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The Visual Impairment of Inherited Retinal Diseases in Portugal as per the National Table of Disabilities

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Purpose: To evaluate the visual impairment of patients with inherited retinal diseases (IRDs), as per the national table of disabilities (TNI).

Design: Retrospective, single-center cohort study.

Participants: Patients with a clinical diagnosis of IRD were recruited at a referral center in Portugal.

Methods: Demographics and clinical data were collected from each individual patient file. The estimated visual disability coefficient was calculated through the evaluation of 7 graduated categories: orbital or eyelid deformities, low vision, visual field change, loss of bi-foveolar fixation, oculomotor palsy, photophobia, and chronic conjunctivitis. The TNI provides minimum and maximum disability values for numerous conditions within each category, which were summed to calculate an overall summary disability coefficient for each patient.

Main Outcome Measures: Demographic/clinical and estimated minimum and maximum visual disability coefficient according to the TNI for each patient.

Results: This study included 253 patients from 214 families, aged 3 to 80 years, with a mean age of 39.8 ± 20.0 years. The mean estimated minimum and maximum visual disability coefficients as per the TNI were 0.6 ± 0.4 and 0.7 ± 0.4, respectively. The low vision was the single most frequent contributor category (21.7%) present in the calculation of visual impairment. Low vision and visual field changes were the most frequent double combination (18.2%), and the addition of loss of bi-foveolar fixation was the most frequent triple combination (8.3%).

Conclusions: This study found that IRD patients had a significant visual disability, with the majority having a disability coefficient ≥0.6, which would qualify them for a "multipurpose disability medical certificate."

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Inherited retinal diseases (IRDs) are a group of degenerative disorders of the retina, characterized by clinical and genetic heterogeneity, which can lead to blindness from birth to late middle age.[1,2] Despite being rare, they are the most common reason for low vision certification amongst the working-age population in some countries, surpassing age-related macular degeneration, diabetic retinopathy, and glaucoma.[3–5] Economic costs and reduction in the quality of life in IRD are significant and have already been described in some countries.[6–9]

The characterization of visual impairment attributed to IRDs is scarce in the literature.[3] There are few data mentioning visual impairment degrees according to age or subtypes of IRDs, as well as descriptions of their main contributors. Knowing the target population is essential to better plan support measures.

The medical and legal assessment of changes in psychophysical integrity is a matter of particular importance. The National Table of Disability due to Work Accidents and Occupational Diseases (TNI), approved by Decree-Law No. 352/2007 of 23 October in Portugal, is used to assess work disability and is intended to protect workers of Labour Law.[10]

This table resulted from a review and update of the previous TNI, carried out for 6 years before its approval, by a permanent commission. This included representatives from various ministries, public services, the Portuguese Insurance Association, labour courts, the National Association of Disabled People Injured at Work, the Portuguese Society of Occupational Medicine, employer associations and trade union associations with seats on the Permanent Commission for Social Coordination, and the National Council for the Rehabilitation and Integration of People with Disabilities.[11]

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The percentages of disability applicable to certain pathologies were adjusted because of technical-scientific work, taking into account the dynamics of the national medical-legal panorama, and what is recommended in several European tables.[12]

With the adoption of this table, the aim was to achieve greater legal precision and safeguard the equality of citizens before the law, respecting the principle that similar repercussions on activities of daily living have identical legal consequences.[10] Therefore, the application to people with hereditary diseases can help us to better understand the quality of life of these patients, in terms of access to socioeconomic care.

The aim of this study was to evaluate the visual impairment of IRD patients as per the TNI in order to better understand this specific population, their visual disabilities, and socioeconomic implications.

Methods

Study Design

A cross-sectional cohort study was conducted on consecutive patients with a clinical diagnosis of IRD, who were observed at the ophthalmology department at the Centro Hospitalar Universitário de Santo António, previously denominated by Centro Hospitalar Universitário do Porto, between January 2021 and May 2023. The study complied with the tenets of the Declaration of Helsinki for biomedical research. An institutional review board/ethics committee approval was obtained. Because the authors ensured that all patients' anonymity was carefully protected, the use of a written and informed consent form was waived by the institutional review board.

Participants

Patients were identified using an electronic medical database created in 2021 and shared between ophthalmologists dedicated to the medical retina, paediatric ophthalmology, and low-vision assessment in Centro Hospitalar Universitário de Santo António. This database includes all patients with IRDs, respective clinical diagnostic subgroups and molecular test results (known, ongoing and not performed). Centro Hospitalar Universitário de Santo António is one of 5 public tertiary hospitals nationwide,[13] and most IRD patients are followed up at this level of care. It is located in the north of the country, together with another tertiary hospital, sharing an area of influence of around 3 428 675 people (based on Census 2011).[14]

The inclusion criteria were clinical diagnosis of IRD and known or ongoing genetic testing results. No patient identified was excluded.

Parameters

Data were collected from the institution's own clinical records for each patient identified from the electronic medical database mentioned previously. The parameters analysed in this study are as follows:

- demographic characteristics of the study's population (gender and age)
- clinical features (the age of onset of symptoms and clinical diagnosis based on classification covered by the national, web-based IRD registry[15]; family history; genetic testing)
- estimated minimum and maximum visual disability coefficient according to the TNI for each patient, calculated by an independent grader (A.M.) after a systematic chart review
- the presence of any low vision aids or rehabilitation intervention measures

Age was divided into subgroups as socioeconomic criteria from the "PORDATA – Statistics about Portugal and Europe" website: young (if $\leq$ 14 years), working-age (between 15 and 64 years), and elderly (if $\geq$ 65 years).

The estimated visual disability coefficient calculation included the evaluation of the following 7 graduated categories available in the ophthalmology chapter of the TNI:

1) Orbital or eyelid deformities
2) Low vision
3) Visual field change
4) Loss of bi-foveolar fixation
5) Oculomotor palsy
6) Photophobia
7) Chronic conjunctivitis

Each category is subdivided into subcategories graded by fixed value ranges with minimum and maximum assignable values possible.[10] Supplement Document 1 shows the ophthalmology chapter of Portuguese TNI translated into English for better visualization of subcategories and how are range graded.

An independent grader (A.M.) did a chart review and systematically collected data on the 7 categories for each patient, and then, using that data, she assigned the minimum and maximum estimated visual disability coefficient possible for each patient, resulting from the sum of minimum and maximum values obtained from each category, respectively, applying Portuguese TNI. Patients with missing data for a category were assumed to have a disability score of zero for that category.

For example, a patient with visual acuity of 0.5 (decimal scale) in both eyes and a concentric decrease in the visual field below 10º in both eyes has the following:

- a minimum estimated visual disability coefficient of 0.73: sum of 0.02 (minimum value assignable because of this level of low vision) with 0.71 (minimum value assignable because of visual field change)
- a maximum estimated visual disability coefficient of 0.84: sum of 0.04 (maximum value assignable because of this level of low vision) + 0.80 (value assignable because of visual field change)

When the minimum or maximum estimated visual disability coefficient exceeded 1, it was rounded down to 1 (maximum possible value).

For example, a patient with visual acuity of 0.05 (decimal scale) in both eyes and a concentric decrease in the visual field below 10º in both eyes has the following:

- a minimum estimated visual disability coefficient of 1: sum of 0.95 (minimum value assignable because of low vision) with 0.71 (minimum value assignable because of visual field change) exceeds 1
- a maximum estimated visual disability coefficient of 1: sum of 0.95 (maximum value assignable because of low

vision) + 0.80 (value assignable because of visual field change) exceeds 1

Therefore, the visual disability coefficient ranges from 0 to 1, and, when multiplicated by 100, it corresponds to the percentage of disability, used in Portugal to obtain a multipurpose disability medical certificate. In this study, we chose to use both the minimum and maximum values because the official final value can vary according to the responsible doctor who attributes the final coefficient.

Statistical Analysis

Statistical analysis was performed using the SPSS program (IBM SPSS Statistics, version 28.0.1.0 for Mac). The normality of the variables was evaluated by the Kolmogorov-Smirnov test. Descriptive statistics are shown as mean ± standard deviation. For categorical variables, descriptive statistics are shown as absolute and relative frequencies. The comparison between continuous variables was performed using the Mann-Whitney and Kruskal-Wallis tests. Significance values were adjusted by Bonferroni correction in multivariate comparative tests. Spearman's bivariate correlation test was used to study linear correlations. A P value < 0.05 was considered statistically significant.

Results

Demographic Data

This study included 253 patients from 214 families, 48.6% male and 51.8% female, aged 3 to 80 years, with a mean age of 39.8 ± 20.0 years, with 33 (13.0%) cases of young age (< 14 years), 189 (74.7%) cases of working-age (between 15 and 64 years), and 31 (12.3%) elderly people (≥ 65 years).

Clinical Features

The most frequent diagnosis was nonsyndromic retinitis pigmentosa (ORPHA 791) with 36.8% of patients, followed by cone and cone-rod dystrophies (ORPHA 1872) with 15.0%, syndromic retinitis pigmentosa (ORPHA 519325) with 14.6%, and Stargardt disease (ORPHA 827) with 9.1%. Distribution by clinical diagnoses including the respective ORPHA numbers is shown in Table 1.

The onset age of symptoms was variable: 10 (3.9%) cases at birth, 73 (28.9%) < 5 years of age, 67 (26.5%) between 6 and 10 years, 33 (13.0%) between 11 and 20 years, 28 (11.1%) between 21 and 30 years, 30 (11.9%) between 31 and 50 years, and 12 (4.7%) > 51 years.

Visual Disability

Data for disability coefficient estimates were extracted from the chart review for all 253 patients, although 126 patients had missing data for the visual field category and 2 had missing data for the low-vision category (Supplemental Document 2).

The mean estimated minimum and maximum visual disability coefficients as per the TNI were 0.6 ± 0.4 and 0.7 ± 0.4, respectively. In 62.5% and 65.6% of patients, the estimated minimum and maximum visual disability coefficients were ≥ 0.6 (cutoff for social benefits in Portugal), respectively. In 17% and 36.8% of patients, the estimated minimum and maximum visual disability coefficients were 1.0 (meaning 100% disability), respectively.

The mean number of categories per patient contributing to their degree of visual disability was 1.8 ± 0.9. The diagram in Figure 1 represents the isolated or combined contribution of the categories, with the absence of the orbital or eyelid deformities that were identified as concomitant in 3 cases (1.2%), oculomotor palsy in 1 case (0.4%), and chronic conjunctivitis in none (0%). The low vision was the single most frequent contributor category (21.7%) present in the calculation of visual impairment. Low vision with visual field changes was the most frequent double combination (18.2%), and the addition of loss of bi-foveolar fixation was the most frequent triple combination (8.3%). In 7.5% of cases, no category contributed to the degree of disability, being asymptomatic patients, or having symptoms not included in the TNI (e.g., nyctalopia).

The median estimated minimum ($P = 0.229$, comparing the 3 groups) and maximum ($P = 0.592$, comparing the 3 groups) visual disability coefficients were similar between age groups (young, working-age, and elderly patients). The median estimated minimum ($r = 0.075$; $P = 0.232$) and maximum ($r = 0.070$; $P = 0.269$) visual disability coefficients had no significant correlation with age.

A graphical representation of the visual disability coefficient by age group and the 2 main categories contributors (low vision and visual field changes) to the final value can be seen in Figure 2. The median estimated minimum ($P = 0.177$, comparing the 3 groups) and maximum ($P = 0.117$, comparing the 3 groups) low vision contributions to the final value were similar between age groups. The median estimated minimum visual field change was different when comparing the 3 groups ($P = 0.040$), with pairwise comparisons suggesting that the visual field change contributions to the final value were lower in patients < 14 years than in patients between 15 and 64 years ($P = 0.033$) and patients > 65 years ($P = 0.035$). The median estimated maximum visual field change was also different when comparing the 3 groups ($P = 0.040$), with pairwise comparisons suggesting that the visual field change contributions to the final value were lower in patients < 14 years of age compared to patients between 15 and 64 years of age ($P = 0.035$) but not compared to patients > 65 years of age ($P = 0.062$).

The median estimated minimum ($P = 0.003$, comparing the 6 groups) and maximum ($P = 0.030$, comparing the 6 groups) visual disability coefficients were different between diagnosis groups ("isolated progressive inherited retinal disorder," "syndromic inherited retinal disorder," "isolated stationary inherited retinal disorder," "chorioretinal dystrophies," "inherited vitreous dystrophies," and "other rare disorders of the posterior segment of the eye"). Patients with "isolated progressive inherited retinal disorders" had a higher median estimated minimum ($P = 0.001$) and maximum ($P = 0.004$) visual disability coefficient than patients with "other rare disorders of the posterior segment of the eye." A graphical representation of the visual disability coefficient by diagnosis groups can be seen in Figure 3.

Table 1. Distribution of Clinical Diagnoses Including the Respective ORPHA Numbers in the Cohort

Inherited Retinal Dystrophies (ORPHA 71862)	Number	%
Isolated progressive inherited retinal disorder (ORPHA 519306)	171	67.6
Nonsyndromic retinitis pigmentosa (ORPHA 791)	93	36.8
Cone and cone-rod dystrophy (ORPHA 1872)	38	15.0
Stargardt disease (ORPHA 827)	23	9.1
Leber congenital amaurosis (ORPHA 65)	7	2.8
Best vitelliform macular dystrophy (ORPHA 1243)	3	1.2
Pattern dystrophy (ORPHA 63454)	5	2.0
Retinitis punctata albescens (ORPHA 52427)	1	0.4
Sorsby macular dystrophy (ORPHA 59181)	1	0.4
Syndromic inherited retinal disorder (ORPHA 519325)	44	17.4
Syndromic retinitis pigmentosa (ORPHA 519325)	37	14.6
Maternally inherited diabetes and deafness (ORPHA 225)	2	0.8
Pseudoxanthoma elasticum (ORPHA 758)	1	0.4
Hypotrichosis with juvenile macular dystrophy (ORPHA 1573)	2	0.8
Papillorenal syndrome (ORPHA 1475)	1	0.4
Sjogren-Larsson syndrome (ORPHA 816)	1	0.4
Isolated stationary inherited retinal disorder (ORPHA 519319)	9	3.6
Achromatopsia (ORPHA 49382)	6	2.4
Early-onset "drusenoid" macular dystrophies (ORPHA 75376)	2	0.8
Fundus albipunctatus (ORPHA 227796)	1	0.4
Chorioretinal dystrophies (ORPHA 519300)	6	2.4
Choroideremia (ORPHA 180)	3	1.2
Gyrate atrophy of choroid and retina (ORPHA 414)	3	1.2
Inherited vitreous dystrophies (ORPHA 519304)	4	1.6
X-linked retinoschisis (ORPHA 792)	4	1.6
Other rare disorders of the posterior segment of the eye (ORPHA 519311)	19	7.5
Ocular and oculocutaneous (ORPHA 284804 e 55) albinism	15	5.9
Foveal hypoplasia (ORPHA 519398)	3	1.2
Posterior microphthalmos	1	0.4

Bold corresponds to items (groups of diseases) that have dependences.

Within the 3 most frequent nonsyndromic diagnoses, the median estimated minimum ($P = 0.533$, comparing the 3 groups) and maximum ($P = 0.533$, comparing the 3 groups) visual disability coefficient was similar between them but not low vision and visual field change contribution for the final value. Patients with nonsyndromic retinitis pigmentosa had a lower minimum ($P < 0.001$) and maximum ($P < 0.001$) low vision contribution and a higher minimum ($P < 0.001$) and maximum ($P < 0.001$) visual field change contribution than patients with cone and cone-rod dystrophy or Stargardt disease. A graphical representation of the visual disability coefficient by the 3 most frequent nonsyndromic diagnoses and the 2 main categories contributors (low vision and visual field changes) to the final value can be seen in Figure 4.

Low vision aids and some rehabilitation intervention measures were identified in 33.6% of patients. Patients with these supports were younger ($P < 0.001$) and had a higher estimated minimum ($P < 0.001$) and maximum ($P < 0.001$) visual disability coefficients than patients without any support.

Discussion

This is the first study evaluating the visual impairment of IRD patients as per the TNI. Knowing the target population is essential for developing public health and social measures.

Overall, this study found that IRD patients had an important visual disability. The estimated minimum and maximum visual disability coefficients were ≥ 0.6 in 62.5% and 65.6% of patients, respectively. The cutoff of 0.6 in Portugal means that they could benefit from a "multipurpose disability medical certificate." This certificate provides for the attribution of multiple social, fiscal, and economic benefits to its holders, namely exemption from user fees in the National Health Service, priority assistance, exemption from car tax, nonurgent transport of patients, protection and social support, and higher education scholarships.[16]

Regarding age, we already know that IRDs also affect young people. In our study, 72.3% of those affected were < 20 years of age. However, there were no correlations between visual disability coefficients and age, meaning that when these diseases begin early, the disability is already high. We also showed that the contributing symptoms are different between age groups. The low vision was a determinant for the final coefficient with any age group, but the visual field change was less determinant if patients were < 14 years of age (Fig 2). Visual field changes may be underestimated at a young age because these patients can have more difficulty performing visual field tests or the changes detected cannot be so valued in the TNI. In fact,

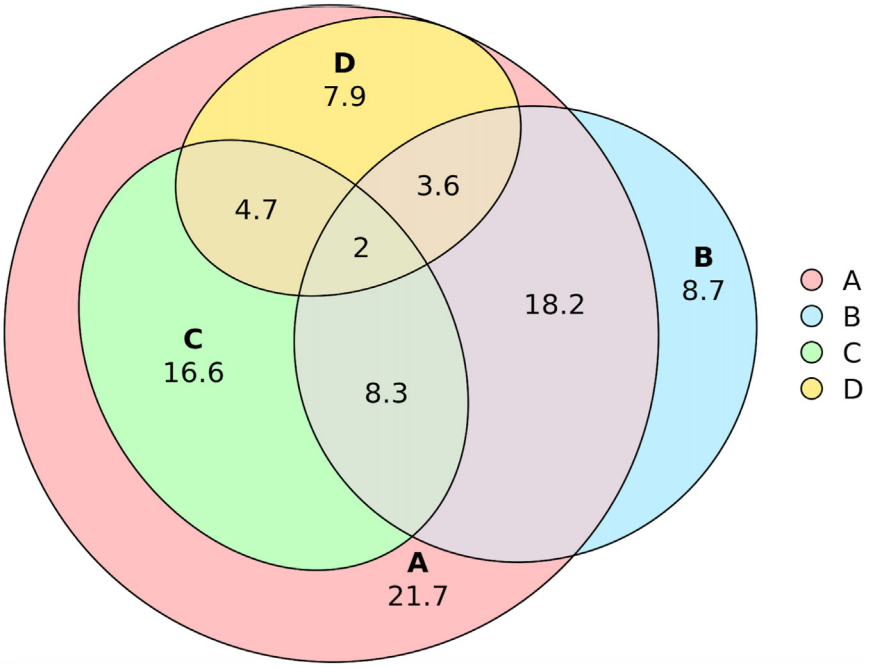

Figure 1. The isolated or combined contribution of the categories for estimated visual disability coefficients. A, Low vision. B, Visual field changes. C, Loss of bi-foveolar fixation. D, Photophobia. Numbers represent percentages.

supporting the first argument, only 21.2% (7/33) of patients < 14 years of age had visual field tests against 54.5% (103/189) and 51.6% (16/31) in the subgroups of patients between 15 and 64 and patients < 65 years of age, respectively.

The visual disability coefficient varied as per diagnostic groups (in the overall test comparing the 6 groups); however, these differences were only significant between only 2 groups (patients with "isolated progressive inherited retinal disorders" vs patients with "other rare disorders of the posterior segment of the eye"), meaning that overall IRDs were very debilitating (Fig 3). Compared among the 3 most frequent nonsyndromic diagnoses, which had a reasonable number for analysis, the visual disability coefficients were similar, although with differences in category contributions (Fig 4). Visual field changes, such as the constriction of the visual field, are easy to document in perimetry and are highly valued in the TNI, scoring higher in patients with nonsyndromic retinitis pigmentosa. On the other hand, visual field changes in the cone and cone-rod dystrophy and Stargardt disease are typically scotomas that, despite being central and

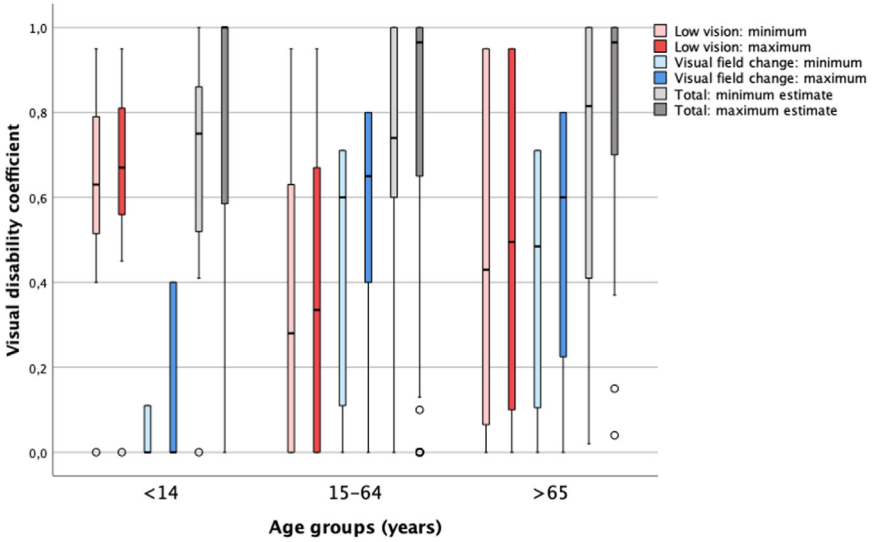

Figure 2. Graphical representation of the visual disability coefficient by age group and the low vision and visual field change contributions to the final value.

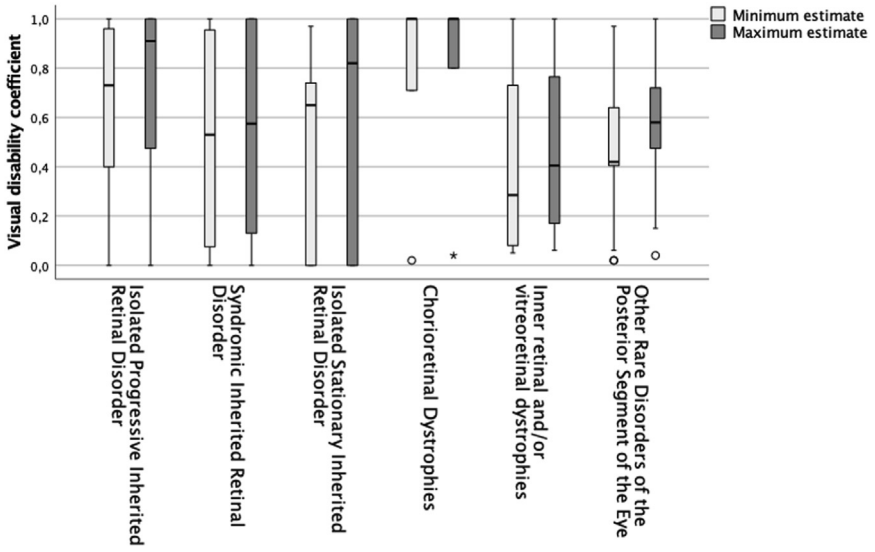

Figure 3. Graphical representation of the visual disability coefficient by diagnosis groups.

debilitating, are not highly valued in the TNI, and their contribution to the final coefficient is scoreless. In retinitis pigmentosa, low vision is of lesser importance since macular atrophy tends to occur late in the disease, allowing patients to maintain good visual acuity despite a tubular field. In contrast, in cone and cone-rod dystrophy and Stargardt, the loss of central visual acuity is characteristic from an early age, being a very important contributor to disability.

In this study, approximately a third of patients had low vision aids and they tend to be younger with lower visual disability coefficients. This demonstrates that rehabilitation is being prioritized among younger individuals, who are typically more adaptable. The inclusion of older patients should not be overlooked, and the earlier inclusion of patients with lower severity of the disease could also prove beneficial for achieving better results in the future.

This study is pioneering as it is the first to document visual impairment using a national table of disabilities for this group of pathologies.

One of the strengths of this study is its legislative framework, which enables us to apply the visual disability impact on the labour market and understand the intervention measures to support these patients. Another strength is the inclusion of a wide range of diagnoses included allowing us to form a complete and comprehensive portrait of the visual disability in IRD patients. Finally, most patients had working-age reinforcing the impact of these diseases on the labour market.

However, this study has some limitations. Only patients followed up at the hospital were included, which may contribute to a sample bias, overestimating the severity of the disease. Asymptomatic patients or those with mild symptoms may not be referred to a tertiary hospital or may

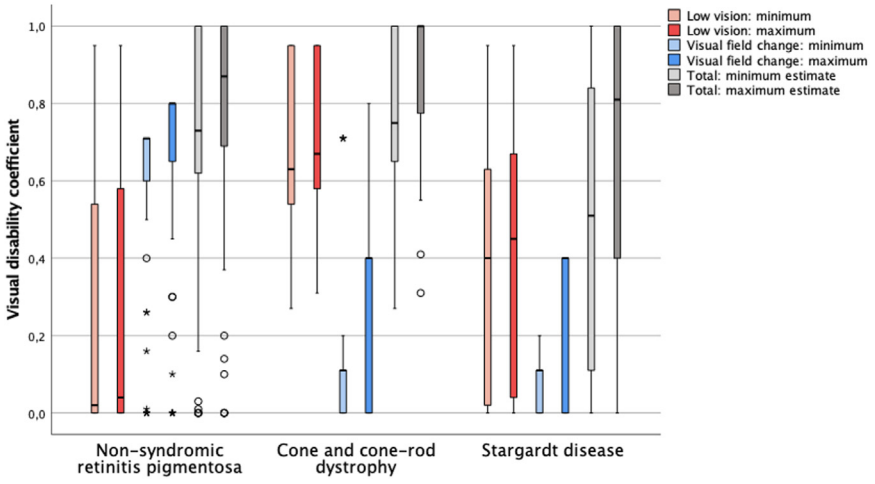

Figure 4. Graphical representation of the visual disability coefficient by the 3 most frequent nonsyndromic diagnoses and the low vision and visual field change contributions to the final value.

not have been diagnosed. Another limitation is missing data in the "visual field change" category, which may underestimate the estimated visual disability coefficient (Supplemental Document 2). The visual field test was not routinely performed in all IRDs. Many patients did not have the capacity to perform it because of syndromic characteristics (such as anatomic limitations, development delay, or intellectual development disorder), eye problems (unstable fixation as nystagmus, very low vision as light perception, or hand motion), age (child or elderly), and so on. Furthermore, the failure to include other types of visual impairments in the TNI (such as dyschromatopsia, nyctalopia, or loss of contrast vision) underestimates the real visual capacity of these patients. However, we chose to keep only the 7 ophthalmology categories included in the TNI to maintain a realistic social framework. Disabilities other than ophthalmological were also not evaluated, which may result in an underestimation of the overall disability, especially in syndromic cases. Nevertheless, the majority of the sample consisted of relatively young patients without concurrent pathologies, reducing the significance of this aspect. Finally, the results of statistical tests should be interpreted with caution, given the number of tests performed in this hypothesis-generating study.

In the future, we hope for the widespread availability of low vision aids for all patients, including older individuals. We also anticipate increased awareness and recognition of these diseases, leading to reduced discrimination within the community, including medical boards. Furthermore, we aspire for improved access to resources and opportunities in the labour market that are specifically tailored to accommodate these patients.

This study can be an example to be replicated in other countries and adapted to their own disability tables. Such an approach would enable us to gain a deeper understanding of this specific population, their visual disabilities and the socioeconomic implications involved.

In conclusion, this study confirmed that patients with IRDs experience significant visual disabilities, with the majority being eligible for a "multipurpose disability medical certificate." Interestingly, no correlations were observed between visual disability coefficients and age, suggesting that the disability is already considerable when these diseases manifest early in life. The disparities in visual disability coefficients observed only between the 2 diagnosis groups further indicate that IRDs as a whole have a profound impact on visual function and overall debilitation.

Footnotes and Disclosures

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No animal subjects were used in this study.

Author Contributions:

Conception and design: Marta

Analysis and interpretation: Marta, Miranda, Lume, Parreira, Soares, Menéres, Lemos, Beirão

Data collection: Marta, Miranda, Lume, Parreira, Soares

Obtained funding: N/A

Overall responsibility: Marta, Beirão

Abbreviations and Acronyms:
IRD = Inherited Retinal Diseases; **TNI** = National Table of Disability due to Work Accidents and Occupational Diseases.

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