de Renseignements sur le médicament, CHU de Saint-Etienne, Saint-Etienne, Auvergne-Rhône-Alpes, France; [8]*Service Hospitalo-Universitaire de Pharmaco-Toxicologie, Hospices Civils de Lyon, Lyon, Auvergne-Rhône-Alpes, France*

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HUMAN SUBJECTS: Human subjects were included in this study. All data are available in the FPVD are reported anonymously by each French regional pharmacovilance centre. The FPVD has an authorization from the "Commission Nationale de l'Informatique et des Libertés." According to current French legislation (loi Jardé, n2012-300), ethics committee approval and patient consent are not required for retrospective database studies with previously collected data (RIPH-4). This study adhered to the tenets of the Declaration of Helsinki.

No animal subjects were used in this study.

Author Contributions:
Conception and design: Thibault, Devilliers, Grandvuillemin
Data collection: Thibault, Rajillah, Freppel, Atzenhoffer, Boulay, Brunel
Analysis and interpretation: Thibault, Ben Ghezala, Rajillah, Auvens, Devilliers, Grandvuillemin
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Overall responsibility: Thibault, Grandvuillemin

Correspondence:
Thomas Thibault, MD, Service de Médecine Interne et Maladies systémiques (Médecine interne 2), CHU Dijon-Bourgogne, 14 rue Paul Gaffarel, BP 77908, 21079 Dijon Cedex, France.

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Diagnostic Odyssey of More than 1000 Patients with Inherited Retinal Diseases

Inherited retinal diseases (IRDs) encompass a diverse group of disorders characterized by varying visual impairment. Their diagnostic process is often referred to as an "odyssey" because of multiple challenges: (1) referrals to various specialists, (2) multimodal evaluations, and (3) difficulties related to the healthcare system, among others.[1] In this study, we describe the results of 2 surveys exploring this odyssey and satisfaction of more than 1000 patients.

This study analyzes the results of 2 anonymous and voluntary online surveys answered by patients or family members of patients with IRD, distributed by not-for-profit organizations, designed by their advisory teams. One was disseminated during 2023 in Latin America by the Argentinian Foundation of Retinitis Pigmentosa and the other in 2022 in the United Kingdom by Retina UK. Ethical approval was provided by the local ethics committees, and the study honored the tenets of the Declaration of Helsinki. Given that patients completed an anonymous online survey, informed consent was not applicable and was not obtained from the participants.

A total of 1096 participants completed the survey: 425 from Latin America (39%) and 671 from the United Kingdom (61%, Table S1, available at www.aaojournal.org).

In the Latin American Survey, most participants were from Argentina (397, 93%); 50 (12%) were younger than 18 years of age, and 375 were adults (88%, Fig 1A). A total of 310 had nonsyndromic retinitis pigmentosa (73%, Fig 1B). The survey was completed within 5 years since diagnosis by 112 participants (26%), between 5 and 20 years by 138 participants (32%), and after more than 20 years by 151 participants (36%).

The mean time between symptoms onset and diagnosis was 6.4 ± 9.1 years. A total of 243 patients (57%) waited more than 1 year, and 156 patients (37%) waited less than 1 year. Seventy-four children waited less than 1 year (29%) versus 82 adults (47%). Children had 2.1 times the odds of waiting more than 1 year for a diagnosis compared with adults.

A total of 120 patients (28%) had 1 or 2 visits until the diagnosis was made, 110 patients (26%) had 3 or 4 visits, and 195 patients (46%) had more than 5 visits. A diagnostic delay of less than 1 year was observed in 54% of patients with 1 or 2 visits until diagnosis, in 42% of those with 3 or 4 visits, and in 28% of those with more than 5 visits. Patients who had more than 5 visits to reach a diagnosis had 3 times the odds of waiting more than 1 year for a diagnosis compared with those with 1 or 2 visits and 1.9 times the odds than those with 3 or 4 visits. Those with 3 or 4 visits had 1.5 times the chance of waiting more than 1 year compared with those with 1 or 2 visits.

A total of 327 participants (77%) had 1 or 2 different diagnoses until receiving the correct one, and 98 participants (23%) had more than 3 diagnoses. A diagnostic period of less than 1 year was seen in 45% of patients with 1 or 2 previous diagnoses and 16% of those with 3 or more diagnoses. Patients with 3 or more diagnoses had 4.2 times the odds of waiting more than 1 year for a correct diagnosis compared with those with 1 or 2 diagnoses.

A total of 331 patients (78%) reported having their diagnosis clearly explained to them, 325 (76%) were told about the

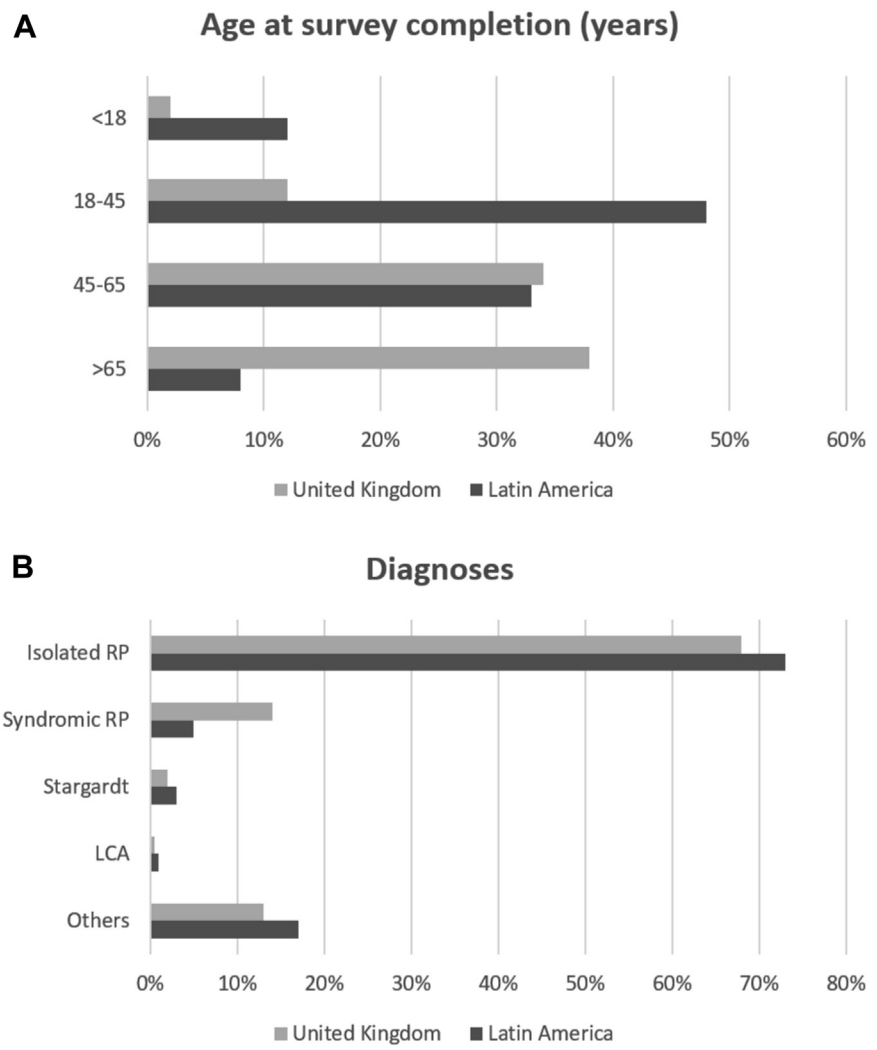

Figure 1. A, Age of patients at the time of survey completion. The Latin American cohort had a larger proportion of younger patients than the cohort from the United Kingdom (60% aged < 45 years vs. 14%). **B,** Diagnoses of patients who completed the survey. In both the United Kingdom and Latin America, approximately 70% of patients had retinitis pigmentosa, which is consistent with this being the most common diagnosis in ophthalmic genetics. LCA = Leber congenital amaurosis; RP = retinitis pigmentosa.

implications of the diagnosis to other family members, and 176 (41%) received recommendations about useful resources online and patient organizations.

In the UK Survey**,** 14 patients (2%) were younger than 18 years of age, and 562 were adults (84%, Fig 1A). Most patients had isolated retinitis pigmentosa (455 [68%]) (Fig 1B). Forty-seven participants (7%) received their diagnosis less than 5 years before completing the survey, 162 participants (24%) received their diagnosis between 5 and 20 years, and 439 participants (65%) received their diagnosis more than 20 years ago.

Regarding their diagnosis experience, 384 patients (57%) mentioned having the opportunity to ask questions, 285 patients (42%) thought that the person diagnosing them had enough knowledge about the condition, 306 patients (46%) considered that the doctor making the diagnosis showed empathy, 230 patients (34%) were informed about available support, and only 67 patients (10%) were offered psychological support.

In this report, we present for the first time the experience of patients with IRD while reaching their diagnosis and getting support.

Rare diseases affecting the eyes and nervous system have been found to have a greater diagnostic delay than those affecting other body systems.[2] In Latin America, 57% of patients described a delay of more than 1 year from symptom onset. This is in keeping with rare eye diseases in Spain (62.9%).[3]

Of note, a larger percentage of pediatric patients (71%) faced longer delays than adults (53%), which has not been the case for other systemic rare diseases.[4] This may be due to the sometimes less clear symptoms of IRD in children (e.g., mistaken as clumsiness or nyctophobia), its slow progression, less marked retinal features in children, and poor compliance with tests. Some of the determinants associated with longer diagnostic delay were young age, multiple visits, and different previous diagnoses.

Regarding the diagnosis experience of patients with IRD in the United Kingdom, approximately 50% did not remember having the opportunity to ask questions and thought that the person

diagnosing them did not have enough knowledge about the condition or showed empathy. In Latin America, more than 20% of patients did not have their diagnosis clearly explained and were not told about the implications of the diagnosis to other family members. In a large European survey, 35% of patients were not satisfied with how they received the diagnosis of a rare disease.[5] The announcement of a rare disease is a significant event in an individual and a family; thus, distinct consideration, empathy, and time are needed from healthcare professionals.[6]

The study limitations include its retrospective nature and the uneven populations, with the Latin American cohort having a larger proportion of younger patients compared with the United Kingdom cohort (60% aged < 45 years vs. 14%). Also, the fact that most UK patients had been diagnosed more than 20 years before completing the survey may introduce recall bias.

By reflecting on these testimonies, it is clear that there are areas of opportunity to improve the patient journey. Clear guidelines and training for ophthalmologists about how to manage patients with IRD are needed to provide more effective and efficient consultations. Improved accessibility to genetic testing and the development of networks of specialized doctors may optimize the diagnostic pathway for individuals with IRD,[7] regardless of their geographic location or socioeconomic background.

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Malena Daich Varela, MD[1,2]
Patricio Schlottmann, MD[3]
Jose Luna Pinto, MD[4]
Michel Michaelides, MD[1,2]

[1]*Moorfields Eye Hospital, London, United Kingdom;* [2]*UCL Institute of Ophthalmology, University College London, London, United Kingdom;* [3]*Charles Centro Oftalmológico, Buenos Aires, Argentina;* [4]*Centro Privado de Ojos Romagosa SA, Córdoba, Argentina*

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Author Contributions:
Conception and design: Daich Varela, Schlottmann, Luna Pinto
Data collection: Daich Varela
Analysis and interpretation: Daich Varela, Michaelides
Obtained funding: Michaelides
Overall responsibility: Daich Varela, Michaelides

Correspondence:
Michel Michaelides, MD, UCL Institute of Ophthalmology, 11-43 Bath Street, London, EC1V 9EL, UK.

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