# Patient-Reported Social Impact of Molecularly Confirmed Macular Dystrophies and Cone-Rod Dystrophies

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Keywords: macular dystrophy; cone-rod dystrophy; quality of life; patient-reported impact; social care; anxiety

*Article*

# Patient-Reported Social Impact of Molecularly Confirmed Macular Dystrophies and Cone-Rod Dystrophies

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**Abstract**

Patient-reported impact of macular and cone-rod dystrophies (MD/CRD) was assessed in a large consecutive cohort of individuals with molecularly confirmed diagnoses, aiming to identify key disease-related disadvantages. Out of 281 patients contacted, 194 (69.0%; 55.2% female) responded to an anonymized survey exploring the effects of MD/CRD on vocational training, professional careers, social participation, family life, personal well-being, and experience with ophthalmologic care. While vocational training was generally less affected, professional careers were frequently disrupted, with 20.6% of patients aged ≥50 retiring early. A majority (54.7%) reported feeling restricted in public life. Financial constraints were noted by 20%. A negative impact on familial life (12.3%) was less frequently reported compared to anxiety (74.2%) and depression (15.8%). Diagnostic delays (≥2 years) were common (34.2%) along with a notable rate of initial misdiagnoses (22.1%). The lack of adequate psychological support was a major complaint in professional care. Compared to a previous study in retinitis pigmentosa, MD/CRD patients reported differing patterns of burden, especially in early retirement and family impact. Our findings underscore the need for ophthalmic and social care providers to accelerate the diagnostic process and enhance access to financial assistance and psychological support as key areas to improve patient care.

**Keywords:** macular dystrophy; cone-rod dystrophy; quality of life; patient-reported impact; social care; anxiety

## 1. Introduction

Inherited macular dystrophies (MD) and cone-rod dystrophies (CRD) predominantly involve the posterior pole of the retina affecting central visual functions like visual acuity, central visual field and color vision [1,2]. Usually, disorders located within the temporal vascular arcades are considered as MD and those additionally affecting peripheral regions are called CRD. However, there are differences between regions affected on ophthalmoscopy, retinal imaging and functional testing; there is a continuous spectrum without distinct means of separation between MD and CRD, and during disease progression several MDs will develop into CRDs [1–3]. MD/CRD account for approximately 30 % of inherited retinal dystrophies (IRD) in Germany [4]. In industrialized countries IRDs are the leading cause of blindness in infants as well as adults under 65 years of age [5–9].

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Progressive loss of visual acuity, central visual fields defects and glare may affect vocational training, professional careers, mobility, social life and communication. Visual disability is associated with financial challenges especially in IRDs due to reduced income [10,11] and increased personal health costs [12]. Mental health can be affected in visually disabled patients and their family members with an increased frequency of anxiety and depression [13,14]. In addition, patients complain about an often lengthy patient pathway towards an established clinical and genetical diagnosis as well as other insufficiencies of ophthalmic and psychological care [15].

In contrast to retinitis pigmentosa (RP), patient reported impact of MD/CRD has not been studied specifically except for two small studies in Sorsby fundus dystrophy and Stargardt disease [16,17]. Usually MD/CRD patients have been included within the group of all IRDs, often without consistent genetic confirmation [13,15,18].

The purpose of the present study was to address this gap by conducting a large, anonymous survey focusing exclusively on patients with molecularly confirmed MD/CRD. The design of the study was similar to our previous study on patients with molecularly confirmed retinitis pigmentosa (RP) [19]. This facilitates a direct comparison of the impact of different visual disabilities on the quality of life in MD/CRD and RP.

## 2. Materials and Methods

By searching two separate databases at two separate specialized centers by two of the authors (G.S. and U.K.) a consecutive series of 318 patients with molecularly confirmed cases of non-syndromic or syndromic MD or CRD was identified. Patients were eligible for inclusion if they had at least one pathogenic or likely pathogenic variant in autosomal dominant or X-linked MD/CRD, or two pathogenic or likely pathogenic variants in a known gene for autosomal recessive MD/CRD; had been examined at one of the centers at least once between January 2018 and January 2024; were at least 18 years old at the time of the survey and resided in Germany (the infrastructure of social help differs between European countries).

More than 60% of these patients had an initial diagnosis of MD and most patients were of Caucasian ethnicity. Clinical assessments and molecular genetic confirmation were performed as previously described [20,21]. Ethics approval for the study was received from the respective ethics committees of the North Rhine (ID: 2024037) and Westphalia-Lippe (ID 2024-234-b-S) medical associations.

The concise, anonymous questionnaire was designed based on previously reported patient concerns and consisted of nine structured questions with predefined answer options (see Supplementary File). Except for the disease terms (MD/CRD vs. RP) it was identical to the questionnaire used for the previous study in patients with molecularly confirmed RP [19]. To ensure anonymity, age and age at onset were recorded in ten-year intervals. Seven questions addressed gender, vocational training, work experience, social life, family life, personal challenges and ophthalmologic care.

With the aim to reach patients in a broad age-range and taking into account the reduced visual function, the survey was conducted using a written questionnaire. To ensure anonymity patients were contacted via postal mail including a cover letter (explaining the study's background, purpose, and the measures taken to ensure anonymity), an unmarked questionnaire, and a prepaid, pre-addressed, unmarked return envelope. Patients were required to respond to questions regarding age-range, gender and age at onset. For the remaining questions either multiple responses or the option to skip the question were allowed. All survey letters were mailed on the same day, and patients had a time frame to respond of four weeks. Patients were asked not to include any personal information, as this would be considered a breach of anonymity and would exclude their responses from the study.

Return letters were handled by two individuals without access to the clinical databases. Questionnaires and envelopes were separated by B.L. to delete traceable postal documentation. Analysis of the original questionnaires was performed by N.ZL..

 doi:10.20944/preprints202509.1998.v1



## 3. Results

### *3.1. Patient Characteristics*

A questionnaire was sent to a consecutive series of 318 patients. A total of 37 out of 318 survey letters were returned due to invalid addresses, resulting in a final patient cohort of 281 individuals. Within the four-week response period, 194 out of 281 patients (69.0 %) submitted completed questionnaires. No responses were excluded due to the inclusion of personal information. The distributions of age groups and gender among respondents is shown in Table 1. The majority (102 out of 194; 52.6 %) were older than 50 years of age. In terms of gender, 107 respondents (55.2 %) identified as female, 85 (43.8 %) as male, and 2 (1.0%) as diverse. Notably, male respondents tended to be younger, with 42.4 % under 41 years of age, compared to 26.2 % of female respondents in the same age group.

**Table 1.** Distribution of patient age.

| Age distribution | All | | Female | | Male | |
|---|---|---|---|---|---|---|
| [years of age] | N=194* | % | N=107 | % | N=85 | % |
| **18-20** | 10 | 5.2 | 5 | 4.7 | 5 | 5.9 |
| **21-30** | 31 | 16.0 | 12 | 11.2 | 19 | 22.4 |
| **31-40** | 23 | 11.9 | 11 | 10.3 | 12 | 14.1 |
| **41-50** | 28 | 14.4 | 17 | 15.9 | 10 | 11.8 |
| **51-60** | 48 | 24.7 | 28 | 26.2 | 20 | 23.5 |
| **61-70** | 36 | 18.6 | 25 | 23.4 | 11 | 12.9 |
| **> 70** | 18 | 9.3 | 9 | 8.4 | 8 | 9.4 |

*Two respondents reported being divers.

The distribution of reported disease onset is presented in Table 2. Most patients (102 out of 194, 52.6 %) experienced initial symptoms within the first three decades of life. However, 73 patients (37.6 %) noted their first symptoms after the age of 40. Gender differences were observed in the reported onset, with female patients most frequently experiencing onset in the fifth decade of life, whereas male patients most frequently reported onset in the first decade.

**Table 2.** Distribution of patient reported age of onset.

| Onset distribution | All | | Female | | Male | |
|---|---|---|---|---|---|---|
| [years of age] | N=194* | % | N=107 | % | N=85 | % |
| **0_10** | 44 | 22.7 | 15 | 14.0 | 29 | 34.1 |
| **11_20** | 32 | 16.5 | 19 | 17.8 | 12 | 14.1 |
| **21_30** | 26 | 13.4 | 15 | 14.0 | 11 | 12.9 |
| **31_40** | 19 | 9.8 | 10 | 9.4 | 9 | 10.6 |
| **41_50** | 41 | 21.1 | 30 | 28.0 | 11 | 12.9 |
| **51-60** | 30 | 15.5 | 17 | 15.9 | 12 | 14.1 |
| **> 61** | 2 | 1.0 | 1 | 0.9 | 1 | 1.2 |

*Two respondents reported being divers.

### *3.2. Vocational Training*

A total of 173 out of 194 patients (89.2 %) provided responses regarding their vocational training experience. Among the 21 non-respondents 14 were over 50 years of age. The majority of respondents (141 out of 173; 81.5 %) reported no issues during their planned vocational training, without a gender

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difference. Some patients (9 out of 173; 5.2%) had not yet started vocational training, including two individuals who were unable to begin their desired program. Additionally, 17 patients (9.8 %) were unable to start their intended vocational training. Three patients had to terminate their first training program (1.7 %). Eight patients reported a prolonged period of more than one year necessary for their vocational training compared to the initial planning. One patient had to change her vocational training more than twice.

### *3.3. Professional Life*

A total of 157 out of 194 patients (80.9%) provided responses regarding their professional life experiences. Six patients who had not yet started vocational training did not respond to this question. The majority (109 out of 157; 69.4 %) reported no significant difficulties in their professional careers (67.4 % of females, 72.5 % of males). However, 28 patients (17.8 %) had to retire early, including 5 individuals who were unable to start their professional careers despite completing vocational training. Among patients 40 years and older 23 out of 130 (17.7 %) had to retire early due to their MD/CRD. In patients 50 years and older this proportion slightly increased with 21 out of 102 (20.6 %). Due to visual-related problems, 9 patients had to change their career more than twice. Fifteen patients (9.6 %) experienced long-term unemployment lasting more than one year.

**Table 3.** Macular dystrophy / cone-rod dystrophy query results.

| | *All* | | *Female* | | *Male* | |
|---|---|---|---|---|---|---|
| | N= | % | N= | % | N= | % |
| Contacted without return of letter | 281 | | | | | |
| Response | 194 | 69.0 | 107 | 55.2 | 85 | 43.8 |
| ***Vocational training response*** | 173 | 89.2 | 91 | 85.1 | 80 | 94.1 |
| No problems | 141 | 81.5 | 74 | 81.3 | 66 | 82.5 |
| Not started | 9 | 5.2 | 4 | 4.4 | 5 | 6.3 |
| Unable to start desired training | 17 | 9.8 | 9 | 9.9 | 7 | 8.8 |
| Termination of desired training | 3 | 1.7 | 2 | 2.2 | 1 | 1.3 |
| Delay > 1 year | 8 | 4.6 | 3 | 3.3 | 5 | 6.3 |
| Changing more than twice | 1 | 0.3 | 1 | 1.1 | 0 | 0.0 |
| ***Professional career responses*** | 157 | 80.9 | 86 | 80.4 | 69 | 81.2 |
| No problems | 109 | 69.4 | 58 | 67.4 | 50 | 72.5 |
| Early retirement all | 28 | 17.8 | 15 | 17.4 | 12 | 17.4 |
| Early retirement > 40 yoa | 23 (130) | 17.7 | 12 (79) | 15.2 | 10 (49) | 20.4 |
| Early retirement > 50 yoa | 21 (102) | 20.6 | 12 (62) | 19.4 | 9 (39) | 23.1 |
| Changing more than twice | 9 | 5.7 | 5 | 5.8 | 4 | 5.8 |
| Unemployed > 1 y | 15 | 9.6 | 10 | 11.6 | 5 | 7.2 |
| ***Social disadvantages responses*** | 190 | 97.9 | 106 | 99.1 | 82 | 96.5 |
| None | 53 | 27.9 | 30 | 28.3 | 22 | 26.8 |
| Limitation in public life | 104 | 54.7 | 59 | 55.7 | 44 | 53.7 |
| Financial restrictions | 35 | 18.4 | 19 | 17.9 | 16 | 19.5 |
| Financial support acquired | 54 | 28.4 | 29 | 27.3 | 24 | 29.3 |
| Financial support sufficient | 24 (54) | 44.4 | 13 (29) | 44.8 | 11 (24) | 45.8 |
| Financial support insufficient | 30 (54) | 55.6 | 16 (29) | 55.2 | 13 (24) | 54.2 |
| ***Family life disadvantages resp.*** | 162 | 83.5 | 91 | 85.1 | 69 | 81.2 |

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| | | | | | | |
|---|---|---|---|---|---|---|
| None | 142 | 87.7 | 76 | 83.5 | 64 | 92.8 |
| IRD reason end of partnership | 7 | 4.3 | 5 | 5.5 | 2 | 2.9 |
| No children due to IRD | 16 | 9.9 | 11 | 12.1 | 5 | 7.2 |
| ***Personal impairment responses*** | 190 | 97.9 | 107 | 100.0 | 81 | 95.3 |
| None | 23 | 12.1 | 12 | 11.2 | 11 | 13.6 |
| Anxiety | 141 | 74.2 | 81 | 75.7 | 58 | 71.6 |
| Depression | 30 | 15.8 | 15 | 14.0 | 14 | 17.3 |
| Other | 80 | 42.1 | 45 | 42.1 | 35 | 43.2 |
| ***Ophthalmic care responses*** | 190 | 97.9 | 105 | 98.1 | 82 | 96.5 |
| Appropriate time | 114 | 60.0 | 65 | 61.9 | 47 | 56.6 |
| Delay all | 65 | 34.2 | 34 | 32.4 | 31 | 37.8 |
| Delay > 2y | 29 | 15.3 | 13 | 12.4 | 16 | 19.3 |
| Delay > 5y | 36 | 18.9 | 21 | 20.0 | 15 | 18.1 |
| Misdiagnosis | 42 | 22.1 | 21 | 20.0 | 21 | 25.3 |
| Insufficient psychological support | 68 | 35.8 | 41 | 39.0 | 27 | 32.5 |

### *3.4. Social Disadvantages*

A total of 190 out of 194 patients (97.9 %) provided responses regarding social disadvantages. A minority of 53 patients (27.9 %) reported experiencing no social disadvantages. However, the majority noted limitations in public life participation as a consequence of their MD/CRD (104 patients; 54.7 %). One fifth complained about financial restrictions (35 patients; 18.4 %). Among those reporting financial difficulties 15 out of 28 who retired early, 9 out of 15 who experienced unemployment of more than a year and 3 out of 9 who had to change careers more than twice, highlighting the impact of employment on financial independence. Additionally, 54 out of 194 patients (28.4%) received financial or other forms of support. Of these, 24 patients (44.4%) considered the support sufficient, while 30 patients (55.6 %) found it insufficient, independent of age, disease duration and gender. Notably, 8 out of 34 patients (23.5 %) who reported financial restrictions did not receive any form of financial support.

### *3.5. Family Life*

A total of 162 out of 194 patients (83.5%) provided responses regarding possible disadvantages in their family life. A vast majority, 142 out of 162 patients (87.7 %), reported no negative impact of MD/CRD on their familial situation, with a higher percentage among males (92.8 %) compared to females (83.5 %). This difference was independent of age. In seven cases, MD/CRD was cited as the reason for the end of a partnership. Additionally, 16 out of 162 patients (9.9 %) reported that MD/CRD influenced their decision to forgo having children. This group included two individuals who otherwise reported no disadvantages in their familial life.

### *3.6. Personal Impairments*

A total of 190 out of 194 patients (97.9%) provided responses regarding personal impairments. Among them, only 23 patients (12.1 %) reported feeling no personal impairments due to MD/CRD, without gender difference. The most frequently reported concern was anxiety about the future due to disease progression, affecting 141 out of 190 patients (74.2%), slightly more frequently females. Depression was reported by 30 patients (15.8 %), slightly more frequently males. Additionally, 80 patients (42.1 %) described experiencing other personal impairments related to MD/CRD.

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*3.7. Ophthalmic Care*

A total of 190 out of 194 patients (97.9 %) provided responses regarding their ophthalmological care. A slight majority, 114 out of 190 patients (60.0 %) felt that their ophthalmologic and molecular genetic diagnosis was provided within an appropriate time frame. However, even among these, 5 patients (4.4 %) still perceived a delay in their MD/CRD diagnosis. A significant delay between the initial ophthalmological examination due to MD/CRD symptoms and the confirmed diagnosis was reported by 65 out of 190 patients (34.2 %). Major concerns regarding the diagnostic process included a delay of more than two years in 29 patients (15.3 %) and more than five years in 36 patients (19.0 %). Additionally, 42 patients (22.1 %) experienced one or more misdiagnoses before receiving a confirmed MD/CRD diagnosis. More than one-third of the patients, 68 out of 190 (35.8 %), expressed that they lacked psychological support throughout the progression of their disease.

*3.8. Comparison with RP Patients*

In a previous study using the same methodology patients with molecularly confirmed retinitis pigmentosa (RP) were contacted and responded with a similar response rate (RP 67.2 %, MD/CDR 69.0 %) [19]. Patient reported impact was similar for RP and MD/CRD in most aspects, however, some differences need to be noted. In the RP study, patients were younger compared to the MD/CDR study (under 51 years of age: RP: 67.3 %, MD/CRD: 47.4 %). Disease onset was later in life in MD/CRD compared to RP, with first symptoms noted after the age of 40 in 17.3 % of RP patients but 37.6 % in MD/CDR. The Differences in a later manifestation of MD/CRD compared to RP affected women more than men. Regarding vocational training no gender difference was observed in MD/CRD in contrast to a female advantage in RP. The response rate regarding professional life was lower in MD/CRD (RP: 88.3 %; MD/CRD: 80.9 %). Early retirement rates were higher in RP patients (RP: 26.6 %, MD/CRD: 17.8 %) especially in patients older than 50 years of age (RP: 50.0 %, MD/CRD: 20.6 %). Social support was more frequently perceived as sufficient by RP patients (RP: 60.0 %, MD/CRD: 44.4 %). The negative impact on family life was lower in MD/CDR (no impact: RP: 78.6 %, MD/CRD: 87.7 %) while the gender difference was similar. In contrast to RP, there was no gender difference regarding personal impairments in MD/CRD.

## 4. Discussion

To the best of our knowledge, this is the first study evaluating the patient-reported impact of MD/CRD on daily life in a large cohort of genetically confirmed MD/CRD patients. The response rate of 69.0% was similar to our previous study in RP patients and higher than that of a similar study in IRD patients [13,19]. This may be due to the selection of patients from two specialized IRD care centers, the short questionnaire or the detailed anonymization measures.

Symptom onset of MD/CRD was noted by most patients in this study within the first three decades of life. In comparison to RP, the onset of MD/CRD was generally later and more patients noted an onset after 40 years of age [19]. Similar to RP, the impact on daily life was limited in the early stages, as they interfered with vocational training in only 13.8 % of patients. Although personal career aspirations do not differ between individuals with and without visual impairment [22], it can be expected that visual loss influences patients to select vocational paths according to their personal abilities. A delay of vocational training contributes to reduced lifetime income [10].

Progression of MD/CRD is associated with increasing challenges in professional life. Patients may be unable to enter the workforce despite completing vocational training or they are forced to leave their jobs. With increasing age, the likelihood of early retirement increased slowly. In contrast to RP patients, rates of early retirement were much lower both in patients 40 years and older (MD/CRD: 17.7 %, RP: 39.8 %) and in patients 50 years and older (MD/CRD: 20.6 %, RP: 50.0 %) [19]. The comparison between both studies must take into consideration, that patients in the RP survey had a younger mean age and a higher response rate. Early loss of central vision in MD/CRD would be expected to interfere with many professional tasks. Nevertheless, MD/CRD patients have a lower

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risk of early retirement when compared to RP patients, which may be explained by the availability of modern low vision aids and rehabilitation measures that help compensate for central vision loss and allow patients to remain employed for a longer period of time.

In contrast, extended periods of unemployment due to delayed vocational training or difficulties securing employment were similar between MD/CRD (9.6 %) and RP (13.3 %). Unemployment due to visual impairment further contributes to reduced lifetime income [10,23].

Social disadvantages as a consequence of MD/CRD manifest primarily in reduced participation in public life (54.7 %). Social isolation has been observed in more than half of individuals in previous IRD studies [13,24]. In RP the risk of bumping into people or objects, challenges with stairs, and an increased likelihood of falls limit mobility and lead to self-imposed isolation [25,26]. Reduced activity and other behavioral changes have been observed already in children with visual impairment [27,28]. Syndromic MD/CRD most likely has an additional impact on the participation in public life [29], but has not been specifically studied in MD/CRD associated syndromes.

Financial restrictions due to MD/CRD (18.4 %) were in a similar frequency as in our RP study (19.4 %), but less frequent compared to other IRD studies (44 %) [13]. Approximately one-third of patients received financial or other forms of support. In contrast to RP patients [19], the majority expressed dissatisfaction with the level of support received, and 23.5 % of those reporting financial difficulties had not received any assistance. This underscores the need for a thorough evaluation and improvement of social and financial support systems to better address patients' needs in all disorders associated with impaired visual function [30,31].

Similar to RP, nearly 80% of patients reported no disadvantages in their family life. It can be expected, that financial challenges affected personal and familiar circumstances. One tenth of the patients chose not to have children due to their MD/CRD. Many of these decisions were likely made prior to molecular genetic testing, underscoring the importance of timely genetic testing and appropriate counselling.

Anxiety and depression are frequently reported in association with IRDs [13,24,32–34]. In the present study, 74.2 % of patients expressed anxiety about their future situation. Previous studies on RP and IRD have reported anxiety rates ranging from 65.6 to 86 % [13,19,25,35]. Similar to detailed anxiety assessments in RP [32,36,37] anxiety rates were independent of patient age or disease duration.

Depression suspected as a consequence of MD/CRD was reported by 15.8 % of patients, slightly lower compared to our previous study with RP patients (24.2 %) [19], and markedly lower compared to studies in non-syndromic (65 %) or syndromic RP (86.2 %) [13,38]. It is important to note that these figures are based on patient-reported diagnoses without formal psychological verification. Different methods have been used to estimate the frequency and severity of depression in RP and other ocular disorders (e.g. specified questionnaires, ICD-10 codes) [26,39–41]. This variation in methodology probably affects the frequency of the diagnosis. The limited access to psychologic care for IRD patients most likely leads to an underdiagnosis of depression.

It remains challenging to determine whether visual impairment or unemployment is the primary cause of depression or if pre-existing depression increases the risk of unemployment in patients with visual impairment. While RP patients with depression have lower employment rates [39], unemployment is associated with higher rates of depression [33]. Depression associated with visual impairment is common across a wide range of ophthalmic disorders [42–44]. Depression itself can exacerbate financial difficulties and negatively impact social life [45].

For decades, IRD patients have complained about the difficulties of the patient journey including the time delay between the symptom onset and the clinical diagnosis, the frequency of initial misdiagnosis as well as the lack of psychological support after establishing the diagnosis and throughout disease progression. In our studies, 34.2 % of MD/CDR patients and 28.6 % of RP patients reported a diagnostic delay of more than two years. Though one would hope that the widespread availability of retinal imaging and novel methods facilitating extensive genetic testing would shorten

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the diagnostic process, recent studies still emphasize the need to further improve the patient pathway in diagnosing IRDs [15,46,47].

The frequency of misdiagnosis is difficult to establish. It is nearly impossible for patients to distinguish between true diagnostic errors (e.g. mistaking MD/CRD for acquired disorders) and variations in terminology used by different ophthalmologists (e.g. macular dystrophy, Stargardt disease, cone-rod-dystrophy). Unfortunately, the lengthy patient journey to a definitive diagnosis suggests that many patients did at least receive one incorrect diagnosis. This has been recognized and diagnostic guidelines for IRDs were recently revised in Germany [48]. Though the resources for improving patient care are in place, a continuous need to raise awareness among healthcare professionals about the importance of early and accurate diagnosis for IRD patients will remain a task for all health care providers associated with IRDs. In addition, discipline-specific directives and guidelines for the clinical and genetic diagnostic process would reduce the psychologic burden associated with unexplained visual loss.

More than one-third of MD/CRD and RP patients missed psychological support after diagnosis and throughout the progression of disease. Continuous psychological support should be an integral part of patient care, provided by a multidisciplinary team [34,49,50]. Such support could reduce the frequency of anxiety and depression and improve patients' social interactions and work capabilities. At least in Germany, the number of providers of psychological support experienced with IRD patients is limited. An interdisciplinary exchange between the separate health care providers involved in care of IRD patients will be important to finally establish an uninterrupted patient care.

The present study has several limitations. Firstly, the limitation to patients with genetically confirmed MD/CRD excludes approximately 30% of MD/CRD patients for whom a genetic cause cannot yet be defined. Molecular genetic testing is usually initiated at specialized centers for rare retinal disease in Germany [51], therefore, the study population may represent individuals who either had the good fortune or personal motivation in pursuing their diagnosis to access these centers. Secondly, due to the anonymity of the survey we cannot exclude that some patients were contacted twice because they were examined at both participating centers. However, given the geographical distance between the two centers, the number of such cases is expected to be rather low and should not significantly impact the study's findings. Thirdly, this study employed a concise questionnaire to assess the impact of MD/CRD on various aspects of patients' lives. To preserve anonymity, the survey did not collect detailed personal comments. While this approach enabled the inclusion of a large patient cohort, it limited the depth of individual responses. In addition, a comparison with clinical findings was not possible. Fourthly, there possibly is a difference between the experience of patients born into a family with other affected family members versus patients without other affected family members. For the latter as well as the family, all experience associated with MD/CRD is novel. Due to anonymization, we could not evaluate this aspect in the present study, but this aspect should be included in future research. Finally, a more detailed analysis especially of anxiety and depression with validated methodology would be desirable. However, these methodologies are more time consuming and often require direct personal communication [36]. This limits the number of study patients due to research resources as well as availability of patients for longer test periods or traveling to research centers. The present study provides a first and valuable overview of current challenges faced by patients with genetically confirmed MD/CRD in Germany.

## 5. Conclusions

In several aspects, patient reported impact of disease was comparable between MD/CRD and RP patients. There was a marked difference in early retirement rates especially in patients older than 50 years of age (MD/CRD: 20.6 %, RP: 50.0 %) [19]. Possible contributing factors are a later disease onset in MD/CRD patients in these two studies, the individual choice of profession, the available means for rehabilitation in a specific profession as well as a difference in severity of visual disability between MD/CRD as well as RP.

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In conclusion, the studies on current challenges faced by patients with genetically confirmed MD/CRD or RP serve as both a guide for key areas requiring attention for decision within the health care system and a possible starting point for future research. There remains a continuous need to enhance a time-efficient diagnostic process for IRD patients. A confirmed diagnosis is mandatory for initiating rehabilitation, psychologic support, and financial assistance. As a possible advantage for the social care system, improved diagnostic processes, rehabilitation and patient care should have an impact on the high health care and societal costs of IRDs [52,53].

Future research should focus on a more detailed analysis of the quality of life in correlation with visual function, the professional confirmation of anxiety and depression and the definition of standards for psychological care in MD/CRD patients. Strategies for a multidisciplinary exchange need to be established and continuously re-evaluated to define a patient pathway beyond the clinical and genetical diagnosis, including a patient-focused individualized rehabilitation process and timely response addressing social needs [50,51,54–56].

**Supplementary Materials:** The following supporting information can be downloaded at the website of this paper posted on Preprints.org, S1: Questionnaire: original German version, S2: Questionnaire: translated English version

**Author Contributions:** Conceptualization, N.ZL., G.S. and U.K.; methodology, N.ZL., G.S. and U.K.; software, N.ZL. and U.K.; validation, N.ZL., H.S., B.H.F.W., G.S. and U.K.; formal analysis, N.ZL., H.S., B.H.F.W., G.S. and U.K.; investigation, N.ZL., B.L., G.S. and U.K.; resources, B.L., G.S. and U.K.; data curation, N.ZL., B.L. and U.K.; writing—original draft preparation, N.ZL. and U.K.; writing—review and editing, N.ZL., B.L., H.S., B.H.F.W., G.S. and U.K.; visualization, N.ZL. and U.K.; supervision, U.K.; project administration, B.L.; funding acquisition, G.S. and U.K. All authors have read and agreed to the published version of the manuscript.

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**Institutional Review Board Statement:** The study was conducted in accordance with the Declaration of Helsinki and approved by the respective ethics committees of the North Rhine (ID: 2024037, date of approval: 27.02.2024) and Westphalia-Lippe (ID 2024-234-b-S, date of approval: 09.04.2024) medical associations.

**Informed Consent Statement:** Patient consent was waived by the ethic committees to preserve anonymization of the query.

**Data Availability Statement:** The data presented in this study are available on request from the corresponding author due to legal restrictions.

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 doi:10.20944/preprints202509.1998.v1



## Abbreviations

The following abbreviations are used in this manuscript:

| | |
|---|---|
| AMD | Age-related macular degeneration |
| CDR | Cone-rod dystrophy |
| DR | Diabetic retinopathy |
| IRD | Inherited retinal dystrophies |
| MD | Macular dystrophy |
| RP | Retinitis pigmentosa |

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