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Quality of life among caregivers of children with achromatopsia: an exploratory cross-sectional study in a rare disease population

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A – Development of the concept and methodology of the study; B – Query – a review and analysis of the literature; C – Submission of the application to the appropriate Bioethics Committee; D – Collection of research material; E – Analysis of the research material; F – Preparation of draft version of manuscript; G – Critical analysis of manuscript draft version; H – Statistical analysis of the research material; I – Interpretation of the performed statistical analysis; K – Technical preparation of manuscript in accordance with the journal regulations; L – Supervision of the research and preparation of the manuscript

Abstract

Introduction and aim. Achromatopsia is a rare congenital retinal disorder that affects children's visual functioning and may impose psychological burdens on their families. Evidence on the quality of life among caregivers of children with achromatopsia remains limited. The study aimed to assess the quality of life among parents and caregivers of children with achromatopsia and to examine selected sociodemographic factors associated with it.

Methods and materials. A cross-sectional online survey was conducted in November 2023 among 62 parents or legal guardians of children with achromatopsia. Data were collected using a sociodemographic questionnaire and the WHOQOL-BREF. Due to the exploratory character of the study and the small sample size, bivariate analyses were performed using the Mann–Whitney U test.

Results. The scores on the WHOQOL-BREF exceeded 60 points in all domains. The highest mean score was observed in the psychological domain, followed by the social, environmental and physical domains. No statistically significant differences were found according to caregivers' gender or professional activity. Caregivers aged 40 years and older, as well as those with higher education, reported higher environmental-domain scores.

Conclusions. Caregivers of children with achromatopsia reported moderate-to-good quality of life, while environmental resources emerged as a potential area of inequality. Younger caregivers and those with lower educational attainment may require particular attention. Public health strategies should include accessible disease-specific information, counselling, referral pathways to visual rehabilitation services and family-centred support.

Keywords: achromatopsia; caregivers; quality of life; rare diseases; vision disorders.

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INTRODUCTION

Achromatopsia (ACHM) is a rare congenital retinal disorder of genetic origin that leads to a complete inability to perceive colours. The condition is characterised by photophobia and reduced visual acuity, which require the use of specialised visual aids [1,2]. The global prevalence of the disease is estimated to be less than 1 in 30,000 individuals [3]. Achromatopsia is caused by mutations in one of six genes and is inherited in an autosomal recessive manner. Generally, only individuals who are homozygous or compound heterozygous for ACHM variants typically present with characteristic symptoms, while carriers generally have normal vision [4].

Patients with achromatopsia experience impaired visual acuity, photophobia, nystagmus, and colour blindness [5]. These symptoms result from a primary functional defect of the cone photoreceptors, which manifests in early infancy [6]. Approximately 90% of patients with ACHM mutations carry variants in the CNGA3 or CNGB3 genes [7]. CNGB3 mutations are the most common cause of ACHM in Europe and the USA, with a prevalence estimated at ≥50% [8], while CNGA3 mutations occur most frequently in the Middle East and China [9]. The functional defect in many patients involves varying degrees of morphological alterations of the cone-rich central retina, ranging from loss of cone outer segments to severe outer retinal degeneration, including loss of the retinal pigment epithelium [6].

The main symptoms of achromatopsia include increased light sensitivity and glare (photophobia) and involuntary eye movements (nystagmus). Currently, there is no approved therapy for ACHM in humans. However, several preclinical studies have provided evidence supporting the rationale for gene therapy using adenovirus-based vectors [7]. The consequences of blindness and visual impairment are profound, affecting most areas of a child's psychosocial functioning. Vision plays a key role in learning and exploring the material, natural, and socio-cultural environment. Therefore, visual impairment significantly hinders the ability to perform many tasks efficiently,

particularly in areas such as communication, independent mobility, spatial orientation, and everyday activities [10].

Studies conducted among parents of children with rare diseases show that caregiving often involves health monitoring, participation in therapy, information seeking and coordination of care [11-13]. Caregivers are therefore at risk of isolation, fatigue, occupational difficulties and financial strain [13]. In achromatopsia, the chronic course of the disorder, the lack of causal treatment and limited access to specialised services may further increase family burden.

AIM OF THE STUDY

This exploratory study aimed to assess the quality of life among parents and caregivers of children diagnosed with achromatopsia and to examine selected sociodemographic factors associated with quality of life. From a public health perspective, rare paediatric visual disorders may expose families to fragmented care pathways, limited rehabilitation access, insufficient disease-specific information and unequal support services. Caregivers' quality of life may therefore indicate unmet family needs and gaps in health and social care systems.

METHODS AND MATERIALS

This exploratory study employed a cross-sectional design. Data were collected using the Computer-Assisted Web Interview (CAWI) method through an online questionnaire distributed in Facebook support groups for parents of children with achromatopsia. A total of 62 questionnaires were completed in November 2023. Because achromatopsia is a rare disease, recruitment was conducted through online support groups to reach a geographically dispersed caregiver population. This approach allowed access to a hard-to-reach population but may have introduced selection bias. In order to facilitate broader participation, the questionnaire was translated into English, allowing parents from countries such as the Netherlands, Hungary and the United States to take part. The study used a purposive sampling approach and included adult parents or legal guardians of children diagnosed with achromatopsia who provided informed consent to participate. Due to the rarity of the condition, recruiting a large and fully representative sample was not feasible.

In total, 62 parents of children with achromatopsia were surveyed. The mean age of the children was 6.31 ± 3.02 years, and the mean age of the parents was 36.92 ± 7.09 years. Most respondents lived in urban areas, held higher education degrees and were employed full-time (Table 1).

TABLE 1. Characteristics of the study group.

Parameter	N = 62 (%) / Mean ± SD
Parents' age, years	36.92 ± 7.09
Children's age, years	6.31 ± 3.02
Place of residence	
• Urban	36 (58.1)
• Rural	26 (41.9)
Caregiver gender	
• Women	40 (64.5)
• Men	22 (35.5)
Child gender	
• Girls	43 (69.4)
• Boys	19 (30.6)
Education level	
• Higher education	32 (51.7)
• Secondary education	21 (33.9)
• Vocational education	9 (14.4)
Marital status	
• Single	1 (1.6)
• In a partnership	11 (17.7)
• Married	49 (79.1)
• Widowed	1 (1.6)
Professional activity	
• Full-time employment	46 (74.2)
• Part-time employment	8 (12.9)
• Retired / disability pension	1 (1.6)
• Unemployed	1 (1.6)
• Unemployed due to child's illness	6 (9.7)

Assessment of parents'/caregivers' quality of life

The research instrument was a questionnaire composed of two parts. The first part, designed by the authors, collected sociodemographic data. The second part used the standardised WHOQOL-BREF questionnaire, in Polish and English versions, developed by the World Health Organisation [14]. The WHOQOL-BREF assesses quality of life across four domains: physical, psychological, social and environmental. Scores were converted to a 0–100 scale, with higher values indicating better quality of life according to the WHOQOL-BREF scoring guidelines. Higher scores indicated better quality of life. No missing WHOQOL-BREF domain data were identified.

The WHOQOL-BREF is a widely used, psychometrically validated instrument with confirmed reliability and validity across diverse populations, including caregivers of children with rare diseases [15]. It was selected as a generic, internationally validated quality-of-life instrument that enables preliminary assessment across physical, psychological, social and environmental domains. Although it is not caregiver-specific and may not fully capture caregiving burden related to childhood visual impairment, its use was considered appropriate for an exploratory study in a rare disease population. Future studies should combine generic quality-of-life instruments with caregiver-specific tools, such as the PedsQL Family Impact Module or the Impact on Family Scale.

Due to the anonymous, voluntary, non-interventional, questionnaire-based design of the study, and the absence of identifiable personal data or clinical intervention, formal approval from a bioethics committee was not sought. All participants were informed about the purpose and procedures of the study and provided informed consent before completing the questionnaire. The study was conducted in accordance with the Declaration of Helsinki.

Statistical analysis

The data were analysed using Statistica 13.3 TIBCO software. Descriptive statistics were calculated for all analysed variables. Quantitative variables were presented as means, standard deviations, medians, ranges and quartiles, and categorical variables as numbers and percentages. Normality and homogeneity

TABLE 2. Clinical characteristics and functional difficulties reported in children with achromatopsia.

Parameter	N = 62 (%)
First symptoms of the disease	
• Nystagmus	39 (62.9)
• Photophobia	10 (16.1)
• Nystagmus and photophobia	13 (21.0)
Diagnosed refractive error	
• Myopia	15 (24.2)
• Hyperopia	5 (8.1)
• Myopia and astigmatism	29 (46.8)
• Hyperopia and astigmatism	13 (21.0)
Functional difficulties	
• Difficulties with spatial orientation	37 (59.7)
• Difficulties maintaining balance	3 (4.8)
• Difficulties with spatial orientation and balance	12 (19.4)
• None reported	10 (16.1)

TABLE 3. Subjective assessment of caregivers' own quality of life.

Subjective quality of life assessment	N	%
Very poor (1)	1	1.6
Poor (2)	3	4.8
Neither poor nor good (3)	21	33.9
Good (4)	28	45.2
Very good (5)	9	14.5
Total	62	100.0
Mean ± SD	3.66 ± 0.85	

TABLE 4. Scores in the WHOQOL-BREF domains among caregivers of children with achromatopsia.

Domain, 0-100 points	N	M	Me	Min	Max	Q1-Q3	SD
Physical domain	62	60.48	63.00	10.00	94.00	50.00–69.00	15.85
Psychological domain	62	65.69	69.00	19.00	100.00	56.00–75.00	15.57
Social domain	62	65.21	69.00	25.00	100.00	56.00–75.00	14.21
Environmental domain	62	60.63	63.00	6.00	100.00	56.00–69.00	17.17

N – group size; M – mean; Me – median; Min – minimum; Max – maximum; Q1-Q3 – interquartile range; SD – standard deviation.

of variance were assessed using the Shapiro–Wilk and Levene's tests. As normality assumptions were not met, group comparisons were performed using the Mann–Whitney U test, with effect size calculated as $r = Z/\sqrt{N}$. Statistical significance was set at $p < 0.05$. Owing to the exploratory character and small sample size, no correction for multiple comparisons or multivariable modelling was applied. Therefore, the findings should be interpreted as descriptive and hypothesis-generating. The study was reported in accordance with the STROBE Statement [16].

RESULTS

In the majority of children, the first symptom of the disease was nystagmus, while the most common refractive error was myopia combined with astigmatism. Difficulties with spatial orientation were reported most frequently (Table 2).

Most respondents assessed their own quality of life as good (n = 28; 45.2%) or as neither poor nor good (n = 21; 33.9%). Nine respondents (14.5%) assessed their quality of life as very good. On average, caregivers rated their quality of life at 3.66 ± 0.85 points on a five-point scale (Table 3).

Scores in the WHOQOL-BREF domains exceeded 60 points in all four domains. The highest mean score was recorded in the psychological domain (65.69 ± 15.57), followed by the social domain (65.21 ± 14.21). Slightly lower scores were observed in the environmental domain (60.63 ± 17.17) and the physical domain (60.48 ± 15.85). The lowest individual value was observed in the environmental domain, while the maximum scores of 100 points were reported in the psychological, social and environmental domains (Table 4).

No statistically significant differences in WHOQOL-BREF domain scores were found according to caregivers' gender or professional activity ($p > 0.05$). Caregivers aged 40 years and older reported higher scores in the environmental domain than younger caregivers ($p = 0.018$, $r = 0.30$). Caregivers with higher education also reported higher environmental-domain scores than those with secondary or lower education ($p = 0.019$, $r = 0.30$). No statistically significant differences were observed in the physical, psychological or social domains according to the analysed sociodemographic variables. Detailed results are presented in Table 5.

TABLE 5. Scores in the WHOQOL-BREF domains according to caregivers' sociodemographic characteristics.

Variable	Group	Physical domain M ± SD	Psychological domain M ± SD	Social domain M ± SD	Environmental domain M ± SD
Gender	Women	61.65 ± 15.23	65.38 ± 12.86	65.93 ± 13.98	63.13 ± 14.09
	Men	58.36 ± 17.07	66.27 ± 19.91	63.91 ± 14.86	56.09 ± 21.31
	Z; p; r	0.77; 0.440; 0.10	-0.39; 0.697; 0.05	1.07; 0.286; 0.14	1.31; 0.190; 0.17
Age	<40 years	57.81 ± 14.26	64.05 ± 13.23	64.26 ± 12.81	57.60 ± 16.01
	≥40 years	66.10 ± 17.84	69.15 ± 19.55	67.20 ± 16.98	67.00 ± 18.18
	Z; p; r	-1.90; 0.058; 0.24	-1.50; 0.134; 0.19	-0.84; 0.399; 0.11	**-2.37; 0.018; 0.30**
Education level	Secondary or lower	58.50 ± 16.14	64.27 ± 16.70	63.97 ± 14.04	55.33 ± 19.61
	Higher education	62.34 ± 15.59	67.03 ± 14.58	66.38 ± 14.50	65.59 ± 12.96
	Z; p; r	-1.32; 0.188; 0.17	-0.94; 0.349; 0.12	-1.32; 0.185; 0.17	**-2.35; 0.019; 0.30**
Professional activity	Full-time employment	61.96 ± 13.76	68.00 ± 14.36	67.11 ± 12.36	62.50 ± 16.11
	Part-time employment or inactive	56.25 ± 20.67	59.06 ± 17.45	59.75 ± 17.89	55.25 ± 19.46
	Z; p; r	0.81; 0.417; 0.10	1.79; 0.074; 0.23	1.29; 0.198; 0.16	1.01; 0.315; 0.13

M – mean; SD – standard deviation; Z – Mann–Whitney U test statistic; p – statistical significance; r – effect size calculated as $Z/\sqrt{N}$. Statistically significant results are shown in bold.

DISCUSSION

A child's disability is associated with the functioning of the entire family. Parents of children with visual impairments experience emotional stress and social isolation, and a sense of being overwhelmed, particularly when support from the environment is limited [17,19]. Studies involving parents of children with chronic conditions, such as achondroplasia, show that these parents often report lower mental health scores compared with the general population [20].

Achromatopsia is a genetic disease that typically manifests in infancy but is often diagnosed later in childhood. It rarely occurs in isolation and is most often associated with other visual impairments [9, 20]. In the present study, the most frequently reported refractive error was myopia with astigmatism, while the first symptom observed was usually nystagmus. In the study by Brunetti-Pierri et al. [21], hyperopia was diagnosed in 42.9% of patients and myopia in 14.3%. In an Israeli Palestinian cohort, hyperopia was found in 72.3% of patients and myopia in only 5.7% [19]. In the general paediatric population, based on a WHO meta-analysis, the prevalence of myopia is 11.7%, hyperopia 4.6% and astigmatism 14.9% [22].

In the present study, caregivers of children with achromatopsia most frequently assessed their quality of life as good or neutral. The mean scores on the WHOQOL-BREF exceeded 60 points in all domains. Previous research shows that parents of children with visual impairments or chronic conditions may report lower quality of life, particularly in the physical and environmental domains, due to organisational, social and financial burdens [17,23,24]. In the present study, the highest scores were recorded in the psychological domain, which may suggest a degree of emotional adaptation. Lower environmental and physical scores may indicate areas requiring systemic and social support.

Scores in the individual domains of the WHOQOL-BREF did not differ significantly according to caregivers' gender or professional activity. This finding should be interpreted with caution due to the small sample size and exploratory character of the study. Previous studies based on the WHOQOL-BREF scale do not always demonstrate a consistent role of gender or employment status in shaping caregiver quality of life. These relationships may depend on socioeconomic context, type of caregiving responsibilities and culturally shaped gender roles [25,26].

Statistically significant differences were observed only in the environmental domain. Caregivers aged 40 years and older and those with higher education reported higher mean scores in the environmental-domain. Both associations were accompanied by medium effect sizes. The environmental domain includes aspects such as financial resources, safety, access to health services and information, housing conditions and opportunities for recreation [14]. Older age may be related to greater economic stability, better occupational standing and more experience in navigating the healthcare system, all of which may contribute to higher environmental-domain scores [27]. Similarly, higher education is often associated with better skills in obtaining medical information, accessing available services and seeking support effectively. Research indicates that education and material resources may protect against the negative effects of caregiving burden [28].

From a public health perspective, the lower environmental-domain scores among younger and less educated caregivers may indicate potential inequalities in access to information, services and supportive resources. In rare diseases, where knowledge is limited, and care pathways are often fragmented, caregivers may be required to act as coordinators of care. Therefore, structured information, counselling and referral to visual rehabilitation services may be particularly important for families affected by achromatopsia.

The literature on parenting children with visual impairments highlights that caregivers' experiences are largely shaped by psychosocial factors, including the quality of information received from specialists, access to counselling, rehabilitation and social support [25,29]. In the context of achromatopsia, two aspects appear particularly relevant: the relative stability of the disease, due to the lack of progressive vision loss, and the limitations stemming from the child's visual deficits, including the need for correction, specialised education and environmental adaptations. These factors are directly related to the environmental domain [30].

Patients with achromatopsia encounter numerous difficulties in performing daily tasks, such as eating independently, using the phone or maintaining personal hygiene. The greatest challenges, however, concern recreational activities that require higher visual competencies, such as travelling or using computers [31]. These difficulties may directly burden families and may partly explain the lower environmental-domain scores observed in the present study. At the same time, increasing importance is being attributed to assistive technologies, such as mobile applications, light filters and magnifying devices. Research suggests that appropriate technological tools can reduce functional limitations in patients with visual impairments and thus ease the burden on families. Implementing such solutions in clinical practice and daily family life could represent an important step toward improving the quality of life of both children with achromatopsia and their caregivers [30].

Achromatopsia affects not only visual functioning but also family adaptation. The relatively high psychological-domain scores may suggest adaptation to the stable disease course, although this was not directly measured and cannot be confirmed in a cross-sectional study.

CONCLUSIONS

This exploratory cross-sectional study suggests that caregivers of children with achromatopsia generally report moderate-to-good quality of life, with relatively lower scores in the physical and environmental domains. Older age and higher education were associated with better environmental-domain scores, which may reflect differences in access to resources, information and support. Although the findings cannot be generalised due to the small, purposive and heterogeneous sample, they highlight the need to recognise caregivers of children with rare visual disorders as a relevant target group for public health interventions, including structured information, counselling, pathways to visual rehabilitation and family-centred support.

Study Limitations

This study has several limitations. The cross-sectional design precludes any causal inference and does not allow the assessment of changes in caregivers' quality of life over time

or evaluation of adaptive processes. Therefore, the findings should be interpreted as descriptive and hypothesis-generating rather than explanatory. The relatively small sample size and the purposive sampling strategy limit the statistical power of the analyses and the ability to detect subtle differences between subgroups. The recruitment through online support groups may have introduced selection bias, as caregivers who are more engaged, have greater access to resources or experience a relatively better quality of life may have been more likely to participate in the study. The international and culturally heterogeneous sample also limits the interpretation of the findings, as countries may differ in healthcare systems, access to visual rehabilitation, social support and educational resources, which could not be controlled for due to the small and uneven country-level numbers. The results cannot be generalised to all caregivers of children with achromatopsia. The absence of a control group limits the interpretation of whether the observed quality-of-life levels are high or low in comparison with other populations. The study did not account for clinical factors that may be associated with caregivers' quality of life, including the severity of the child's visual impairment, the presence of comorbid conditions or the time elapsed since diagnosis. These factors may play an important role in shaping caregivers' experiences and cannot be examined within the scope of the present study. The use of the WHOQOL-BREF questionnaire, while psychometrically validated and widely applied, represents a generic assessment of quality of life and may not fully capture caregiver-specific burdens related to childhood visual impairment, such as decision-making stress, concerns about the child's future or cognitive and organisational overload. Future research would benefit from the use of caregiver-specific instruments and longitudinal designs to provide a more comprehensive understanding of caregivers' quality of life in the context of achromatopsia.

Public health implications

The findings highlight the need to recognise caregivers of children with rare visual disorders as a group that may require targeted informational, psychological and organisational support. Routine assessment of caregiver quality of life may help identify families at risk of reduced well-being, particularly younger caregivers and those with lower educational attainment. Public health strategies should include accessible information about achromatopsia, referral pathways to visual rehabilitation services, family counselling and support groups. Although the present findings are preliminary, they suggest that environmental resources may play an important role in shaping caregiver well-being in rare paediatric conditions.

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