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ORIGINAL RESEARCH

The Landscape of Genomic Services for Inherited Retinal Degenerations (IRDs) Across Europe

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Purpose: To map the existing genomic services available for patients with IRDs across Europe.

Methods: A survey was conducted to 24 ophthalmic and/or genetic specialists across 19 European countries. The survey was conducted in an interview style via zoom for participants from 17 out of 19 countries. Interviewees were clinical/medical/ophthalmic geneticists, ophthalmologists/retina specialists and internal medicine specialists. The survey focused on referral pathways, genetic counseling, insurance coverage, awareness of genetic testing and counseling for IRDs among practitioners and patients, and preferred testing methodologies.

Results: Genomic services (testing and counselling) for IRDs vary among countries from an awareness, availability and insurance coverage perspective. Affordability could be a barrier for patients in countries without any payment scheme (eg, Poland) and in countries where only a targeted population is covered (eg, Bulgaria). Genetic counseling via qualified genetic counsellors did not exist in many countries. The level of awareness regarding the benefits of genetic testing in IRDs among healthcare professionals (HCPs) and patients was perceived as low in some countries. Panel-based next-generation sequencing (NGS) was the first test of choice for genetic testing in 68% of the studied countries.

Conclusion: There is some disparity in the approach to genetic testing for IRDs across Europe. Greater awareness of genetic testing services is required among the eye care professional community. A revised approach to the provision of genetic testing services such as centralized free genetic testing with associated interpretation and genetic counselling may help in ensuring equitable access and reimbursement, which will empower patients through improved access to clinical trials, expedite innovation, improve access to therapy and the delivery of care.

Keywords: genetic testing, genetic counselling, access, cost coverage, equitable, inherited retinal diseases

Introduction

Inherited retinal degenerations (IRDs) represent a myriad of vision-threatening conditions characterized by progressive or non-progressive degeneration of retinal photoreceptors and/or other retinal cells such bipolar cells, RPE and choroid. There are over 100 subtypes of IRDs caused by defects identified in over 300 genes.[1,2]

IRDs may be severe and early onset (eg, Leber congenital amaurosis), predominantly macular (eg, Stargardt or Best diseases), rod-cone (eg, retinitis pigmentosa), cone-rod (eg, Bardet–Biedl syndrome) or stationary (eg, achromatopsia, congenital stationary nightblindness).[3] IRDs are the leading cause of visual impairment and blindness among working-age populations in several developed countries.[4–6] The prevalence of IRDs in Europe and North America is approximately 1 in 3500 individuals.[7] Societal and economic impact of IRDs is a substantial burden to economies in the UK and Ireland, the total cost attributable to IRDs was estimated to be £42.6 million and £523.3 million, respectively, in the year 2019.[8] Similarly, in the USA and Canada, the total cost attributable to IRDs was estimated to be between US$13,414.0 and US$31,797.4 million and CAN$1637.8 and CAN$6687.5 million, respectively.[9]

IRDs present with a considerable genetic and phenotypic heterogeneity. Even within the same genetic subtype, the rate of progression, severity of visual impairment and age at onset of symptoms can vary. Defects in different genes can cause a similar clinical presentation, while defects in the same gene can cause a different clinical presentation. For example, PRPH2 variants can

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cause RP and macular dystrophy, while over 90 genes are associated with RP phenotypes.[3] Furthermore, the prevalence of genetic defects varies across different geographic regions.[10–12] Therefore, a proper genetic diagnosis via genomic testing is crucial to identify the exact gene responsible for the condition. Besides allowing the precise diagnosis of the condition, there are several additional benefits of genomic testing.[13] The outcome of genomic testing also allows clinicians to a) stratify clinical risk in terms of prognosis and co-morbidities, b) assemble the correct multidisciplinary team (for syndromic cases) for appropriate care and management plan and c) determine eligibility of the individual to be enrolled in emerging research studies and clinical trials. However, the benefits of genetic testing extend far beyond the clinical and therapeutic impact.[14] Having a precise molecular diagnosis through identifying the causative gene and the specific inheritance pattern of one's IRD empowers patients and their families to make informed life and family planning choices. via cascade testing. Genetic testing also allows other family members with less severe disease or carrier status to be identified via cascade testing.[15] These choices optimize the continued wellbeing of patients and their families, which can often be severely impacted by progressive vision loss.

The American Academy of Ophthalmology advises clinicians to perform a comprehensive medical, ocular and family history along with referral for genetic testing and counselling for all patients with inherited retinal diseases.[16] However, the integration of genetic testing protocol into routine eye care practice has been slower than expected. There is an understandable concern among eye care professionals that patients who undergo a genetic test for an IRD may ultimately be disappointed because the gene specific to their condition cannot be found, no research has been undertaken into their gene or there is no treatment available, causing inadequate referral of IRD cases for genetic diagnosis. Nevertheless, given the increasing number of pre-clinical and clinical studies on IRDs, therapeutic clinical trials and novel treatments on the horizon, it is now imperative for all patients with IRDs to receive a referral to a specialist in IRDs and receive genetic testing. Although it must be acknowledged that only approximately 60–80% of patients living with IRDs get confirmatory genetic diagnosis after genetic testing.[17] To effectively advocate for equitable, affordable, accessible, and timely genomic services for IRDs, it is first necessary to investigate the current genomic service landscape from a processes and systems perspective. Therefore, the overarching aim of this study is to explore the current landscape of existing genomic services for IRDs across Europe.

Methods

A survey was designed to assess the existing processes and system available for genomic services (genetic testing and counselling) for Inherited Retinal Diseases (IRDs). Survey questions and answers are provided in Supplementary Material File 1. The scope of IRDs discussed included the full list of IRDs outlined in the subject of the Retina thematic area of the European Reference Network for Rare Eye Diseases (ERN-EYE), a unique and innovative cross-border cooperation platform between specialists for the diagnosis and treatment of rare eye diseases within Europe.[18] The survey included questions in relation to the existing pathway to obtain genetic diagnosis, the state of genetic counseling, insurance coverage of genomic services, health care professionals' perceived awareness on the benefits of genetic services in IRDs among patients and practitioners and the most common testing methodologies for genomic testing. Surveys were conducted in an interview style via zoom video conferencing with questions provided in advance except for Belgium and Czech Republic where only written answers were obtained. The total duration of the interview lasted between 30 minutes to 1 hour. Purposive sampling was conducted to identify key informants in each country. The selection of key informants was determined by their active participation in the field of diagnosing and managing inherited retinal diseases. The expert advisory committee of Retina International, composed of three IRD specialists, provided recommendations for potential informants. Interviewees included clinical/medical/ophthalmic geneticists, ophthalmologists/retina specialists and internal medicine specialist from the following 19 countries across Europe: Bulgaria, Croatia, Czech Republic, France, Germany, Ireland, Italy, Lithuania, Netherlands, Norway, Poland, Portugal, Romania, Russia, Slovenia, Spain, Belgium, Sweden, and the UK. Eleven countries were full members of the ERN-EYE.[18]

Results

A total of 24 health care practitioners (12 clinical/medical/ophthalmic geneticists, 11 ophthalmologists/retina specialists and 1 internal medicine specialist) were identified and interviewed. More than one practitioner participated from some of the countries, hence the total number of practitioners was greater than the number of countries studied. Countries and associated key informants are presented in Table 1.

Table 1 Professionals Involved in the Study and Their Respective Cities and Countries

Country	City	Profession (n)
Bulgaria	Sofia	Geneticist (2)
Czech Republic	Prague	Ophthalmologist (1)
Croatia	Rijeka	Ophthalmologist (1), Geneticist (1)
France	Paris	Ophthalmologist (1)
Germany	Giessen	Ophthalmologist (1)
Ireland	Dublin	Ophthalmic Genetic Specialist (2)
Italy	Rome	Geneticist (1)
Lithuania	Vilnius	Clinical geneticist (1)
Netherlands	Nijmegen	Geneticist (1)
Norway	Oslo	Ophthalmologist (1)
Poland	Lublin	Ophthalmologist (1)
Portugal	Coimbra	Geneticist (1), Ophthalmologist (1)
Romania	Timisoara	Geneticist (1), Ophthalmologist (1)
Slovenia	Ljubljana	Clinical geneticist (1)
Spain	Madrid	Internal medicine specialist (1)
Sweden	Lund	Ophthalmologist (1)
UK	London	Ophthalmologist (1)
Belgium	Ghent	Ophthalmic genetic specialist (1)
Russia	Moscow	Ophthalmologist (1)

Current Practice of Genomic Services

The results of the survey demonstrated that the fundamental process to access genomic services for IRDs is consistent across all countries studied wherein patients are referred to a genomic testing center of excellence by one of the following health care professionals - an ophthalmologist, a pediatrician, or a general practitioner. The process of referral for genetic testing for syndromic and non-syndromic IRD cases was similar except a multidisciplinary approach is taken for the management of syndromic cases whereby a team of experts are consulted based on the organ systems affected. Common sources of referral relevant to syndromic IRD are ENT for hearing loss, looking for Usher syndrome, diabetes and kidney disease coming from nephrology looking for signs of Bardet Biedl Syndrome.[19] In relation to the availability of genetic counseling, respondents from 12 countries (67%) indicated that they do offer pre-test genetic counseling before genetic testing. However, the qualifications of the health care professionals offering genetic counseling varied slightly across countries. In the majority of countries genetic counselling was provided either by the ophthalmologist/retina specialist or clinical/medical geneticist having either a medical degree or genetics training. Only three countries (UK, Ireland and Belgium) specified that genetic counseling was offered by genetic counsellors. However, the respondent from Belgium stated that genetic counselors are not yet recognized as health care professionals in the country. Respondents from Spain raised a concern that medical genetics specialty is non-existent in the country; therefore, genetic counseling is offered by medical doctors who only have basic training in genetics.

Insurance Coverage of Genomic Services

In relation to the insurance coverage of genetic testing and counseling services, the majority of the countries surveyed had either full (67%) or partial coverage (22%) by their national health insurance or government/national pay scheme (Table 2). Partial coverage includes partial reimbursement of genomic services or full reimbursement to only a targeted patient population (eg, in Bulgaria only under 18s are covered, in Norway only under 16s are covered for genetic counseling). Poland was the only country among the countries included in this study where genetic services for IRDs were not covered by any of the payment schemes.

Perceived Awareness Among Health Care Practitioners and Patients

In relation to the participants' perceived level the awareness of genetic services for IRDs among ophthalmologists in their respective countries, respondents from 8 countries (42%) reported that the ophthalmologists are adequately aware, respondents from 5 countries (26%) reported that the ophthalmologists are aware but there is room for improvement and the respondents from the rest of the countries (32%) reported that they were either unsure or thought that there was inadequate awareness among ophthalmologists. Countries where the respondents reported they were either unsure or think that the ophthalmologists have inadequate awareness in relation to genetic services for IRDs include Bulgaria, Croatia, Czech Republic, Italy, Lithuania, Romania and Portugal.

Table 2 Insurance Coverage of Genomic Services for IRDs Across the Surveyed 19 European Countries

Country	Payment Coverage of Genetic Testing (GT) and Counselling (GC)
Belgium	National Health Insurance (small nominal fee applies)
Bulgaria	National Health Insurance (under 18s only)
Czech Republic	National Health Insurance
Croatia	National Health Insurance
France	National Health Insurance
Germany	National Health Insurance
Ireland	GT partly covered by the national or governmental payment scheme on request basis (case by case and occasionally by batch) GC covered by a mix of national health cover and charitable cover
Italy	National/ Government scheme
Lithuania	National Health Insurance
Netherlands	National Health Insurance
Norway	National/Government scheme (GT) Government scheme but only for under 16s (GC)
Poland	Out of Pocket
Portugal	National Health Insurance
Romania	National Health Insurance
Russia	Only a small proportion by National health insurance, charitable
Slovenia	National Health Insurance
Spain	National Health Insurance
Sweden	National Health Insurance
UK	National Health Insurance

In relation to the participants' opinion on the awareness of genetic services for IRDs among the patient population, participants from 11 countries (58%) thought that there is an adequate awareness, participants from 5 countries (26%) (Bulgaria, Czech Republic, Romania, Russia, and Slovenia) reported that there is low or very little awareness and participants from two countries (France and Portugal) said they were unsure about the level of awareness among the patient population.

Diagnostic Techniques for Genomic Testing for IRDs

According to the survey responses, respondents from 68% of the countries stated that panel-based NGS was their first test of choice for genomic testing for IRDs. Whole exome sequencing was the first test of choice in 26% of the countries.

Discussion

This study explored the current landscape of genomic services for Inherited Retinal Diseases in 19 European countries via a survey questionnaire administered to HCPs. Our findings revealed that the process to obtain genomic services by patients with IRDs varied slightly across the European countries. However, the pathway for referral of patients with IRDs for genomic services to the center for excellence and the management plan for syndromic and non-syndromic IRD cases was similar.

Most of the studied countries offered genetic counseling service, but it was offered either by clinicians (retina specialists) trained in genetics or medical geneticists. The profession of genetic[20] counseling was nonexistent in many countries. Genetic counseling is "the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease".[20] This process integrates: Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence. Education about inheritance, testing, management, prevention, resources, and research. Post- test counseling to promote informed choices and adaptation to the risk or condition.[20,21] In countries such as the USA, the profession of genetic counselling has existed for over 40 years.[22] In Europe, countries such as Denmark, France, Ireland, the Netherlands, Norway, Portugal, Romania, Spain, Sweden, Switzerland and the UK offer genetic counseling degree up to Masters level. In the rest of the world, the profession is continuing to develop, with countries in various stages of development.[23,24] Despite having educational courses that train genetic counsellors, genetic counseling by qualified genetic counselors for IRDs was not evident in our survey except for Ireland, the UK and Belgium. This could be due to the limited number of genetic counselors and inadequate funding allocated for these services. The limitation of available genetic counsellors is typically due to the large workload from cancer and cardiac genetic screening.[25] Worldwide, the number of trained genetic counsellors per million population varies widely with only a few countries having the recommended amount.[24] In Ireland and Canada, for example, there is approximately one genetic counsellor per 1 million, Denmark 4 per 1 million, Netherlands 3 per 1 million and Spain 1.5 per million (These numbers are clearly inadequate to fulfill the needs of the IRD community in these countries).

In relation to the cost coverage of genomic services for IRDs, over 85% of the studied countries had a full coverage of genomic services for IRDs either via national health insurance scheme or via a national/government payment scheme with additional research and charitable opportunities providing molecular diagnostics for free for the IRDs patients. This is a very encouraging finding. However, there are countries where the cost is partly covered (Russia, Ireland), only covered for a targeted group of people (Bulgaria, Norway) or not covered at all (Poland). For patients with IRDs in the aforementioned countries where no coverage is available or only partial coverage is available, there is a possibility of participating in a clinical trial to obtain genetic diagnosis and access emerging therapies; however, the number of patients who could benefit from this opportunity is limited.

Even in countries where the cost of genomic services is covered by a national health insurance or a government scheme, there is a general understanding that patients often experience delay in diagnosis leading to poor access to treatment.[26,27] This delay in diagnosis could be due to several reasons. One of the reasons could be due to health care practitioners' lack of knowledge regarding the benefits of genetic testing for IRDs, which leads to inadequate referrals of patients. This study suggested that a considerable proportion of health care professionals may not be aware of the benefits of genomic testing for IRDs. Similarly, the lack of awareness among the patient population regarding the

benefits of genetic testing may also contribute to the delay. This study revealed the perceived level of awareness regarding the benefit of genetic testing for IRDs among patient population can be improved in some countries with participants from Bulgaria, Czech Republic, Romania, Russia, and Slovenia stating that they think the level of awareness among patient population is low to very little. These findings, however, need to be confirmed via a large-scale survey targeting directly to the relevant health care practitioners and patients with IRDs although some previous reports have revealed that the knowledge of eye care professionals regarding the benefits of genomic services for patients with IRDs is inadequate.[28,29]

With the advancement of technologies, several diagnostics tests are available for genomic testing for IRDs. The type of the test that is ordered for genetic diagnosis depends upon various factors such as the patient's clinical diagnosis, the pattern of inheritance, the turnaround time required, and the patient's insurance coverage.[29] In this study next-generation sequencing was the first test of choice for genomic testing across the studied countries. This signifies that most of the studied countries have state-of-the-art genomic testing services, and those services are covered by the national health insurance programme.

Limitations

This study has some limitations. Firstly, we were able to approach key informants in 19 European states and therefore were only able to capture information from these states. The inclusion of additional European states would have provided us with a complete picture of the genomic services available to IRD patients in all Europe. Secondly, we determined the perceived level of awareness on the benefits of genomic testing for IRDs among HCPs and patients via an interview with a small number of HCPs. The results of this survey cannot be generalized to represent the level of awareness among the overall HCPs and patients in respective countries. A direct survey targeted at the HCPs and patients would need to be conducted to explore the actual level of awareness among the targeted population. Nevertheless, this study provided valuable insight regarding the existing system and processes of genomic services for IRDs in several European countries.

Conclusion

Some form of genomic services for IRDs that are either fully or partially covered by national health insurance or government payment scheme exist in several European countries. However, genetic counseling by qualified genetic counsellors is still unavailable in the majority of European countries surveyed. The level of awareness regarding the benefit of genomic testing for IRDs among health care practitioners and patients' needs to be improved. The emergence of novel therapies for IRDs necessitates all IRD patients to obtain genomic testing. There is a pressing need to integrate affordable genomic services into health care systems in states where genomic services do not exist or are not covered by any mechanism.[30]

Ethics Statement

This study did not require ethics approval as no personal, socioeconomic, or demographic information was collected from the interviewees.

Author Contributions

All authors made a significant contribution to the work reported, whether that is in the conception, study design, execution, acquisition of data, analysis and interpretation, or in all these areas; took part in drafting, revising or critically reviewing the article; gave final approval of the version to be published; have agreed on the journal to which the article has been submitted; and agree to be accountable for all aspects of the work.

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Disclosure

The authors report no conflicts of interest in this work.

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