

Quality of Life Analysis in Patients with Retinitis Pigmentosa

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Keywords

Retinitis pigmentosa · Quality of life · Depression

Abstract

Introduction: Retinitis pigmentosa (RP) is a chronic progressive disease causing loss of visual acuity and ultimately blindness. This visual impairment can contribute to psychiatric comorbidity and worse overall quality of life (QOL). Our goal was to assess the relationship between the severity of disease for people with RP and QOL as it pertains to mental health, social support, disability resources, and financial factors. **Methods:** This was a survey study conducted from June 2021 to February 2022 including 38 people with RP. QOL was assessed through a survey questionnaire focusing specifically on demographics, visual function, family, employment, social support, and mental health/well-being. Statistical analysis was conducted using a χ^2 test for significance. **Results:** A best corrected visual acuity (BCVA) of less than 20/200 ($p = 0.0285$) and living alone ($p = 0.0358$) were both statistically significant independent risk factors for experiencing depressive symptoms. Highest education level attained and unemployment rate were not found to be related to the development of depressive symptoms. Subjects had a higher unemployment rate (64% vs. US rate of 3.6%) and a high likelihood of reporting depressive symptoms (47.4%). **Conclusion:** People with RP are more likely to be unemployed and to develop depressive symptoms when compared to the

general population. Similar to previous studies' findings, those with a BCVA of less than 20/200 were statistically more likely to experience depressive symptoms; living alone is a novel risk factor that is also associated with the presence of depressive symptoms. Contrary to prior findings, highest education level and unemployment status were found not to be related to the development of depressive symptoms. These patients may benefit from regular depression screenings and optional establishment of care with a psychiatrist or psychologist if they live alone or their BCVA is 20/200 or worse.

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Introduction

Retinitis pigmentosa (RP) is a group of genetically heterogeneous disorders of the retina characterized by a gradual degeneration of the photoreceptors resulting in loss of vision and visual field [1–3]. People with RP can initially present with night blindness and decreasing peripheral visual fields [4]. RP is the most common inherited disease of the retina, impacting approximately 1 in 4,000 people in the USA and 1 in 5,000 people worldwide [1]. Currently, with limited treatment options available that may slow the progression of the disease, management of RP largely focuses on supportive care. With progressive vision loss, the level of disability

increases with the progression of their disease, and patients may find themselves requiring more visual aids, experiencing limited mobility, and losing some of their independence. It follows intuitively, and has been confirmed in the literature, that this would lead to higher rates of depressive disorders as their disease advances [5, 6].

Of any type of major sensory loss, vision impairment is the most psychologically, socially, and potentially financially debilitating. Prior studies have found that worsening vision or visual impairment was associated with reports of poor job security [7] and high rates of depression and/or anxiety [6, 8–12]. Specifically in people with RP, worsening visual acuity has been associated with a lower quality of life (QOL) and emotional state deterioration [13]. According to studies done in several countries, several factors that may contribute to this population's lower QOL include difficulty with day-to-day tasks and concerns about personal safety [14]; worry about disease progression and uncertainties of the future [14]; difficulty adjusting to their visual loss in their social environment and family relationships [15]; lower education levels [16]; and difficulty maintaining employment [17, 18]. Unemployment has been associated with not only a lower income in a population that already has considerable health expenditures [19], but it also has substantial health impacts of both adults and their children [20–23]. The current literature suggests that globally, people with RP may be at higher risk of experiencing negative impacts on their mental health and well-being than those with normal vision. However, the research quantifying this relationship between mental health and other components of the QOL in people with RP is limited in the USA. Thus, further investigation into the factors comprising this patient population's QOL is critical in order to assess whether the current disability aid and health care resources in the USA are sufficient to address the needs of those living with RP. The objectives of this study were to better understand the components contributing to the diminished QOL in people with RP through investigating the relationships between severity of visual impairment, quality of healthcare and access to disability resources, education level and employment rate, living situation and social support, and mental health and personal well-being.

Methods

This study was designed as a cross-sectional survey questionnaire on a sample of 38 patients followed at the University of Florida Health Department of Ophthalmology at Gainesville. This study was conducted in accordance with the University of Florida Institutional Review Board policies, guidelines, and guidance.

Collection of Data

Of 142 patients found eligible for the study from the University of Florida Shands Health System, 38 subjects were successfully consented and enrolled to participate. Eligibility criteria included patients who had a confirmed diagnosis of RP by ICD-10 codes seen at the University of Florida Health Eye Clinic in Gainesville and excluded patients who did not have a diagnosis of RP by ICD-10 codes. Demographic information and best corrected visual acuity (BCVA) were obtained through chart review of electronic medical records. An attempt was made to collect visual field data to better assess visual function; however, most patients did not have recent visual field testing done, and most visual field data available were from the time of diagnosis. QOL was assessed through a specifically designed survey questionnaire focusing primarily on demographics, vision specifics, family, employment, healthcare, social support, and mental health/well-being (Table 1).

Statistical Analyses

Results are reported as the mean \pm SD unless otherwise indicated. Statistical significance was defined as $p < 0.05$. Categorical variables were compared using a χ^2 test for significance with the corresponding odds ratio. For quantitative variables, comparisons were done with unpaired, two-tailed t tests. Analysis was carried out using Excel software.

Results

Thirty-eight subjects including 28 males and 10 females were successfully consented, enrolled to participate, and surveyed for the study. The age of subjects ranged from 14 to 91 years with a mean age of 50.8 ± 21.9 years. Four subjects were minors, and 1 patient was still in school. Patients were surveyed regarding their disease, lifestyle, socioeconomic factors, and education. Regarding the presence of depressive symptoms, 37 of 38 responded and 1 preferred not to comment. Visual field data in the form of Goldmann V-4e perimetry were available for only 13/38 patients, and none of the visual field testing had been done within the past 5 years. Thus, the decision was made to use BCVA as a surrogate of visual function. Of the 37 who responded about depressive symptoms, BCVA was available for 35 of 37 patients. The subjects were surveyed from June 2021 to February 2022.

Visual Function, Household Data, and Disability Status

As summarized in Table 2, of the 38 subjects with a confirmed diagnosis of RP in the study, 78.9% (30/38) reported first noticing visual difficulties under the age of 18 years old, and 13.2% (5/38) first started noticing visual difficulties at age 35 or older. With regards to visual function, 42.2% (14/33) patients had BCVA of 20/40 or better, and 51.4% (18/35) had BCVA of 20/200 or better.

Table 1. QOL questionnaire components

Survey category	Data collected from questionnaire
Demographic information	Age Gender BMI Education level of completion Degree specification
Vision-specific questions	Age with noticeable visual difficulties Use of visual aids Need of visual aids to complete education
Family	Family history of ocular disease Living alone or with others
Income/finances	Employment Seeking employment Visual aids needed that is specific to line of work Household income contribution Job satisfaction Income
Healthcare, health insurance, access to healthcare resources	Health insurance Health insurance satisfaction Low vision aid coverage Difficulty obtaining visual aids Seeking mental healthcare Additional health conditions
Disability resources and transportation	Disability insurance Disability benefits Means of transportation Ability to drive Access to public transportation Difficulty walking along
Social support and experiences	Preferred type of communication (text, phone call, email) Difficulty communicating Attendance of social events Issues at social events Finding others to vent and/or sympathize with
Personal care and well-being	Difficulty with activities of daily living (ADLs) Difficulty reading on electronic devices versus paper Difficulty traversing home Ability to exercise Types of hobbies Difficulty with hobbies Emotional stress Mental care and therapy Physical distress due to vision Disease acceptance

Two of 38 patients (5.3%) had syndromic RP; one had Usher syndrome and the other had Bardet-Biedl syndrome. While the patient with Bardet-Biedl syndrome reported anxiety and depression because of their condition, the patient with Usher syndrome did not report psychological stress. As seen in Table 2, it was established

that 57.9% (22/38) of subjects had a family history of RP; of the 22 patients with a family history of RP, 68.1% (15/22) had a first degree relative with RP.

With regards to the subjects' living situations, 76.3% (29/38) reported living with another person whether it was partner, parents, children, roommate, church

Table 2. Demographics, disease- and visual-specific information, household data

Surveyed participants, n/n_T (%)	
Adults	34/38 (89.5)
Minors	4/38 (10.5)
Age, years	
Age range	14–91
Mean age	50.8±21.9
Biological sex, n/n_T (%)	
Females	28/38 (73.7)
Males	10/38 (26.3)
BMI	
Mean BMI	26.5±6.4
Age at diagnosis, n/n_T (%)	
<18 years	30/38 (78.9)
18–24 years	1/38 (2.6)
25–34 years	2/38 (5.3)
35–44 years	3/38 (7.9)
45–54 years	2/38 (5.3)
Family history, n/n_T (%)	
Family history of RP	22/38 (57.9)
Immediate family member with RP	15/22 (68.2)
No family history of RP	16/38 (42.1)
Household and living situation, n/n_T (%)	
Living with another person(s)	29/38 (76.3)
Living alone	9/38 (23.7)

community members, or in a group home. The remainder of subjects, 23.7% (9/38), reported living alone.

As demonstrated in Figure 1, the percentage of subjects that reported using glasses/contact lenses or a reading magnifier was 71.1% (27/38) or 23.7% (9/38), respectively. It was found that 45% (17/38) of subjects reported utilizing a cane, 15.8% (6/38) received help with a service dog, and 44.7% (17/38) had a personal assistant. Overall, it was established that 65.8% (25/38) of subjects used at least 2 kinds of visual aids and 42.1% (16/38) used at least 3 different kinds of visual aids. Less than half of the subjects, 44.7% (17/38), reported using a cane for visual support (Fig. 1). Although those with worse visual acuity tended to have a higher number of visual aids used on average, statistical significance was not appreciated (Table 3).

Education Level and Employment Status

Because 4 patients were minors who had not yet completed their education, they were excluded from analysis of education level and employment status. Of 34 adult subjects, the percentage of those who had achieved at least a high school degree was 91.2% (31/34), comparable to 91.1% of people nationally who had at least

completed high school in 2021. The percentage of subjects who had obtained bachelor's degree or higher was 52.9% (18/34), noticeably higher than the 37.9% of people nationally who had completed at least a bachelor's degree in 2021 [24].

Of the 34 adult participants, the percentage of subjects who reported being unemployed was 41.1% (14/34). Of the 79% (11/14) of subjects who were unemployed and not seeking employment, 54.5% (6/11) of them reported that it was due to their vision impairment. The remaining adult subjects who reported being employed or retired were 23.5% (8/34) and 35.3% (12/38), respectively. Of the 14 unemployed subjects, 57.1% (8/14) reported depressive symptoms.

Transportation

Of the 27 adult subjects that reported utilizing a car for their mode of transportation, 70.3% (19/27) reported that they do not personally drive the car. Overall, the percentage of adult subjects who confirmed that they do not personally drive due to their vision impairment was 73.5% (25/34). The percentage of subjects utilizing a lift service or the public bus were 23.7% (9/38) and 5.3% (2/38), respectively.

Mental Health and Wellness

It was found that 47.4% (18/38) of subjects had experienced depressive symptoms to some degree due to their vision, including but not limited to, feelings of anxiety, depression, irritation, anger or anger outbursts, and/or isolation. Additionally, 10.5% (5/38) of subjects reported having a learning disability secondary to their vision deficit and 5.3% (2/38) of subjects reported poor nutrition attributed to their vision deficit. Of the 18 subjects who reported depressive symptoms, 16.7% (3/18) of them explained that they do not seek mental healthcare due to their vision.

Whether or not the subjects lived alone or lived with others in their household was found to have a significant association on whether they had experienced depressive symptoms. Of the nine subjects who reported living alone, 66.7% (6/9) of them had experienced depressive symptoms as compared to 41.3% (12/29) of subjects living with others ($p = 0.036$).

As depicted in Figure 2, worsening visual function as defined as a visual acuity of less than 20/200 was statistically significantly associated with the presence of depressive symptoms ($p = 0.0285$) as compared to subjects with a visual acuity of 20/200 or greater in this subject population. However, there was not statistical significance appreciated for the number of subjects with a visual acuity of less than 20/40 experiencing depressive symptoms compared to those with a visual acuity of 20/40 or greater ($p = 0.409$).

Increasing education level, measured through increasing degree levels from high school, college, or university

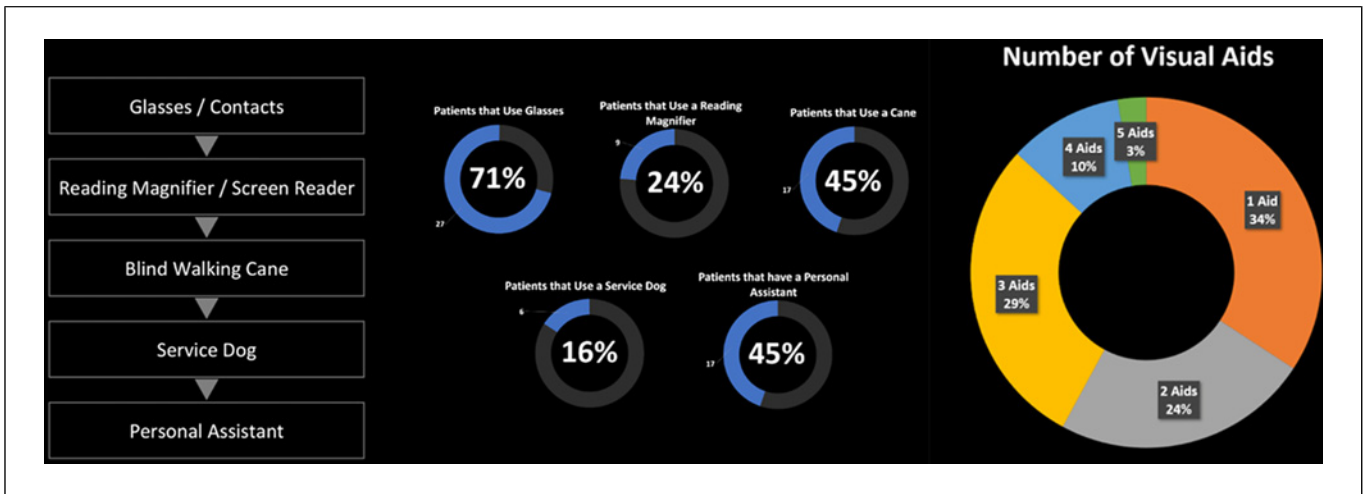


Fig. 1. Patient-specific visual support and number of visual aids utilized.

Table 3. Visual acuity and number of visual aids used

Visual acuity	Mean and standard deviation (SD)	<i>p</i> value
≥20/40	Mean: 2.00 SD: 1.07	0.4216
<20/40	Mean: 2.28 SD: 0.94	
≥20/200	Mean: 1.89 SD: 0.90	0.1254
<20/200	Mean: 2.41 SD: 1.06	

bachelor's degree, graduate master's degree, or a doctorate degree, was not found to be statistically significantly associated with the presence of depressive symptoms in this subject population (Table 4). Although 57.1% (8/14) of the subjects who reported being unemployed had reported depressive symptoms, this was not found to be statistically significant ($p = 0.2264$) when compared with subjects reporting depressive symptoms who were actively employed or retired (36.8% (7/19)). The five subjects who were minors or still in school full time were not included in this employment analysis.

Discussion

This QOL assessment demonstrated that almost half of the subject population (45.9%) experienced symptoms of depression. This is significantly higher than the national

average according to the National Institute of Mental Health, which states that approximately 8.4% of adults over the age of 18 in the USA have a history of at least 1 major depressive episode [25]. Not surprisingly, previous studies have described that people with vision impairment, and specifically RP, were shown to be at a higher risk of developing problems with their mental health compared to the general population [6, 8, 11, 12].

While it would have been preferable to correlate visual field data with the presence of depressive symptoms, other studies have used visual acuity as a surrogate of visual function in people with RP [6, 9, 10]. Similar to findings from previous studies [6, 9, 10], our findings also demonstrated that those with a lower visual acuity, specifically less than 20/200 in this subject population, were statistically more likely to experience depressive symptoms with an OR of 4.767 ($p = 0.0285$). Azoulay et al. [13] also found in their study that a logMAR VA worse than 0.3, or 20/40, in people with RP was associated with a significant drop in the QOL; poor VA was also reported to affect the emotional state of the participants. While these findings support worsening visual acuity in RP being a predictor for the development of depression, it remains unclear whether the development of depressive symptoms is secondary to the psychosocial and financial burdens associated with vision impairment or is associated with the disease process directly [26].

Our study findings demonstrated that the unemployment rate of 41.1% of subjects with RP is substantially higher than the unemployment rate in the USA of 3.8% per US Bureau of Labor Statistics data from February of 2022, supporting findings from prior studies. A study by Sherrod et al. [27] in 2014 found that even after correcting

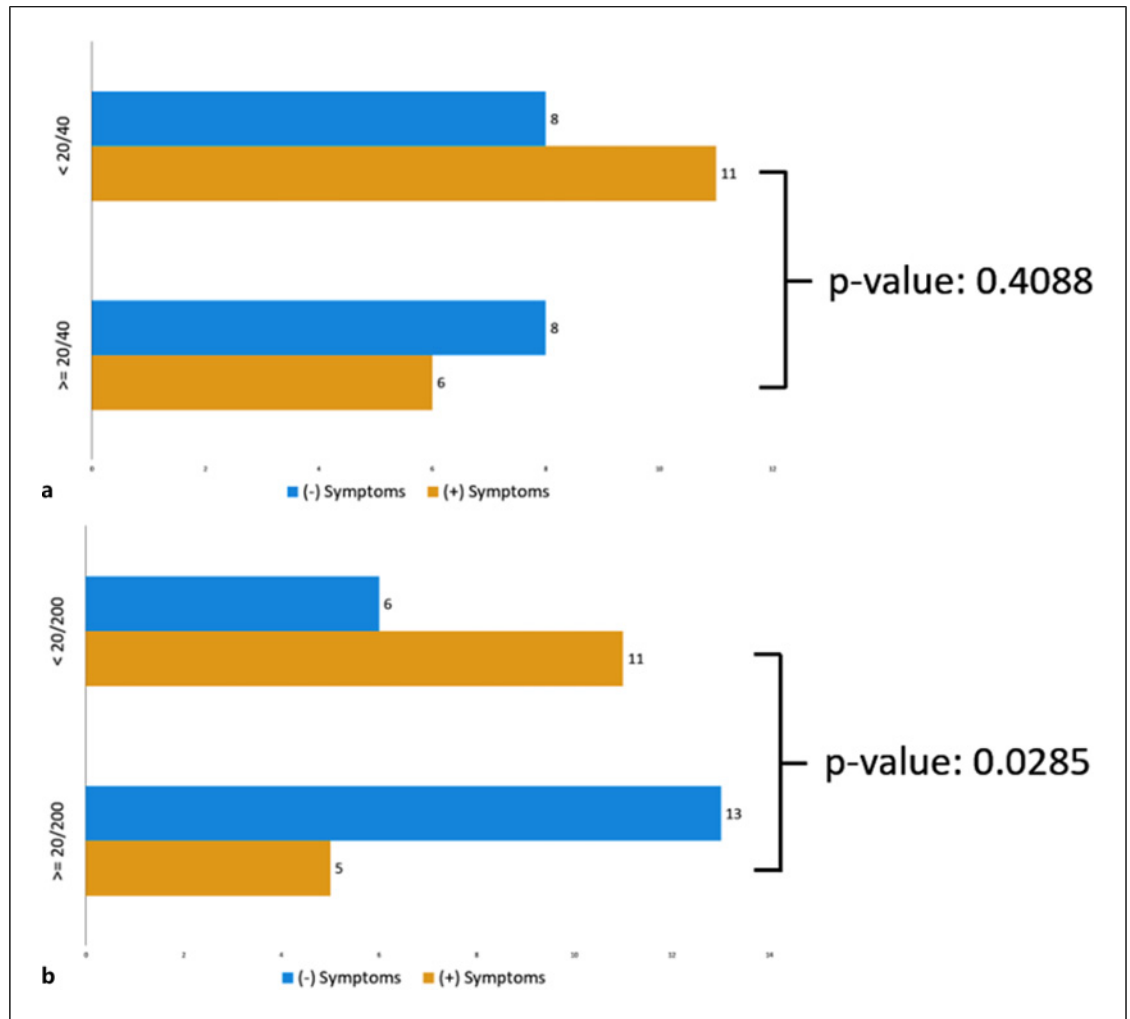


Fig. 2. a Graph summarizing the number of subjects with and without depressive symptoms who had a visual acuity of worse than 20/40 and those with a visual acuity of 20/40 or greater. **b** Graph summarizing the number of subjects with and without depressive symptoms who had a visual acuity of worse than 20/200 and those with a visual acuity of 20/200 or greater.

for age, sex, race and ethnicity, and chronic disease status, those with uncorrected refractive errors and visual impairments were more likely to be unemployed. There is a considerable amount of evidence highlighting a positive association between unemployment and the development of depression and other mental health issues [9, 28–30]. Indeed, in this study, unemployment was associated with an OR of 2.40 for the presence of depressive symptoms in people with RP, although this finding was not statistically significant ($p = 0.2264$). However, this finding is supported by Chaumet-Riffaud et al. [18], which found that unemployment rate was not found to be related to the development of depressive symptoms in this subject population. When insurance providers were compared, there was no statistically significant

difference appreciated, but some interesting trends were observed. It was found that having Medicaid coverage was associated with an OR of 1.93 for the presence of depressive symptoms, although this was not found to be statistically significant ($p = 0.4977$). This could be owed to the target population of Medicaid generally being younger, more vulnerable individuals. Having private insurance was protective against the presence of depressive symptoms, with an OR of 0.779 ($p = 0.7174$). This is likely a confound factor given that those with private insurance have the disposable income to afford it rather than having to rely on government insurance.

This study population was more highly educated than the general population. The percentage of subjects who

Table 4. Summary of examined risk factors for depression with corresponding odds ratio and *p* value for significance

Risk factor	Odds ratio	<i>p</i> value
Age >30	4.86	0.1569
Age >65	0.61	0.4948
Female	0.43	0.2581
Completed high school	0.47	0.5595
Completed bachelor's degree	1.0	1.0
Completed master's degree	1.61	0.6406
Completed doctorate	1.0	1.0
BMI >25	1.19	0.7956
Unemployed	2.40	0.2264
Medicare insurance	0.92	0.9003
Medicaid insurance	1.93	0.4977
Private insurance	0.77	0.7174
Family history of RP	1.83	0.3682
BCVA <20/40	1.83	0.4088
BCVA <20/200	4.77	0.0285
Lives alone	8.50	0.0359

achieved higher education, as defined by obtaining a bachelor's degree or higher, was 52.9% (18/34), as compared to 31.5% of the general population in Florida and 37.9% of the general population in the USA [31]. In study done by Chaumet-Riffaud et al. [18], it was also seen that level of education was not significantly impaired by RP. Given that 45.2% of the Alachua County population has obtained a bachelor's degree or higher [32], it is possible that our study population's higher education is due to the nature of enrollment at a university health center. Despite this, level of education completed was not significantly associated with the presence of depressive symptoms (Table 4). This contrasts findings from prior studies showing that more years of education have a protective effect against depression [5, 33].

Our study findings demonstrated that BMI >25 was not a statistically significant risk factor for the presence of depressive symptoms (*p* = 0.7956). While the average BMI in this study was 26.5 ± 6.4, the average BMI in the USA is 26.6, falling within the 95% confidence interval [31]. To the best of our knowledge, this is the first examination of BMI as a risk factor for depression in people with RP. As there is overwhelming evidence in the current literature of an association between elevated BMI and the presence of mood disorders such as depression [34–36], it is possible that the lack of statistical significance found in our study is a result of small sample size rather than an absence of a relationship. Family history of RP was also found to be not significantly linked to the presence of depressive symptoms (*p* = 0.3682). Although patients over the age of 30 have an

OR of 4.86 for the presence of depressive symptoms, age was additionally found not to be a significant risk factor in people with RP. Interestingly, being older than 65 years was demonstrated to be protective against the presence of depressive symptoms with an OR of 0.61. Among an age group which has had more time to accept and adapt to this lifelong disease process, this study finding is not a surprise. It is also possible that depressive symptoms may have been underreported in this age group, in part due to the stigma or perceived shame that may be associated with depression and other symptoms of mood disorders.

Our results additionally demonstrated that subjects who reported living alone were also statistically more likely to report symptoms of depression with an OR of 8.50 (*p* = 0.0359). Similar data have been exhibited in prior studies. Papadopoulos et al. [5] found that negative emotional social support in adult subjects with vision impairment was significantly associated with development of depressive symptoms. Overall, this finding speaks to the importance of having a strong social network in support of mental health and well-being for not only this population with RP, but anyone with a significant visual disability. In addition, given that this questionnaire was done during a period shortly following the height of the COVID-19 pandemic, those living alone who once had social networks outside the house may have experienced an abrupt adjustment to social isolation. Overall, while it is likely that the development of depressive symptoms primarily stems from vision impairment itself, other factors including stress related to social isolation, employment and relationships, family history, and genetics may also play a role.

Depression is often underreported and undertreated due to stigma or lack of education about signs and symptoms [37], and this is only made worse by a visually disabling condition. In our patient sample, 16.7% (3/18) of them admitted that they do not seek mental healthcare due to their vision. Only 33.3% (6/18) were actively seeking treatment in support of their mental health. Thus, it may be beneficial for people with RP to undergo annual or biannual screenings for major depressive disorder that they may be identified and referred for treatment. Especially in people with RP who reach the threshold of having a visual acuity of 20/200 or less, it may be reasonable to not only establish a screening protocol for major depressive disorder such as inquiring about symptoms of depression or administering the Patient Health Questionnaire-9 (PHQ-9) [38] but also to consider a referral to establish further care with a psychiatrist or psychologist. This referral could be considered earlier if the individual lives alone or reports significant decreases in vision, regardless of BCVA. Furthermore, the

results of this study emphasize the importance of encouraging the medical professionals caring for a person with RP to regularly inquire about their local support network and resources they are currently utilizing and to educate them on coping strategies.

With better QOL, vision-related QOL, and reduced comorbidities [6, 10, 39, 40], the impact of treating depressive disorders in those with RP cannot be understated. However, the societal implications of untreated depression in people with RP are not to be overlooked. While this is relevant for people of all ages, it is most evident in younger individuals diagnosed with RP. People with RP have higher rates of depression than their healthy counterparts [6]. As their vision declines and their visual disability worsens, symptoms of depression can increase their risk of being withdrawn from society. If depressive symptoms are experienced in people with RP at a young age and are not addressed promptly and adequately, their ability to develop into high-functioning, involved members of society and achieve any personal, academic, and career goals becomes challenging. The recognition and possible treatment of depressive symptoms or depressive disorders would allow this patient population to have one less barrier to overcome. For older patients, proper management of depressive symptoms or depressive disorders would likely improve QOL and reduce comorbidities. In all patients, and particularly those with RP, BCVA is a readily available and easily measured variable that can be used as a screening tool identify those at higher risk for mental health disorders. Additionally, the social history obtained from people with RP should include information about a patient's support network and cohabitation status, as this may help identify those at higher risk of developing depressive disorders.

Limitations of this study included the lack of a control group with normal vision who were matched for age and education, the small sample size, and the subject population being limited to those receiving care in Gainesville, FL, USA. While the Visual Function Questionnaire-25 (VFQ-25) has commonly been employed to assess QOL in patients with visual impairment [13, 18], it mainly focuses on visual function as a surrogate for QOL. As it stands, there are no validated QOL questionnaires specifically for RP. Additionally, the VFQ-25 does not address such socioeconomic topics as employment, education, or insurance coverage. The questionnaire employed here does overlap with the VFQ-25 by addressing visual function. Future directions for research should focus on implementing a validated major depressive disorder screening such as the PHQ-9 to better quantify the impact of RP on mental health and QOL.

Conclusions

Overall, the findings of this study demonstrate that people with RP in the USA are more likely to be unemployed and to have depressive symptoms when compared to the general population. Similar to previous studies' findings, subjects with a visual acuity of less than 20/200 or those who reported living alone were statistically more likely experience depressive symptoms. Contrary to prior findings, highest education level and unemployment rate was found not to be related to the development of depressive symptoms in this subject population. Thus, further research utilizing matched controls is warranted to investigate how socioeconomic factors, disability benefit utilization, and specific disease features impact mental health in this patient population. Ultimately, while effective and affordable treatment for RP is still limited, these people may benefit from regular depression screenings and optional establishment of care with a psychiatrist or psychologist if visual acuity reaches below 20/200. Lastly, these data emphasize the difference that medical professionals caring for these people can make by regularly inquiring about one's local support network, checking in on the disability resources being utilized, and offering education on various coping strategies.

Statement of Ethics

This study protocol was reviewed and approved by the University of Florida Institutional Review Board, approval number 202100119. This study was granted exemption from requiring informed consent by the University of Florida Institutional Review Board.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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Author Contributions

J.S. primarily served to survey patients and collect the relevant data points and served as the principal investigator and helped coordinate the project to its completion. T.N. and M.D. both

oversaw the project development and execution. T.N. also contributed with statistical analysis. A.H. and L.B. contributed equally to the production and revision of the manuscript. J.C. and S.G. revised the manuscript and provided expert knowledge.

Data Availability Statement

All data generated or analyzed during this study are included in this article. Further inquiries can be directed to the corresponding author.

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