

## Quality of Life of Achromatopsia Patients

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### Abstract

**Purpose:** To ascertain the aspects of life, which are the most affected by achromatopsia. It is significant for discovering their vision rehabilitation needs and creating effective management strategies to improve the quality of life for achromatopsia patients.

**Methods:** We combined several investigation methods to reveal which tasks and goals in daily life are the most affected by color-blindness, low visual acuity, photophobia and nystagmus. Therefore, we chose the Mass of Activity Inventory questionnaire and additionally performed unstructured interviews, which helped to capture patients' unique experience of their disease.

**Results:** The respondents found that the most important and difficult is to reach goals of daily living without anyone else's assistance: to be able to eat, manage finances, use phone independently, perform personal hygiene, household tasks. Only half of the respondents found their abilities to be independent in social interactions as important. Most of the respondents found it difficult and important not to be able to perform the goals of recreation like traveling, use of computers or baking without anyone's assistance.

**Conclusion:** We found, that it would be useful for choosing the best individual management strategy in clinical practice, to improve the healthcare and create assistive technologies for people with achromatopsia. Keywords. achromatopsia, color blindness, quality of life, rehabilitation, questionnaire.

**Keywords:** Achromatopsia; Color blindness; Quality of life; Rehabilitation; Questionnaire

### Introduction

Achromatopsia is an autosomal recessive disorder, defined as a loss of cone function in the retina and presenting with color blindness (CB), photophobia, nystagmus and low visual acuity. [1,2] Pathological changes in the retina are determined by mutations in the

genes CNGA3, CNGB3, GNAT2, PDE6C, and PDE6H, that are responsible for cone phototransduction cascade. It is a rare condition, which affects 1 in 30,000 people worldwide. [3] Clinical diagnosis is based on symptoms, morphological and functional findings, and confirmed by molecular genetic testing. [1] As there is still no

specific treatment, a better understanding of the impact that achromatopsia has on everyday life from the patient-perspective is crucial for providing the best possible symptomatic care. [4]

In this study, we aim to ascertain the aspects of life, which are the most affected by achromatopsia. It is significant for discovering their vision rehabilitation needs and creating effective management strategies to improve the quality of life for these patients.

## Methods

As there is no specific questionnaire to measure vision-related quality of life in patients with achromatopsia, we combined several investigation methods. We chose the Mass of Activity Inventory questionnaire to investigate which tasks and goals in daily life are the most affected by color-blindness<sup>5</sup>. However, this tool does not cover other aspects of achromatopsia – low visual acuity, photophobia and nystagmus. Therefore, we have also performed unstructured interviews, which helped to capture patients' unique experience of their disease.

The questionnaire consisted of 3 activity objectives (daily living, social interaction, and recreation), which were specified by activity goals. The most important of them are listed below:

- Goals of daily living: personal hygiene, dressing, personal health care, eating, daily meal preparation, household tasks, personal communication, correspondence, management of finances, shopping, child care, driving, telephone use.
- Goals of social interaction: social functions, dining out, spectator events, attending of meetings, playing games, performing in public.
- Goals of recreation: leisure entertainment, exercise, sewing or needlework, painting or drawing, cooking or baking, electrical work, playing musical instrument, travels, fishing, hunting, outdoor activities, sports, computer use, collecting, reading the newspaper, photography.

A subject was asked to rate each goal on 4-point scale ranging from not important to very important. If the goal was rated as not important – question was skipped to another goal. Otherwise, the subject was asked to rate the difficulty of the goal on a 5-point scale ranging from “not difficult” to “impossible to do without someone else's help”. If the subject rated the goal as “not difficult”, we also moved on to the next Goal, otherwise the patient was asked to rate the difficulty of Tasks on the same 5-point difficulty scale.

All patients gave their written informed consent to use their anonymized clinical and genetic data for publication. The study conformed to the Declaration of Helsinki of 1984 and subsequent amendments. Ethics committee approval was obtained.

## Results

Questionnaire was responded by 8 patients (2 females, 6 males) with achromatopsia. Their age ranged from 5 to 29 years. Some of them needed family member's help to fill in the questionnaire. Relevant clinical data is listed in Table 1. The photopic ERG was non recordable for all the patients. SD-OCT showed different amount of EZ changes or existing ERM. The ophthalmological data is shown in Table 1.

### Goals of daily living

In the first set of questions, respondents were asked how important it is for them to be able to reach goals of daily living without anyone else's assistance. 100% respondents found it very important to be able to eat, manage finances and use phone independently. On the other hand, these activities were troublesome: 33,3% of respondents found it slightly difficult to locate food on a plate or a table; 75% of respondents rated reading a bill, financial or bank statements, filling tax form while managing finances to be slightly difficult.

80-85,7% patients rated being able to independently take care of personal hygiene, household tasks, dressing, personal communications, correspondence and shopping as very important. These goals of daily life are also associated with challenges. 80% of respondents find it slightly/moderately difficult to brush and floss teeth, clean dentures, care for skin, shave, do makeup. Slightly difficult household tasks for 75% of patients were detecting stains on clothes, threading a needle, sewing a hem or a button, hammering a nail. 75% of patients also found matching clothes and accessories slightly difficult.

Personal communications are limited due to issues seeing facial expressions, gestures, body language, recognizing people and making eye contact. 40% of respondents found these aspects moderately difficult, 20% - impossible without help, others - not difficult. Correspondence (sending or reading mails and e-mails) was slightly/moderately difficult for 66,6% of patients, e-messages were preferred to handwritten communication. Shopping was rated as difficult by all patients (by 25% of them – very difficult), the most challenging aspects of shopping were arranging transportation, locating products in the store, reading product labels, receipts, price

tags, nutritional information on food packaging, counting/identifying currency. All respondents classified independency while driving as neither important nor possible. 80% of respondents found daily meal preparation very important, yet slightly difficult. 60% of

respondents rated being able to care for young children as very important. 66,6% of them found it to be slightly/moderately difficult, especially – supervising, dressing and reading to children. Only 42,9% of respondents selected being able to take care of personal health needs without anyone’s assistance very important.

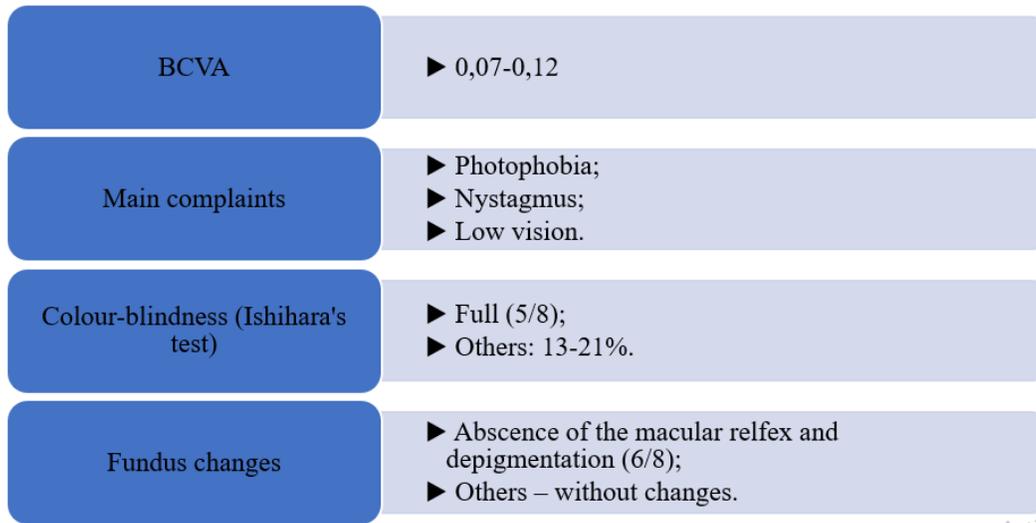


Table 1: Clinical data of the participants.

**Goals of social interaction**

Second set of questions was designed to determine achromatopsia’s impact on social interactions. We present only the goals, which were difficult for the patients (Table 2).

**Goals of recreation**

Third set of questions was designed to determine achromatopsia’s impact on recreation. We present only the goals, which were important for the patients (Table 3).

Goal of social interaction	How important it is for you to be able to reach the goal without anyone else’s assistance?	How difficult it is for you to be able to reach the goal without anyone else’s assistance?	The most difficult aspect of the goal
Social functions (attend parties and other functions)	40% - very important, 20% - moderately important, 40% - slightly important,	25%- moderately difficult 50% - slightly difficult, 25% - not difficult,	Recognizing people, seeing gestures, food, arranging transportation, finding the way and walking around at social function
Attending meetings	40% - very important 60% - slightly important,	25% - slightly difficult 75% - not difficult	Reading agenda, seeing charts, projection screens
Dinning out	33,3% - very important 50% - moderately important, 16,7% - slightly important	50% - moderately difficult, 25% - slightly difficult, 25% - not difficult	Reading menus, receipts, bill, counting currency
Playing games	16,7% - very important 16,7% - moderately important 50% - slightly important, 16,7% - not important	33,3% - very difficult, 66,7% - moderately difficult	Reading cards, instructions, seeing board games, partner’s signals

Table 2: Overview of the impact on social interactions.

Goal of recreation	How important it is for you to be able to reach the goal without anyone else's assistance?	How difficult it is for you to be able to reach the goal without anyone else's assistance?	The most difficult aspect of the goal
Travels	80% - very important, 20% - slightly important	50% - Impossible to do without someone else's help, 25% - very/slightly difficult	Reading signs, maps, brochures, using transport, walking around, seeing displays at museum
Computer use	66,7% - very important, 15,7% - moderately important 15,7% - not important	66,7% - moderately difficult, 33,3% - not difficult	Filling out forms/ spreadsheets, editing pictures with Photoshop, Paint, etc., playing computer games
Leisure entertainment	50% - very important, 50% - moderately important	50% - slightly difficult, 25% - not difficult, 25% - not applicable	Playing cards, word puzzles, reading numbers on TV, viewing scenery, using camera
Cook or bake for recreation	20% - very important 20% - moderately important 20% - slightly important 40% - not important,	50% - slightly difficult 50% - not difficult	Reading recipes, measuring ingredients
Exercise	28,6% - very important. 28,6% - Slightly important 14,6% - moderately important 28,6% - Not important	50% - slightly difficult, 50% - not difficult	Seeing an exercise instructor, following movements; setting controls on exercise equipment
Outdoor activities	50% - moderately important, 33,3% - slightly important, 16,7% - not important	33,3% - not difficult 33,3% - slightly difficult 33,3% - moderately difficult	Boating, biking, skiing, swimming
Sports	16,7% - moderately important 50% - slightly important 33,3% - not important	50% - moderately difficult 50% - not difficult	Playing golf, tennis, bowling

**Table 3:** Over view of the impact on recreation.

During the unstructured interviews, patients elaborated on the main aspects of the disease, support they were receiving, rehabilitation options. Answers showed that patients with achromatopsia sometimes feel like their disease and daily struggles are not fully understood even by family or close friends. From their point of view, achromatopsia is understood not well enough in the society, which is an obstacle when meeting new people, searching for a job. They tend to avoid communicating with new people because of nystagmus and low vision acuity as well as being outside on a sunny day because of photophobia.

Respondents who have been found to have one of the common gene mutations are very interested in ongoing clinical trials and optimistic about possible gene therapy. Rehabilitation clinics do not always meet patient's expectations or can suggest targeted therapy for specific visual disability.

## Discussion

For decades, tests of visual function (e.g. visual acuity, visual fields, color vision) were the most important measures to determine patient's condition, effect of treatment and prognosis in ophthalmology. However, now the focus is shifting to understanding each patient's subjective appraisal of his/hers condition and outcomes and individualized approach towards rehabilitation and support measures. The first step in the implementation of this patient-centered approach is to collect patient-reported-outcomes (PROs), such as health related quality of life, patient-reported disability, function, satisfaction and others. [6-9] PROs offer complementary information to the objective measurements and is useful for monitoring individual patient, determining impact of an intervention, predicting outcomes, assessing needs of patients, screening for specific health disorders. It supports patient-centered decision making as well as helps improve provision of services in healthcare systems based on patients' needs. [8-11] One of the best ways to collect PROs is by using disease specific quality of life questionnaire. [9-12]

Barry JA et al. study created and validated a questionnaire measuring the CB impact on quality of life (CBQoL). This study showed that color blind people score significantly lower on scales of emotional well-being (feel anxious, lacking self-esteem), work (even if an occupation does not obviously rely on color), health and lifestyle (e.g., experience difficulties taking colored medication). [13] McLean et al. study developed a nystagmus-specific QOL questionnaire (NYS-29) and found nystagmus to have a negative impact on person's ability to create relationships, explore new places, it also causes to feel self-conscious or excluded, restricts choice of occupation. [14] Another study found these six domains of living to be adversely affected by nystagmus: visual function, movement, standing out/not fitting in, feelings about the inner self, negativity with regards to the future and relationships. [15]

However, there is no quality of life questionnaire, which would cover all the achromatopsia's symptoms. The main aim of the study was to reflect the major difficulties experienced by patients with achromatopsia, who are suffering from CB, nystagmus, photophobia and low visual acuity at the same time. Most important goals for patients with achromatopsia in daily living sections were - being able to eat, manage finances and use phone independently. In social interaction section - entertaining guests and social functions. In recreation section - traveling, using a computer and leisure entertainment. It can be noticed, that the most difficult tasks are the ones, requiring color-coding (deciding if the meal is well baked, playing games, dressing up), precise movements (shaving, pouring liquids, setting up decorations), good visual acuity (seeing face expressions, reading, counting, traveling).

Furthermore, respondents reported lack of awareness about achromatopsia in general society, which contributes to stigma around visual impairment and low-vision aids usage, sets barriers when creating relationships, searching for a job, can lead to mental health issues. [16-17] Respondents also expressed concerns about rehabilitation facilities lacking programs, that would meet patients' with achromatopsia needs. Other studies also show, that people's with visual impairment needs are not fully understood nor met in general and medical society, but it could be changed with information collected from PROs. [16-18] As there is still no treatment for achromatopsia, improving rehabilitation programs should be a high priority.

We imply that this research could be used for developing a standardised achromatopsia-specific quality of life questionnaire, which would be useful in everyday clinical practice as well as for improving rehabilitation programs, adjusting housekeeping and personal hygiene tools, creating user-friendly software for digital gadgets, as well as spreading awareness about this condition. This is the first study to research the impact achromatopsia has on quality of life, which provides information, helpful for developing a standardised achromatopsia-specific quality of life tool. It would be useful for choosing the best individual management strategy in clinical practice (e.g. rehabilitation program). It is also important to raise awareness about this complex condition, improving healthcare and creating assistive technologies for people with achromatopsia.

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#### Declaration of interest statement

The authors declare no competing interests.

The manuscript has been read and approved by all the authors, the requirements for authorship as stated earlier in this document have been met, and each author believes that the manuscript represents honest work.

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