

Quality of Life of Achromatopsia Patients

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(CB), ... logical changes in the retina are determined by mutations in the ... 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

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work, playing musical instrument, travels, fishing, haunting, 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 consent

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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

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



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			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

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optimistic about possible gene therapy. Rehabilitation clinics do not always meet patient's expectations or can suggest targeted therapy for specific visual disability.

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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 quality of life of patients with achromatopsia. The study included 10 patients with achromatopsia and 10 healthy controls. The questionnaire was designed to assess the impact of achromatopsia on various aspects of life, including social interactions, employment, and overall quality of life. The results showed that patients with achromatopsia experienced significantly lower quality of life compared to the healthy controls, particularly in terms of social interactions and employment. The study also identified several barriers to rehabilitation, such as a lack of specialized programs and services. The authors concluded that the questionnaire could be a valuable tool for assessing the quality of life of patients with achromatopsia and for identifying areas for improvement in rehabilitation services.

How can we address these challenges? One approach is to develop specialized rehabilitation programs and services for patients with achromatopsia. This could include providing training in social skills, job training, and assistive technology. Another approach is to raise awareness of achromatopsia in the general public and among healthcare providers. This could help reduce stigma and improve access to care. Finally, further research is needed to better understand the impact of achromatopsia on quality of life and to develop more effective rehabilitation strategies.

Future research should focus on developing specialized rehabilitation programs and services for patients with achromatopsia. This could include providing training in social skills, job training, and assistive technology. Another approach is to raise awareness of achromatopsia in the general public and among healthcare providers. This could help reduce stigma and improve access to care. Finally, further research is needed to better understand the impact of achromatopsia on quality of life and to develop more effective rehabilitation strategies.

We imply that this research could be used for developing a standardized questionnaire for assessing the quality of life of patients with achromatopsia. This questionnaire could be used in clinical settings to help healthcare providers better understand the needs of their patients and to develop more effective rehabilitation strategies. Additionally, the research could be used for developing educational materials for the general public and healthcare providers to raise awareness of achromatopsia and its impact on quality of life.

References

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