/15	4	Ant. caps.; lens asp.; PC IOL	
III-16	RE	11	6/15	6/7.5	10	Ant. caps.; lens asp.; PC IOL	
	LE	17	6/21	6/9	4	Ant. caps.; lens asp.; PC IOL	Nd:YAG laser
III-17	RE	11	6/21	6/7.5	2	Ant. caps.; lens asp.; PC IOL	
	LE	7	6/15	6/7.5	4	Ant. caps.; lens asp.; PC IOL	
III-18	RE	9	6/21	6/9	5	Ant. caps.; lens asp.; PC IOL	PP ant. vit.; post. caps.
	LE	13	6/15	6/9	0.6	Ant. caps.; lens asp.; PC IOL	Nd:YAG
III-20	RE	17	6/60	6/9	7	PP lensectomy; ant. vit.; post. caps.	
	LE	17	6/15	6/9	7	PP lensectomy; ant. vit.; post. caps.	
III-21	RE	28	6/60	6/7.5	0.6	Ant. caps.; lens asp.; PC IOL	
	LE	21	6/21	6/7.5	7	Ant. caps.; lens asp.; PC IOL	Nd:YAG laser
III-23	RE	17	6/21	6/9	2	Ant. caps.; lens asp.; PC IOL	
	LE	25	6/18	6/9	0.2[b]	Ant. caps.; lens asp.; PC IOL	
III-27	RE	9	6/30	6/7.5	7	Ant. caps.; lens asp.; PC IOL	
	LE	14	6/30	6/9	2	Ant. caps.; lens asp.; PC IOL	
IV-2	RE	6	6/15	6/7.5	1.5	Ant. caps.; lens asp.; PC IOL	PP ant. vit.; post. caps.
	LE	7	6/18	6/9	1	Ant. caps.; lens asp.; PC IOL	PP ant. vit.; post. caps.
IV-4	RE	–	6/10	–	–	Not operated	
	LE	–	6/10	–	–	Not operated	

VA, visual acuity; RE, Right eye; LE, left eye; U, unknown; ECCE, extracapsular cataract extraction; PC IOL, posterior chamber intraocular lens implantation; PP, pars plana; ant., anterior; post., posterior; vit., vitrectomy; caps., capsulectomy or capsulorrhexis; asp., aspiration.

[a]See Fig. 1.

[b]Currently under follow-up.

examination, visual acuity varied from 6/15 to counting fingers at 1 m. None of the patients had nystagmus.

All patients had central bilateral posterior subcapsular lens opacities of 2.5 to 3.5 mm in diameter, located at the visual axis (Fig. 2). None of the patients had lenticonus. In some cases a translucent circular or star-shaped area of varying extent was visible at the periphery of the central opacity. The density of the lens opacity increased with age. Fig. 2 shows the morphology of lens opacity in three different patients at different ages. Progression was relatively slow over the years in all patients except for one (III-15), in whom the cataract progressed rapidly and led to remarkably poor vision (counting fingers at 3 m) in the first years of life (Table 1). This patient was also affected by systemic disorders. He suffered from haemophilia A and coeliac disease, and died at the age of 22 years from acquired immune deficiency syndrome.

The criteria for surgery were the following: (1) visual acuity – surgery was not performed if visual acuity was better than 6/15; (2) individual needs; (3) the patient's desire to achieve an improvement in visual functions. Assessment of visual functions and criteria for surgery in preverbal and non-compliant children were based on the fixation pattern and the morphology and location of the cataract, as well as on the amount of opacity observed on slit-lamp examination, the intensity of the fundal red reflex observed by retinoscopy and fundus visualisation by direct and indirect ophthalmoscopy. Age-related visual performance and behavioural observations were also taken into account in deciding on the timing of surgery.

Patients' ages at the time of surgery ranged from 6 to 58 years (mean 20.7, median 17 years) (Table 1). The earliest diagnosis of cataract was at 13 months and the oldest patient was diagnosed at the age of 58 years. The pre-operative follow-up period ranged from 1 week to 13 years. The mean pre-operative period from initial examination to first eye operation was 4.2 years. In cases of bilateral surgery, the time between operations ranged from 2 months to 9 years. Post-operative follow-up

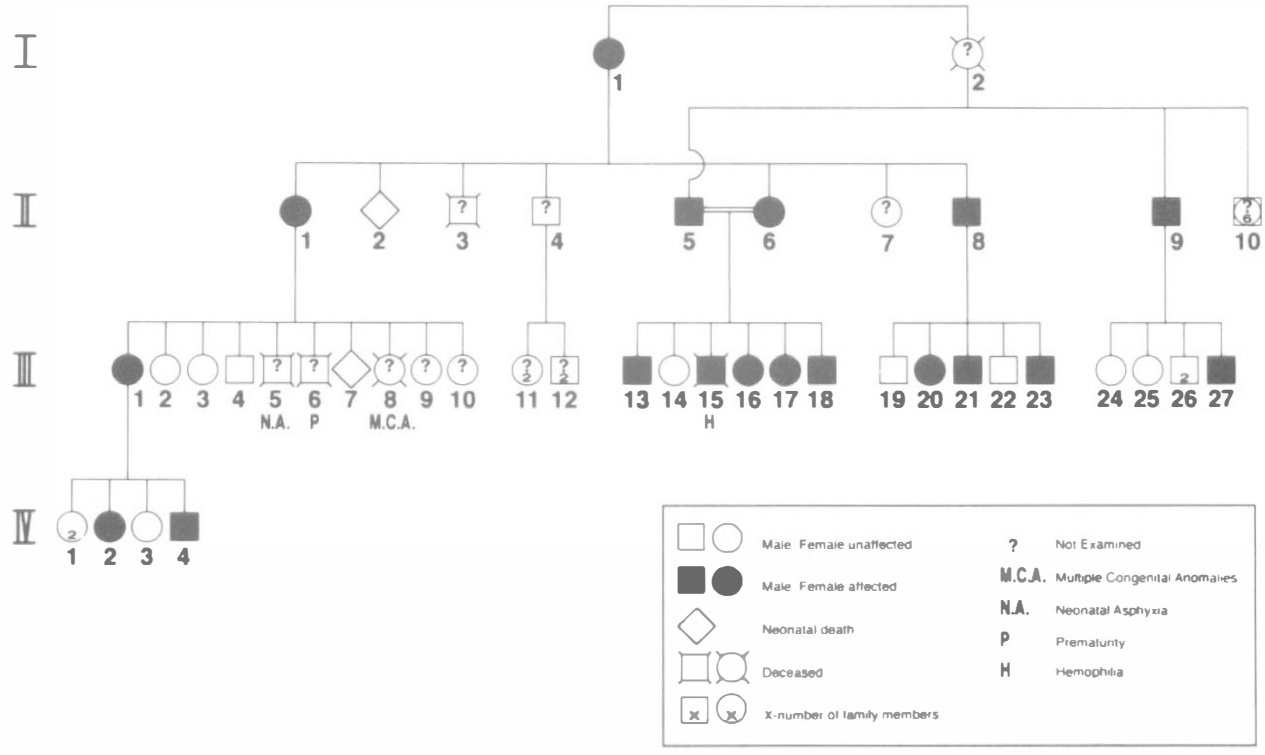

Fig. 1. *Pedigree of the affected family.*

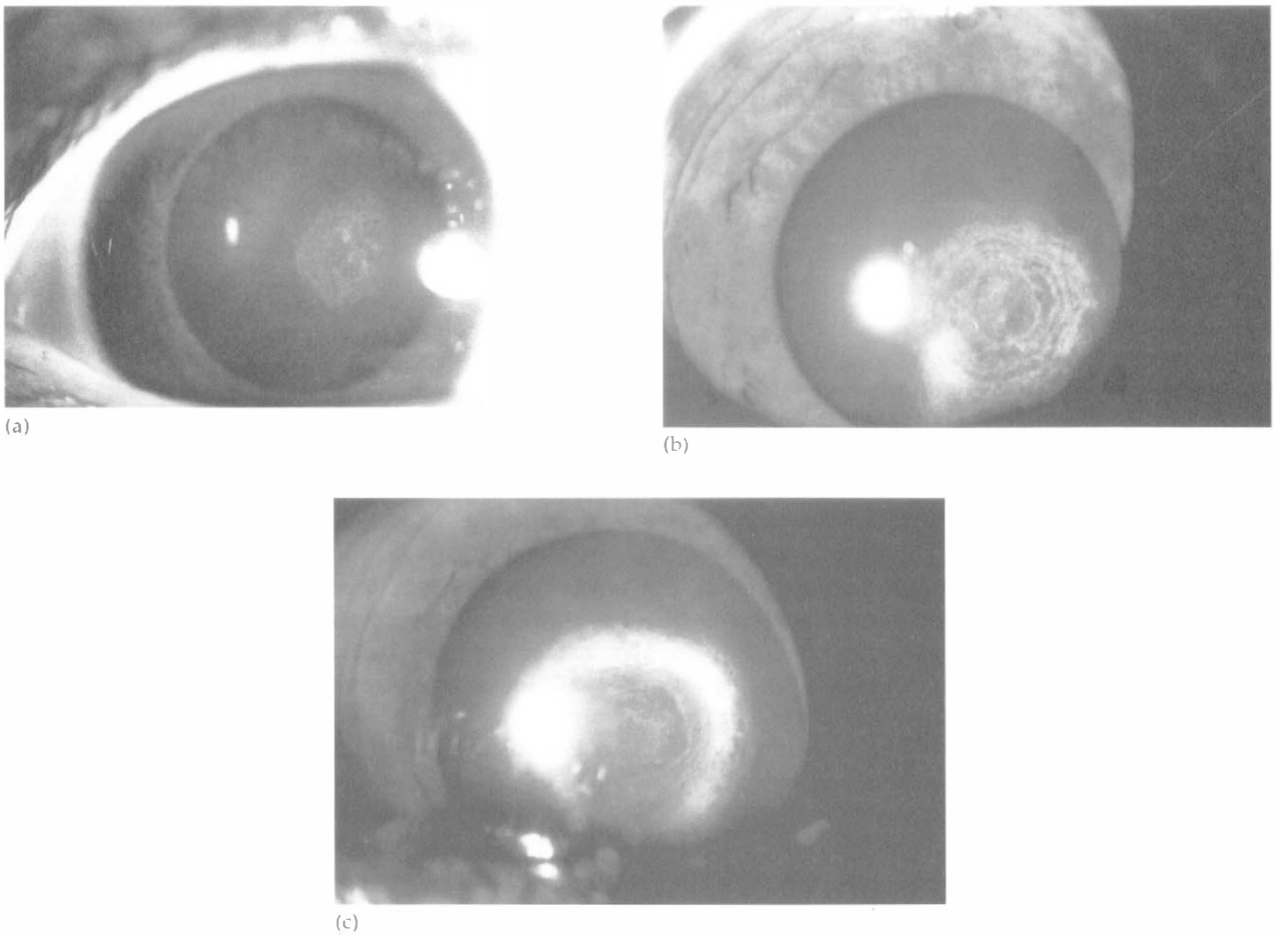

Fig. 2. *Cataract morphology in three patients, showing an increase in density with age. (a) Right eye of patient IV-4 at the age of 4 years; (b) right eye of patient III-18 at the age of 9 years; (c) left eye of patient III-16 at the age of 17 years.*

ranged from 2 months to 13 years (mean 4 years). Patients under the age of 18 years were followed up for at least 6 months.

As the operations on these 26 eyes spanned the years from 1978 to 1996, different surgical techniques were used, as detailed in Table 1.

Results

Of the 26 operated eyes, 24 (92%) achieved a post-operative visual acuity of 6/9 or better. Of the remaining 2 eyes, one (II-1, LE) achieved a final visual acuity of 6/12, and the other (the second eye of another patient, III-15, LE) of 6/15. The fellow eye of this latter patient had been operated on 9 years earlier when he was 6 years old. There was no relationship between post-operative visual acuity and patient age at surgery.

In all patients, with the exception of patient IV-2, good ocular alignment was maintained both before and after surgery. Patient IV-2 has an intermittent basic exotropia with stereopsis of 80′ as measured by the Titmus stereoacuity test.

Discussion

The treatment of infantile partial cataract is often problematic for paediatric ophthalmologists because the decision as to the optimal timing of surgery depends on different and frequently equivocal indications. In the past a conservative approach was generally adopted.[10–12] Later there was a shift towards early surgical intervention, mainly on the basis of visual development studies[4,13–17] as well as significant improvements in contact lens technology[18–21] and the safety of surgical techniques.[9,22–24] Very early surgery is indicated in cases of dense bilateral congenital cataract. A moderately cloudy unilateral cataract also needs a more aggressive approach, because of the higher risk of development of intractable amblyopia.[1–3] However, in children with bilaterally equal partial lens opacification and visual acuity, the risks of developing abnormal visual functions are smaller.

In verbal children, a visual acuity of 6/21 or worse has been accepted as an indication for surgery. However, this should not be considered as an absolute indication for intervention, since many authors have reported excellent visual rehabilitation after aspiration with posterior chamber intraocular lens (IOL) implantation in children.[25–28] A disadvantage of surgery in early infancy is the unavoidable consequence of accommodation loss, which probably has a critical influence on the development of visual acuity. Early surgery may also disrupt binocular vision.[29,30] Moreover, since IOL implantation is becoming widely accepted in infants and children, the growth of the infant eye is an additional factor in favour of delaying surgery, because the changing refractive power of the eye in the first years of life may, with time, induce an overcorrected refractive error. Surgery has its attendant short-term and long-term complications and is more hazardous in infants and young children than in older children.

The series of cases in this report provides an overview of the operative results in family members whose ages differed widely at the time of surgery. The youngest patient was operated on at the age of 6 years and the oldest at 58 years. All of the affected family members had a history of visual difficulties from early childhood. Moreover, all the cataracts were bilateral and were very similar in morphological appearance, consisting of a central posterior polar opacity located at the visual axis (Fig. 2), and having a density directly related to age.

We never examined newborns in the affected families. Moreover, only a minority of the patients were examined very early in life, when much of the critical patterning of visual perception is developed. The earliest age at which a patient with cataract was examined and diagnosed was 13 months (III-15). Two patients (III-23; IV-4) were seen and diagnosed at the age of 4 years; all other patients in this family were first examined at older ages.

As shown in Table 1, 24 of the 26 operated eyes (92%) achieved a post-operative visual acuity of 6/9 or better. Eight eyes achieved a visual acuity of 6/7.5, 16 eyes achieved 6/9, one eye 6/12 and one eye 6/15. After this last patient (III-15) underwent surgery in his right eye at the age of 6 years, his visual acuity went from counting fingers at 3 m pre-operatively to 6/7.5. Because of his poor general condition (he suffered from haemophilia A and coeliac disease), there was a delay of 9 years before the second eye was operated on. As his parents (II-5; II-6) were first-degree cousins, and both had bilateral cataracts, there was a 25% probability that he would be homozygotic for the dominant mutation, inheriting one mutated allele from each parent. This patient died before this hypothesis could be tested. Theoretically, any of these factors might explain the very early maturation of his cataracts, unlike the relatively slow progression observed in other members of the family. Since a complete late post-operative examination, including fundal examination, was without any positive findings, we cannot exclude the possibility of mild amblyopia in this eye.

Dense axial lens opacities have been shown to be at least potentially amblyogenic.[1,7] The axial lens opacities seen in infants in this study were of relatively mild density, which may be a reason for the apparent absence of amblyopia. It is possible that the visual axis may have been clear during the critical period for the appearance of deprivation amblyopia.[4]

Merin and Crawford[7] have emphasised the need for an objective way to analyse and grade the severity of partial cataracts. With regard to amblyopia, Freeman and Lovick[6] suggest the use of diagnostic technologies such as A-scan ultrasonography, standardised pre-operative photography, fluorescein angiography and electrophysiological monitoring, which may contribute to our assessment of the severity of the amblyogenic factors in such cases. These authors proposed a large collaborative study, with the object of identifying factors

predictive of good vision after cataract surgery. The report of Freeman and Lovick[6] as well as the present report and our knowledge of other similar unreported families, may support the predictive value of family data concerning cataract morphology, clinical findings, and pre- and post-operative visual acuity. The findings of the present study, in which cataract surgery was performed over a wide age range, led us to conclude that in this family there was little or no evidence that failure to operate during the first years of life was harmful to the development of visual functions. It is possible that these findings may be applicable also to other families and that better final results may be obtained if, early in life, a child is allowed to develop normal vision with the fine and accurate tuning mechanism of accommodation. Furthermore, delay of surgery probably results in better stabilisation of binocular vision and enables more precise evaluation of the dioptric power of the implanted IOL, with better approximation to emmetropia in later life.

In deciding when to schedule surgery for patients with familial infantile bilateral partial cataract, the family data on cataract management can be of substantial help. Paediatric ophthalmologists should consider the possibility that when operations are performed in infancy or early childhood, their success may not be attributable to the early time of surgery.

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