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# Evaluation of functional vision and eye-related quality of life in children with congenital ectopia lentis: a prospective cross-sectional study

Xiaolin Liang,[1] Danying Zheng ,[1] Charlotte Aimee Young,[2] Yiyuan Ma,[1] Lirong Ling,[1] Minjie Zou ,[1] Siyuan Liu,[1] Xinyu Zhang,[1] Guangming Jin [1]

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[1]State Key Laboratory of Ophthalmology, Zhongshan Ophthalmic Center, Sun Yat-sen University, Guangdong Provincial Key Laboratory of Ophthalmology and Visual Science, Guangdong Provincial Clinical Research Center for Ocular Diseases, Guangzhou, China
[2]Albany Medical College, Albany, New York, USA

**Correspondence to**
Dr Guangming Jin; guangming27050103@126.com and
Dr Xinyu Zhang; zhangxinyu0294@163.com

## ABSTRACT

**Objectives** This study aims to evaluate the effect of congenital ectopia lentis (CEL) on functional vision and eye-related quality of life (ER-QOL) in children and their families using the Paediatric Eye Questionnaire (PedEyeQ).

**Design** A questionnaire survey administered via in-person interviews of patients with CEL and their parents.

**Participants** 51 children with CEL and 53 visually normal controls accompanied by 1 parent completed the survey questionnaires for the study from March 2022 to September 2022.

**Outcome measures** PedEyeQ domain scores. Functional vision and ER-QOL of children and their families were evaluated by calculating and comparing the Rasch domain scores of the PedEyeQ.

**Results** PedEyeQ domain scores were significantly worse with CEL compared with controls (p<0.01 for each), with the exception of the Proxy Social domain among children aged 0–4 years (p=0.283). Child PedEyeQ greatest differences were in the functional vision domain (5–11 years, −20 points (95% CI −27 to −12)) and frustration/worry domain (12–17 years, −41 (95% CI −37 to −6)). Proxy PedEyeQ greatest differences were in the functional vision domain (0–4 years, −34 (95% CI −45 to −22)) and frustration/worry domain (5–11 years, −27 (95% CI −39 to −14); 12–17 years, −37(95% CI (−48 to −26))). Parent PedEyeQ greatest difference was in the 'worry about child's eye condition' (−57 (95% CI (−63 to −51))).

**Conclusions** In this study, children with CEL had reduced functional vision and ER-QOL compared with controls. Parents of children with CEL also experience reduced quality of life.

## STRENGTHS AND LIMITATIONS OF THIS STUDY

⇒ This study used an appropriate questionnaire to develop in-depth insight into the quality of life issues experienced in congenital ectopia lentis (CEL) from the perspectives of children and their parents or legal guardians.
⇒ The Paediatric Eye Questionnaire used in this study is a comprehensive instrument designed to simultaneously evaluate the functional vision and eye-related quality of life in children with eye conditions.
⇒ A wide range of age groups of CEL patients were included and comparative analysis was conducted among these age groups.
⇒ The in-person interviews were conducted in a large-scale ophthalmology hospital in China, which might result in potential sample selection bias.
⇒ As a single-centre study, selection biases may exist in this study.

## INTRODUCTION

Congenital ectopia lentis (CEL) is defined as the displacement of the crystalline lens from its normal position caused by congenital dysplasia of the zonule. The prevalence rate of CEL is approximately 6.4/10 million.[1] CEL may present with symptoms such as blurred vision, fluctuating vision and monocular diplopia,[2–4] and can also occur in conjunction with abnormalities in the skeletal and cardiovascular system, as seen in Marfan's syndrome (MFS), homocystinuria and Weill-Marchesani syndrome.[5–7] MFS accounts for the largest proportion of systemic diseases in CEL patients, while cardiac manifestations associated with MFS can potentially lead to death.[8–10] Additionally, surgical intervention and long-term follow-up visits are usually needed for patients with CEL, which may affect their daily lives with mental health repercussions.[11 12] Unfortunately, objective clinical examinations provide only a limited measure of visual performance and may not capture the full impact of eye diseases on children and their families. Thus, patient-reported outcome measures in paediatric eye conditions are widely advocated for.[13]

In the previous studies, a series of questionnaires has been designed to evaluate the impact of eye diseases on patients. Khadka *et al* developed the Cardiff Visual Ability Questionnaire for Children to assess self-reported visual ability in children and young people with visual impairment.[14] The LV

Prasad-Functional Vision Questionnaire can also serve as a valid measure of self-reported functional vision performance for visually impaired children.[15–17] However, these questionnaires were specifically designed for children with vision impairment but not for other eye conditions. Reduced quality of life (QOL) was found among children with different ophthalmic disorders using the PedsQL (Paediatric Quality of Life Inventory),[18–20] a generic questionnaire that systematically assesses the health-related QOL (HRQOL) of paediatric patients with chronic conditions,[21] but the PedsQL is reported to be less sensitive to the concerns of children with eye diseases compared with newer eye-specific questionnaires.[22 23] Recently, Hatt *et al* developed the Paediatric Eye Questionnaire (PedEyeQ) to assess functional vision and eye-related QOL (ER-QOL) in children with eye conditions and QOL of parents.[24 25] PedEyeQ is a validated questionnaire that contains age-specific questions to evaluate the impact of eye diseases on daily life and well-being from the child's and parent's perspectives. Additionally, this survey has a high quantity of question items than most other measures and can simultaneously examine the two broad areas of functional vision and ER-QOL in children of any age with any eye condition.

To date, the majority of prior studies on the association between childhood eye diseases and ER-QOL mainly focused on common diseases, such as congenital cataract, refractive errors, strabismus and amblyopia, but few studies have focused on eye diseases with systemic abnormalities such as CEL.[22 23 26 27] As a progressive disease, CEL could lead to visual acuity defects at an early age and subsequently develop cardiovascular, skeletal and other systemic complications. Though many previous studies have reported the treatment strategies for this disease, the life and psychological burden of patients and their families caused by long-term follow-up visits and worries about the eye condition is rarely reported. Therefore, we conducted this study to evaluate the functional vision and ER-QOL in children with CEL, aiming to raise ophthalmologists' attention to the mental health impact of CEL on children and their families. We hope our findings will be impactful to the proposal and development of improved treatment strategies.

## METHODS

### Subjects

Participants, accompanied by one parent or legal guardian, were prospectively recruited both in outpatient and inpatient ward from March 2022 to September 2022. Patients were enrolled in this study according to the following inclusion criteria: (1) be diagnosed as CEL; (2) under 18 years old and (3) be able to complete the survey questionnaire. Exclusion criteria were as follows: (1) a history of ocular surgery (incisional or laser); (2) lens dislocation caused by trauma or other reasons; (3) those with cataract, secondary glaucoma, strabismus or other associated ocular complications except refractive error and (4) subjects who were unable to respond to or complete the survey. Additionally, age-matched and sex-matched individuals, with normal visual acuity,[28 29] no visual disorders except mild refractive error, and no history of ophthalmic surgery were prospectively enrolled as the control group.

### The Paediatric Eye Questionnaire

The PedEyeQ was developed to assess the functional vision and ER-QOL in children with any eye conditions and their parents.[24 25] The questionnaire is composed of three components (child, proxy and parent), each with distinct, separately scored domains, using a three-point frequency scale for responses ("never', 'sometimes' and all of the time'). This instrument has different versions according to the child's age. The Child PedEyeQ component has two versions (5–11 or 12–17 years old versions) and is completed by the children. The Proxy and Parent PedEyeQ components have three versions (0–4, 5–11 or 12–17 years old versions) and are answered by one parent or caregiver of each child. The Child 5–11 and 12–17 PedEyeQ consists of 'functional vision', 'bothered by eyes/vision', 'social' and 'frustration/worry' domains. The Proxy 0–4 PedEyeQ consists of three domains: 'functional vision', bothered by eyes/vision' and 'social'. The Proxy 5–11 and 12–17 PedEyeQ consists of five domains: 'functional vision', 'bothered by eyes/vision', 'social', 'frustration/worry' and 'eye care'; and the Parent PedEyeQ assesses the effect of the child's eye condition on the parent and family in four domains: 'impact on parent and family', 'worry about child's eye condition', 'worry about child's self-perception and interactions' and 'worry about child's functional vision'.

### Statistical analysis

Statistical analyses were performed by using SPSS (V.25.0, IBM). For each participant, on each PedEyeQ domain, Rasch scores were calculated using previously published Rasch lookup tables and converted to a scale from 0 (worst) to 100 (best). Domain scores were analysed by age group (0–4, 5–11 and 12–17) separately. Median scores were compared between CEL and control cohorts using Wilcoxon tests as data were not normally distributed. Values of $p \leq 0.05$ were considered statistically significant. Mean differences with a 95% CI were also calculated to represent the precision of our estimates. Spearman correlation analysis was used to assess the relationship between PedEyeQ Domain Scores and patients' best-corrected vision acuity (BCVA).

### Patient and public involvement

Patients and the public were not involved in the development of the research questionnaire, outcome measures, design, recruitment and implementation of the study. The results will be disseminated through scientific journals.

## RESULTS

### Demographics and clinical characteristics of participants

A total of 104 subjects were included in this study. Fifty-one children with CEL were included, of whom 14 (27.5%) were aged 0–4 years (median, 3.6 years), 28 (54.9%) were 5–11 years (median, 7.5 years) and 9 (17.6%) were aged 12–17 years (median, 14.6 years). Fifty-three children with normal visual acuity were then enrolled as a normal control group. Seventeen of 53 (32.1%) were aged 0–4 years (median, 3.5 years), 25 (47.2%) were aged 5–11 years (median, 7.6 years) and 11 (20.8%) were aged 12–17 years (median, 14 years). Demographics and clinical characteristics for all enrolled children and their parents are shown in table 1 and online supplemental table 1.

**Table 1** Demographics and clinical characteristics of respondents (51 children with congenital ectopia lentis and their parents)

| Demographic characteristics | Age 0–4 years (n=14), No (%) | Age 5–11 years (n=28), No (%) | Age 12–17 years (n=9), No (%) |
|---|---|---|---|
| Sex | | | |
| Female | 9 (64) | 9 (32) | 6 (67) |
| Sibling relationships | | | |
| Only child | 3 (21) | 4 (14) | 2 (22) |
| Parent/legal guardian completing questionnaires | | | |
| Father | 5 (36) | 6 (21) | 3 (33) |
| Mother | 9 (64) | 22 (79) | 6 (67) |
| Legal guardian | 0 (0) | 0 (0) | 0 (0) |
| Parent/legal guardian age, years | | | |
| Under 21 | 0 (0) | 0 (0) | 0 (0) |
| 21–30 | 4 (29) | 5 (18) | 0 (0) |
| 31–40 | 8 (57) | 17 (61) | 2 (22) |
| 41–50 | 2 (14) | 5 (18) | 7 (78) |
| 51–60 | 0 (0) | 1 (4) | 0 (0) |
| Older 60 | 0 (0) | 0 (0) | 0 (0) |
| Parent/legal guardian highest level of education | | | |
| Primary school graduate | 0 (0) | 2 (7) | 1 (11) |
| Junior-high graduate/technology secondary school graduate | 1 (7) | 11 (39) | 4 (44) |
| High school graduate/junior college degree | 5 (36) | 10 (36) | 3 (33) |
| College graduate | 2 (14) | 5 (18) | 0 (0) |
| Postgraduate/professional degree | 0 (0) | 0 (0) | 0 (0) |
| Not reported | 0 (0) | 0 (0) | 1 (11) |
| Annual household income | | | |
| Under 30 000 | 3 (21) | 5 (18) | 3 (33) |
| 30 000–80 000 | 4 (29) | 10 (36) | 3 (33) |
| 80 000–300 000 | 6 (43) | 10 (36) | 3 (33) |
| ver 300 000 | 1 (7) | 2 (7) | 0 (0) |
| Not reported | 0 (0) | 1 (4) | 0 (0) |
| Previous treatment | | | |
| No previous treatment | 6 (43) | 6 (21) | 0 (0) |
| Glasses wearing | 2 (14) | 20 (71) | 7 (78) |
| Amblyopia treatment | 1 (7) | 6 (21) | 3 (33) |
| BCVA in better eye (logMAR) | 0.6±0.4 | 0.2±0.3 | 0.2±0.2 |
| BCVA in worse eye (logMAR) | 0.8±0.5 | 0.4±0.4 | 0.5±0.5 |

BCVA, best-corrected vision acuity; logMAR, logarithm of minimum angle of resolution.

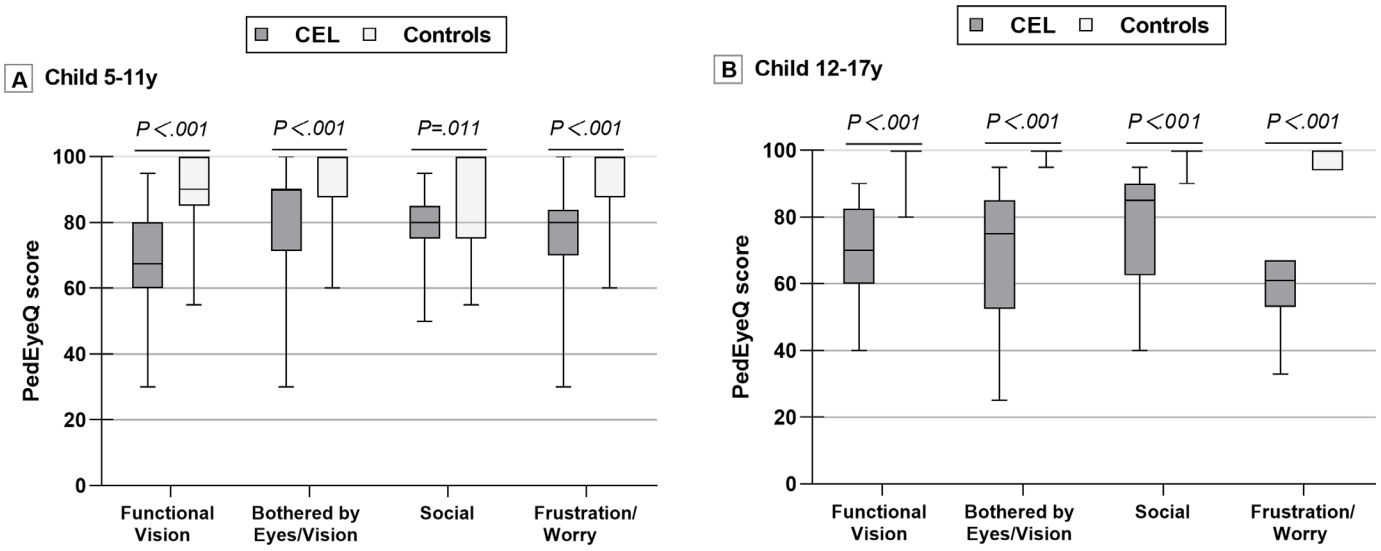


**Figure 1** Child Paediatric Eye Questionnaire (PedEyeQ) domain scores in children with CEL and in normal controls. (A) PedEyeQ scores for children aged 5–11 years (functional vision, bothered by eyes/vision, social and frustration/worry). (B) PedEyeQ scores for children aged 12–17 years (functional vision, bothered by eyes/vision, social and frustration/worry). The boxes represent the first quartile, median and third quartile values; whiskers represent extreme values. CEL, congenital ectopia lentis.

### Comparing child PedEyeQ scores in children with CEL and normal controls

Child PedEyeQ scores were lower with CEL compared with visually normal controls for all domains (figure 1A,B). The greatest difference among children aged 5–11 years was in 'functional vision' and among children aged 12–17 years was in 'frustration/worry' (p<0.001 for each domain, table 2).

### Comparing proxy PedEyeQ scores in CEL and control cohorts

Using the Proxy PedEyeQ, CEL children had lower scores across all domains than visually normal controls (figure 2A–C), except in the 'social' domain in 0–4 years (p=0.283, online supplemental table 2). The greatest difference among children aged 0–4 years was in 'functional vision', among children aged 5–11 years was in 'frustration/worry', and among children aged 12–17 years was also in 'frustration/worry' (p<0.001 for each, online supplemental table 2).

### Comparing parent PedEyeQ scores in parents of CEL and control cohorts

Parents of CEL children had significantly negative PedEyeQ domain scores (figure 3). The greatest mean difference was in the 'worry about their child's eye condition' domain (p<0.001, table 2). We also found that parent PedEyeQ scores of those with children aged 0–4 years were generally lower than those of children aged 5–17 years in the 'worry about their child's eye condition' and 'worry about child's visual function' domains (p<0.05 for each, online supplemental table 2).

### Correlations between PedEyeQ domains and patients' BCVA by age group

All correlations are shown in online supplemental table 3. There were no associations between patients' BCVA with all PedEyeQ domain scores among children aged 0–4 years. Among children aged 5–11 years, there was a significant association between poor worse-eye's visual acuity and lower PedEyeQ score in only one of the four child domains (functional vision: −0.427, p<0.05). Among children aged 12–17 years, poor worse-eye's visual acuity was associated with lower PedEyeQ scores in all child domains (p<0.05 for each). A negative association was detected for the social domain both with better-eye's BCVA (r=−0.688, p<0.05) and with worse-eye BCVA (r=−0.753, p<0.05).

## DISCUSSION

This prospective study aimed to evaluate the impact of CEL on children and their parents, using the newly developed and validated instrument PedEyeQ. The results showed that according to the sell-reporting of children, the presence of CEL resulted in significantly poorer functional vision and ER-QOL (Child PedEyeQ). Additionally, results showed that older paediatric patients are more likely to be psychologically affected. By proxy report, we also found generally reduced functional vision and ER-QOL in CEL children compared with normal controls. Notably, there was no significant difference in the social domain of children aged 0–4 years from parents' perspective. In addition, parents of CEL patients also reported a reduced QOL when evaluating impact on the parents themselves (Parent PedEyeQ) and parents of children aged 0–4 years with CEL have generally lower PedEyeQ scores than those of the other two age groups.

We found reduced functional vision and ER-QOL in children with CEL using the Child component of the PedEyeQ. The functional vision domain reflects concerns about problems with seeing, learning, concentrating, schoolwork and sports (full questionnaires available at

Table 2 PedEyeQ domain scores for children with congenital ectopia lentis and visually normal control

| PedEyeQ domains (by age group for child and proxy) | Median (range) PedEyeQ scores | | P value for difference, median scores | Mean difference with 95% CI |
|---|---|---|---|---|
| | CEL | Controls | | |
| Child 5–11 years | **N=28** | **N=25** | | |
| Functional vision | 67 (30–95) | 90 (55–100) | < 0.001 | –20 (–27 to –12) |
| Bothered by eyes/vision | 90 (30–100) | 100 (60–100) | < 0.001 | –13 (–21 to –5) |
| Social | 80 (50–95) | 100 (55–100) | 0.01 | –9 (–17 to –1) |
| Frustration/worry | 80 (30–100) | 100 (60–100) | < 0.001 | –16 (–23 to –9) |
| Child 12–17 years | N=9 | N=11 | | |
| Functional Vision | 70 (40–90) | 100 (80–100) | < 0.001 | –27 (–39 to –15) |
| Bothered by eyes/vision | 75 (25–95) | 100 (95–100) | < 0.001 | –31 (–48 to –14) |
| Social | 85 (40–95) | 100 (90–100) | < 0.001 | –21 (–37 to –6) |
| Frustration/worry | 61 (33–67) | 100 (94–100) | < 0.001 | –41 (–37 to –6) |
| Proxy 0–4 years | N=14 | N=17 | | |
| Functional vision | 52 (30–80) | 100 (55–100) | < 0.001 | –34 (–45 to –22) |
| Bothered by eyes/vision | 55 (25–90) | 100 (50–100) | 0.002 | –24 (–39 to –9) |
| Social | 89 (44–100) | 94 (50–100) | 0.283 | –2 (–17 to –13) |
| Proxy 5–11 years | N=28 | N=25 | | |
| Functional vision | 65 (25–100) | 95 (50–100) | 0.001 | –19 (–29 to –9) |
| Bothered by eyes/vision | 75 (40–100) | 100 (50–100) | 0.003 | –13 (–22 to –3) |
| Social | 69 (44–100) | 100 (50–100) | 0.01 | –12 (–24 to –1) |
| Frustration/worry | 60 (10–100) | 100 (50–100) | < 0.001 | –27 (–39 to –14) |
| Eye care | 58 (42–100) | 92 (50–100) | < 0.001 | –21 (–30 to –12) |
| Proxy 12–17 years | N=9 | N=11 | | |
| Functional vision | 65 (35–85) | 100 (85–100) | < 0.001 | –30 (–42 to –18) |
| Bothered by eyes/vision | 60 (45–100) | 100 (95–100) | < 0.001 | –35 (–50 to –21) |
| Social | 81 (38–100) | 100 (75–100) | 0.001 | –23 (–37 to –9) |
| Frustration/worry | 62 (44–94) | 100 (94–100) | < 0.001 | –37 (–48 to –26) |
| Eye care | 67 (58–92) | 100 (83–100) | < 0.001 | –23 (–32 to –14) |
| Parent | N=51 | N=53 | | |
| Impact on parent/family | 60 (10–100) | 100 (50–100) | < 0.001 | –30 (–37 to –23) |
| Worry about child's eye condition | 45 (0–80) | 100 (65–100) | < 0.001 | –57 (–63 to –51) |
| Worry about child's self-perception and interactions | 64 (21–100) | 100 (50–100) | < 0.001 | –34 (–41 to –28) |
| Worry about child's visual function | 50 (0–100) | 100 (50–100) | < 0.001 | –47 (–53 to –40) |

CEL, congenital ectopia lentis; PedEyeQ, Paediatric Eye Questionnaire.

https://public.jaeb.org/pedig/view/Other_Forms).[23] Our results are consistent with previous studies involving children with visual impairment and confirm the presence of functional impairment experienced by CEL patients, showing poorer reading speed and reduced physical competence.[15 23] Additionally, we directly measured child-reported ER-QOL and found scores were significantly lower across 'bothered by eyes/vision', 'social' and 'frustration/worry' domains compared with visually normal controls, confirming that children with lens dislocation experience a range of QOL concerns in their everyday life. Handisides *et al* have also reported lower overall, psychosocial and physical HRQOL among children and adolescents with MFS using the PedsQL Generic Core Scales.[30] Notably, apart from MFS patients, there are still part of CEL children experiencing ocular and other systematic abnormalities. Nevertheless, the association between CEL and QOL has not yet been well studied. Our findings are important in confirming the reduced QOL of CEL patients, especially in the psychological domain. CEL may have more adverse effects on patients compared with other eye diseases since in addition to ocular impairment, CEL patients may have skeletal abnormalities that may lead them to feel isolated, discriminated against or socially devalued.[31–33] What's worse, CEL can be a progressive, life-threatening disorder since cardiac

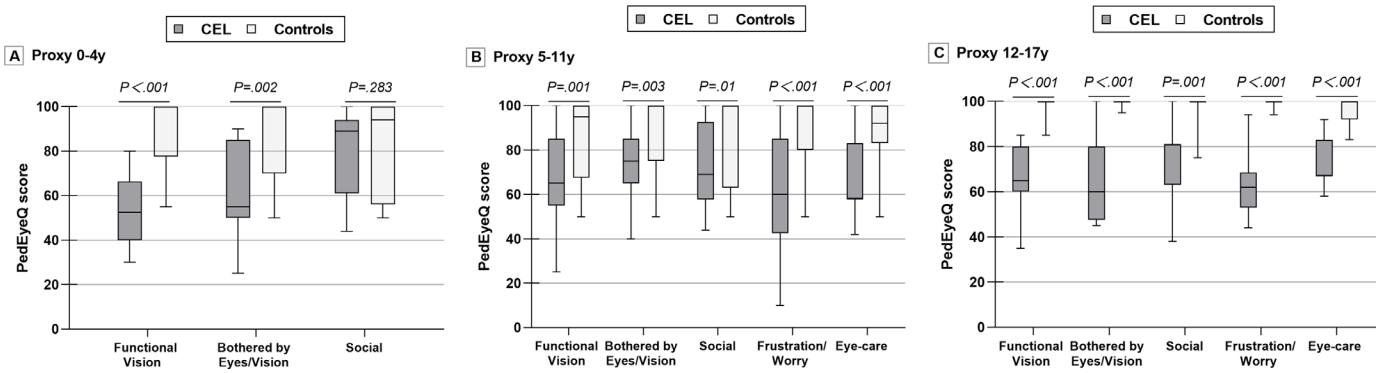


**Figure 2** Proxy Paediatric Eye Questionnaire (PedEyeQ) domain scores in children with CEL and in normal controls. (A) Proxy PedEyeQ scores for children aged 0–4 years (functional vision, bothered by eyes/vision and social). (B) Proxy PedEyeQ scores for children aged 5–11 years (functional vision, bothered by eyes/vision, social, frustration/worry and eye care). (C) Proxy PedEyeQ scores for children aged 12–17 years (functional vision, bothered by eyes/vision, social, frustration/worry and eye care). The boxes represent the first quartile, median and third quartile values; whiskers represent extreme values. CEL, congenital ectopia lentis.

complications are the leading causes of mortality,[9 34] which puts paediatric patients at much higher risk for anxiety or depression.[35]

Regarding proxy report, we also found CEL significantly affected the functional vision and ER-QOL of paediatric patients, as was paralleled by self-reporting children. This finding indicates that parents/guardian could be aware of difficulties encountered by their children and they play a key role in observing daily activities of CEL patients. Proxy evaluation of a child's QOL remains an important approach of assessment, especially in young children where self-reporting may be limited. However, in our current study, there was no statistical difference between CEL and control cohorts in the social domain among 0–4 years. In contrast, Leske *et al* detected significant difference between children with variable ocular disorders and controls in this domain across 1037 children.[36] The discrepancy in results may be explained by the racial and ethnic differences between the study population. In addition, it is possible that children in 0–4 years category may be too young for parents to perceive they are affected socially by this disease.

Having a child with CEL also had a marked impact on parental QOL. Parents of CEL children had significantly negative PedEyeQ scores in the worry about their child's eye condition domain. We also found that parent

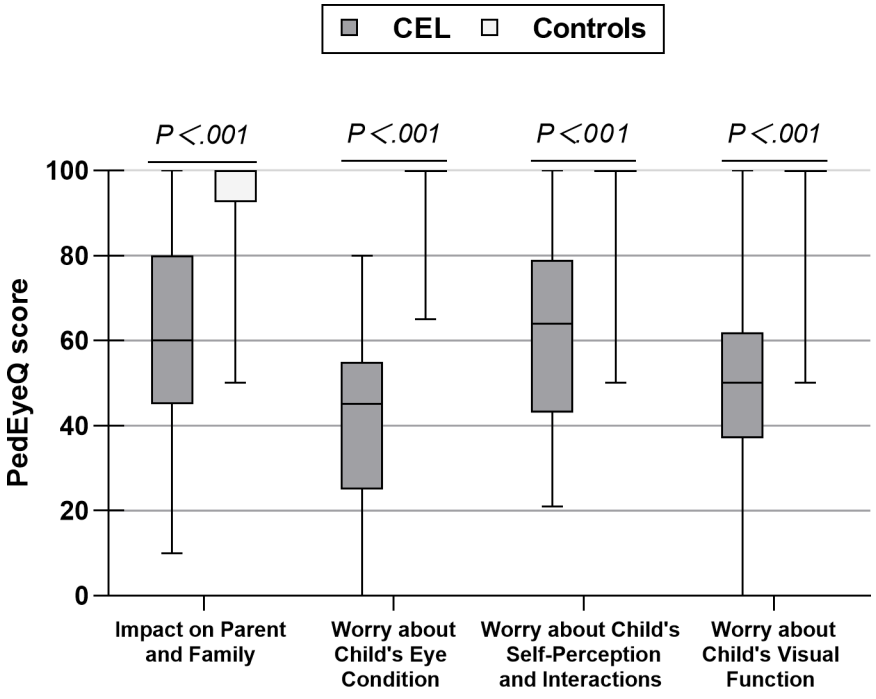


**Figure 3** Parent Paediatric Eye Questionnaire (PedEyeQ) domain scores in children with CEL and in normal controls. Parent PedEyeQ domains include the impact on parent and family, worry about child's eye condition, worry about child's self-perception and interactions, and worry about child's functional vision. The boxes represent first quartile, median and third quartile values; whiskers represent extreme values. CEL, congenital ectopia lentis.

PedEyeQ scores of those with children aged 0–4 years were generally lower than those of children aged 5–17 years. Specifically, the scores were significantly lower for parents of children aged 0–4 years with CEL in the 'worry about their child's eye conditions' domain. Our findings indicate that parents tend to overestimate the influence of CEL on their children as they may suffer from ocular as well as other systematic abnormalities. CEL patients especially those with cardiac complications are at risk of sudden death, which may be the cause of a higher degree of anxiety. Furthermore, it is likely that paediatric patients do not openly communicate with their parents about their school life, which then adds to the parents' psychological concerns with regard to their children's learning and social life. Moreover, parents of children diagnosed at a young age experience a higher financial burden, which may create additional stress on the parents. Finally, long-term expectations and their understanding of their children's health may also affect parents' QOL.

We found an overall association between poorer worse-eye's BCVA but not better-eye's BCVA and worse functional vision scores and with worse specific ER-QOL domain scores among CEL children aged 5–17. This result is partly in accordance with prior studies. Leske *et al* reported that both poorer better-eye and worse-eye's visual acuity were associated with functional vision and ER-QOL in children aged 5–11 with eye conditions.[37] Our finding may be explained by the fact that CEL is usually a binocular disease, poor eyes often develop ectopia lentis first and its impact occurs earlier and more significantly. Our findings highlight the importance of recognising the effect of worse-eye's visual acuity on QOL among CEL children and suggest that timely and appropriate treatment of the worse-eye may help improve functional vision and QOL effectively for CEL patients.

In this study, we used the recent age-specific, patient-derived PedEyeQ to investigate how CEL affects both paediatric patients and their caregivers. The PedEyeQ is a comprehensive instrument that has been developed to evaluate the impact on functional vision and ER-QOL in children of any age with any eye conditions. Additionally, it has more items than most other measures and can simultaneously evaluate respondents' functional vision and ER-QOL. Construct validity, reliability and responsiveness of the new instrument have been previously evaluated among children with bilateral visual impairment.[38] Moreover, the newly eye-specific PedEyeQ is tested to be more sensitive to the concerns of children with specific eye diseases compared with the more generic PedsQL.[22] Hence, chose to use PedEyeQ and included a wide range of age groups of CEL patients in our research. To our knowledge, the current study is the first to evaluate functional vision and ER-QOL in children with CEL and their parents using PedEyeQ.

There are limitations to this study. First, this study is a single-centre study, which may result in selection biases. Second, the sample size in 0–4years group was relatively small and to some extent, the accuracy of the results will be affected. However, despite the rarity of CEL, we have made every attempt to enrol individuals in this study, making it a relatively large sample. In addition, further work exploring associations with different clinical factors such as the degree of lens and treatment timing and methods remains a critical next step for future research.

In conclusion, children with CEL have reduced functional vision and ER-QOL compared with normal controls, as reported by the child themselves and by proxy reporters. Additionally, parents of CEL patients also experience lower QOL. In the management of CEL, we should comprehensively consider the impact of this disease on patients' QOL and propose improved treatment strategies. Children with CEL and their families can be expected to benefit from health education, clinical treatment and psychological intervention.

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**Contributors** XL, DZ, XZ and GJ contributed to the study conception and study design. Data collection, analysis and interpretation were undertaken by XL, LL, YM, MZ and SL. The first draft of the manuscript was written by XL and CAY. All authors commented on previous versions of the manuscript. All authors read and approved the final manuscript. DZ and GJ act as guarantors for this manuscript.

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**Competing interests** None declared.

**Patient and public involvement** Patients and/or the public were not involved in the design, or conduct, or reporting, or dissemination plans of this research.

**Patient consent for publication** Not applicable.

**Ethics approval** This study involves human participants and was approved by the Institutional Review Board of Zhongshan Ophthalmic Center in Sun Yat-sen University, Guangzhou, China (2022KYPJ207). Participants gave informed consent to participate in the study before taking part.

**Provenance and peer review** Not commissioned; externally peer reviewed.

**Data availability statement** Data are available on reasonable request. Requests can be submitted to the corresponding author.



**ORCID iDs**
Danying Zheng http://orcid.org/0000-0003-1315-7130
Minjie Zou http://orcid.org/0000-0001-7706-6663
Guangming Jin http://orcid.org/0000-0001-9994-6338

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**Supplemental Table1. Demographics of 53 Visually Normal Children Completing the Pediatric Eye Questionnaire, and Their Parents.**

| Demographic characteristics | Age 0-4 y (n=17), No. (%) | Age 5-11 y (n=25), No. (%) | Age 12-17 y (n=11), No. (%) |
|---|---|---|---|
| **Sex** | | | |
| Female | 8（47） | 9（36） | 7（64） |
| **Sibling Relationships** | | | |
| Only Child | 8（47） | 10（40） | 7（64） |
| **Parent/Legal Guardian Completing Questionnaires** | | | |
| Father | 3（18） | 4（16） | 3（27） |
| Mother | 14（82） | 21（84） | 8（73） |
| Legal guardian | 0（0） | 0（0） | 0（0） |
| **Parent/Legal Guardian Age, y** | | | |
| Under 21 | 0（0） | 0（0） | 0（0） |
| 21-30 | 2（12） | 2（8） | 0（0） |
| 31-40 | 14（82） | 18（72） | 6（54） |
| 41-50 | 2（12） | 5（20） | 4（36） |
| 51-60 | 0（0） | 0（0） | 1（9） |
| Older 60 | 0（0） | 0（0） | 0（0） |
| **Parent/Legal Guardian Highest Level of Education** | | | |
| Primary school graduate | 0（0） | 2（8） | 0（0） |
| Junior-high graduate/technology secondary school graduate | 5（29） | 6（24） | 0（0） |
| high school graduate/junior college degree | 5（29） | 2（8） | 7（64） |
| College graduate | 7（41） | 14（56） | 4（36） |
| Postgraduate/professional degree | 0（0） | 0（0） | 0（0） |
| Not Reported | 0（0） | 1（4） | 0（0） |
| **Annual household income** | | | |
| under 30,000 | 0（0） | 3（12） | 0（0） |
| 30,000-80,000 | 8（47） | 11（44） | 6（55） |
| 80,000-300,000 | 7（41） | 12（48） | 5（45） |
| over 300,000 | 0（0） | 0（0） | 0（0） |
| Not Reported | 2（12） | 0（0） | 0（0） |

**Supplemental Table 2. Parent PedEyeQ Scores in Parents of CEL Children aged 0-4 years，5-11 years and 12-17 years**

| PedEyeQ domains | Median(Range) PedEyeQ scores | | | P value for difference，0-4y vs 5-11y | Mean difference (95% CI), 0-4y vs 5-11y | P value for difference，0-4y vs 12-17y | Mean difference (95% CI), 0-4y vs 12-17y |
|---|---|---|---|---|---|---|---|
| | Age 0-4 y (n=14), No. (%) | Age 5-11 y (n=28), No. (%) | Age 12-17 y (n=9), No. (%) | | | | |
| **Impact on Parent / Family** | 55（35-90） | 60（10-100） | 70（45-95） | 0.562 | -4(-16 to 8) | 0.071 | -14(-30 to 2) |
| **Worry about Child's Eye Condition** | 28（0-65） | 50（5-80） | 50（40-80） | 0.058 | -13(-26 to 0) | 0.014 | -23(-37 to -8) |
| **Worry about Child's Self-Perception and Interactions** | 57（36-86） | 61（21-100） | 71（21-100） | 0.494 | -5(-17 to 7) | 0.204 | -10(-28 to 9) |
| **Worry about Child's Visual Function** | 37（0-62） | 56（6-100） | 50（31-69） | 0.012 | -15(-27 to -3) | 0.079 | -13(-26 to 0) |

**Supplemental Table 3. Spearman's ρ correlation results of relationship between PedEyeQ Domain Scores and patients' best corrected vision acuity**

| PedEyeQ domains (by age group for child and proxy) | Better-eye BCVA (logMAR) | Worse-eye BCVA (logMAR) |
|---|---|---|
| **Child 5–11 years (N=28)** | | |
| Functional Vision | -0.204 | **-0.427** |
| Bothered by eyes/vision | 0.008 | -0.110 |
| Social | -0.190 | -0.314 |
| Frustration/worry | -0.271 | -0.281 |
| **Child 12–17 years (N=9)** | | |
| Functional Vision | -0.353 | **-0.689** |
| Bothered by eyes/vision | -0.300 | **-0.791** |
| Social | -0.523 | **-0.776** |
| Frustration/worry | -0.363 | **-0.429** |
| **Proxy 0–4 years (N=14)** | | |
| Functional Vision | -0.153 | -0.093 |
| Bothered by eyes/vision | -0.084 | -0.275 |
| Social | -0.028 | -0.474 |
| **Proxy 5–11 years (N=28)** | | |
| Functional Vision | -0.191 | -0.294 |
| Bothered by eyes/vision | -0.141 | -0.256 |
| Social | -0.024 | -0.135 |
| Frustration/worry | -0.070 | -0.23 |
| Eye-care | -0.028 | -0.307 |
| **Proxy 12–17 years (N=9)** | | |
| Functional Vision | -0.332 | -0.167 |
| Bothered by eyes/vision | -0.198 | -0.294 |
| Social | **-0.688** | **-0.753** |
| Frustration/worry | -0.094 | -0.207 |
| Eye-care | -0.128 | -0.186 |
| **Parent (N=51)** | | |
| Impact on Parent / Family | -0.168 | -0.242 |
| Worry about Child's Eye Condition | -0.275 | -0.199 |
| Worry about Child's Self-Perception and Interactions | -0.217 | -0.213 |
| Worry about Child's Visual Function | -0.285 | -0.096 |

Bold values indicate $P < 0.05$.