European Society of Ophthalmology

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Project Abstract

Please include the headings if appropriate: ***Title, Purpose, Methods, Results and Conclusion***

Title of Project: Mapping the Portuguese IRD population: The IRD-PT3000 project

Purpose: Inherited retinal diseases (IRDs) represent a clinical and genetic heterogeneous group of rare eye disorders. The IRD-PT registry (www.retina.com.pt) is a national, web-based, pseudonymized, interoperable registry for IRDs, designed to generate scientific knowledge and collect high-quality data on the epidemiology, genomic landscape and natural history of IRDs in Portugal. Despite a consistent growth since its debut in 2020, the registry has a lower-than-expected adoption rate, with only 6 centers actively enrolling patients. Considering the Portuguese population (~10 million individuals) and a recent survey on the management of IRD patients in Portugal, we estimate an IRD prevalence of 1:3000 individuals. As of February 2024, the registry has 1696 patients enrolled. The aim of the IRD-PT3000 project is to map all Portuguese IRD patients in order to develop natural history studies, facilitate access to novel therapies and clinical trials.

Methods: We will develop strategies to increase the adoption and sustained use of the registry by all centers managing IRD patients in Portugal. Lack of time, individual attitudes and beliefs and low technological literacy were shown to be the most significant challenges and barriers to a nationwide embracement of the registry. Our approach for the future involves further dissemination in scientific events; making data capture easier and less time-consuming for the users with the development of additional training materials; and publication of multicenter studies with data from the registry, aiming to increase the attractivity of the platform.

Results: The registry has grown since its national debut in 2020 and as of February 2024, 1696 patients from 6 Portuguese public health care providers have been enrolled. Still, this number is far from the estimated total number of IRD patients in the country (~3300).

Conclusion: Rare disease registries increase research accessibility for patients, while providing clinicians/investigators with a coherent data ecosystem necessary to boost research and patient care. Since IRD incidence rates are low, national collaborations are the first step to get an overview of cohort sizes, genetic landscape, and the matching clinical features. Even though the registry has been available since 2020, its adoption has been far from ideal. Sustainability in the long run can only be met by fostering a culture of communication and cooperation between users and adopting realistic strategies to overcome challenges and barriers.