Major Article

Functional vision and quality of life in children with microphthalmia/anophthalmia/coloboma—a cross-sectional study

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PURPOSE To determine the child's and parental perception of functional visual ability (FVA), vision-related and health-related quality of life (VR-QoL, HR-QoL) in children with microphthalmia/anophthalmia/coloboma (MAC).

METHODS Between June 25, 2014, and June 3, 2015, we carried out a cross-sectional observational study at Moorfields Eye Hospital, London, UK, enrolling 45 children 2-16 years of age with MAC attending our clinics, and their parents. To assess FVA, VR-QoL, and HR-QoL we asked participants to complete three validated tools, the Cardiff Visual Ability Questionnaire for Children (CVAQC), the Impact of Vision Impairment for Children (IVI-C) instrument, and the PedsQL V 4.0. The main outcome measures were the FVA, VR-QoL, and HR-QoL scores, reported by children and parents.

RESULTS In children with MAC, FVA is moderately reduced, with a median CVAQC score of −1.4 (IQR, −2.4 to 0.4; range, −3.0 [higher FVA] to +2.8 [lower FVA]). VR-QoL and HR-QoL are greatly reduced, with an IVI-C median score of 63 (IQR, 52-66; normal VR-QoL, 96), a median self-reported PedsQL score of 77 (IQR, 71-90; normal HR-QoL, 100) and parental score of 79 (IQR, 61-93), and a family impact score of 81 (67-93). Psychosocial well-being scores are lower than physical well-being scores. Parents and children have a different perception of the impact of the condition on the child's HR-QoL.

CONCLUSIONS MAC has a significant impact on a child's FVA and QoL, similar to that described by children with acute lymphoblastic leukaemia and chronic systemic conditions. Children and families may benefit from psychosocial support. (J AAPOS 2018;■:1-5)

The microphthalmia/anophthalmia/coloboma (MAC) spectrum of congenital eye malformations is rare, with an estimated prevalence of anophthalmia at 0.6-4.2, microphthalmia at 2-17, and coloboma at 2-14 per 100,000 live births.[1,2] In most children the condition is bilateral, and in around one-third of children it is part of a syndrome associated with extraocular abnormalities, such as brain, craniofacial, cardiac, renal, and urogenital defects.[1-3] The extent of the malformation determines the visual acuity in children with MAC. Vision is often poor, and children with bilateral MAC often have severe sight impairment and require developmental support.[4] It is estimated that MAC is responsible for approximately 15% to 20% of severe visual impairment and blindness in children worldwide.[5] The published literature on the clinical management of MAC is scant.

The growth of the orbital cavity and the development of the maxilla can be significantly affected in the absence of a normal-sized globe. Therefore, in infants, orbital conjunctival conformers of progressively increasing size are applied to expand the orbital tissues; fitting and exchanging expanders may require multiple anesthetics. Cases of marked orbital asymmetry may require orbital reconstruction

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surgery to reduce cosmetic disfigurement. Despite the burden of MAC and its management on children and their families, no study has explored functional visual ability (FVA), vision-related and health-related quality of life (VR-QoL, HR-QoL) in this population. Two studies of adults with MAC reported low HR-QoL, increased anxiety and psychosocial impact from feelings of shame, shyness, sadness, and fear.[6,7] Validated tools to measure FVA, VR-QoL, and HR-QoL in children include the Cardiff Visual Ability Questionnaire for Children (CVAQC) for FVA,[8] Impact of Vision Impairment for Children (IVI-C) instrument for VR-QoL,[9] and PedsQL V 4.0 for HR-QoL.[10,11] The present study aimed to describe the impact of MAC on FVA, VR-QoL, and HR-QoL from a child's perspective. Parental views on the impact of MAC on their child's and family's HR-QoL are also assessed.

Subjects and Methods

This study was approved by the National Research Ethics Committee South Central–Oxford A (14/SC/1052) and adhered to the tenets of the Declaration of Helsinki. Between June 25, 2014, and June 3, 2015, children 2-16 years of age attending Moorfields Eye Hospital were prospectively enrolled. Exclusion criteria were inability to communicate in English and surgical intervention within 1 month (before or after) of completing questionnaires. We screened the medical records of all children attending clinics in advance to identify those who met inclusion criteria. We consecutively approached these children and families. For those who did not wish to take part, we noted the reasons given. We provided parents and children age-appropriate information material and addressed any questions. Parents gave written consent, and children could sign an assent form.

Data Collected

We recorded age at study participation, sex and ethnic background, ocular and systemic diagnoses, laterality and best-corrected visual acuity with both eyes open in logMAR on the day of study participation. Where best-corrected visual acuity was counting fingers, we assigned a value of 2.1 logMAR; hand movements, 2.4 logMAR; light perception, 2.7 logMAR; and no light perception or ocular prosthesis/artificial eye, 3 logMAR.[12] We also recorded details of previous and current treatment, such as number of previous surgical interventions and number of general anesthetics.

To assess FVA, children who were at least 5 years of age completed the CVAQC.[8] The CVAQC was developed with focus groups of children with and without visual impairment and validated in children with visual impairment to assess difficulties in performing daily activities. Designed to be completed by the child, it consists of 25 questions with answers selected on a four-point scale covering education, near and distance vision, getting around, social interaction, leisure, and sports. Using a Rasch conversion calculator provided, we transformed the raw scores into logarithmic scores. The resulting scores range from −3.0 (higher FVA) to +2.8 (lower FVA).

To evaluate VR-QoL, a subgroup of children aged ≥8 years of age enrolled after August 1, 2014, when required agreements and permissions were granted, completed the IVI-C tool.[9] The IVI-C was validated in visually impaired and normally sighted children. It consists of 24 questions with 5 possible answers plus an additional option of "no, for other reasons," covering areas of school, mobility, social interaction, and emotion. We scored the IVI-C responses using the relevant scoring sheet, which allocates values of 0-4, and did not allocate a score when the response "no, for other reasons" was selected. The resulting raw scores range from 0 to 96, with the highest score indicating normal VR-QoL. No Rasch conversion table is available for this tool as yet.

For HR-QoL, we used age-specific versions of the PedsQL Inventory (www.pedsql.org), which allows children >5 years of age to express their views on different aspects of their physical and emotional state and their social and school life.[10,11] Furthermore, parents completed two questionnaires, one regarding their child ("parental report") and one about the impact on the condition on the family ("family report"). The parental report was specific to the age of the child and consisted of 21-23 questions covering children aged 2-4, 5-7, 8-12, and 13-18 years. The family report contained 36 questions. Children from the age of 5 self-administered the questionnaire (PedsQL administration guidelines), and answers were given on a Likert scale of 0-4. We calculated the PedsQL scores following the scoring instructions. If items were left blank, we adjusted the denominator, using the number of completed items instead of the number of total items. It is recommended to remove questionnaires from the analysis if 50% or more of the items have been left blank; this did not occur in our sample. Scores range from 0 to 100, with 100 indicating normal HR-QoL.

All questionnaires were completed on the same day, during a regular clinic appointment. When children needed help, they were assisted by a member of the research team or play leaders, but not by family members.

Statistics

We aimed for an overall sample size of 50, the smallest sample size required for Bland Altman limits-of-agreement analysis. Where data were missing for individual items in the PedsQL and IVI-C, we adjusted the denominator accordingly. For the CVAQC, the Rasch analysis–based calculator takes into account missing data. Where data were missing, datasets were excluded from the relevant analyses. We applied descriptive statistics throughout, reporting means and standard deviations for normally distributed data or median and interquartile range (IQR) for data non-normally distributed. We assessed relationships between age at participation, age at diagnosis, best-corrected visual acuity in the better-seeing eye, sum of surgical interventions, sum of general anesthetics and CVAQC, IVI-C, and Peds QL scores using Spearman rank correlation, and relationships with uni/bilaterality using the Mann-Whitney test. Agreement between parent and child PedsQL scores was assessed using Bland-Altman techniques. Statistical significance was set at the 5% level and all tests conducted were two-tailed. We did not adjust for multiple comparison testing in our exploratory investigations of associations