

## Patient data: Recognition, evaluation, incorporation and practice

### Organizers

Avril Daly, CEO, Retina International and Orla Galvin, PhD, Director of Stakeholder Engagement, Retina International

### Description

Without real world evidence providing the true value of the impact of disease on patients and of the benefits therapy can provide to patients; access to diagnosis, interventions and clinical trials will remain a challenge for most across disease areas. This course will cover why and how patient data can be utilized to access diagnosis and provide a better understanding of complex and rare conditions. This course will cover why and how collaboratively all stakeholders in the patient journey can aid in the collection of patient data to be included in centralised registers. This course will cover how all of the above will enable the development of multi-centre Clinical Trials for small populations. This course will cover how to incorporate patient engagement in research from bench to bedside.

### Learning objectives

After attending this course, participants will be able to:

- Recognize, describe and discuss what patient data is
- Evaluate and interpret patient data
- Illustrate the importance of Patient Reported Outcome Measures
- Incorporate and practice patient engagement in their research

### Agenda

Presenters and presentations may change.

#### 1 – 1:15pm

#### Introduction

**Avril Daly, CEO, Retina International, Dublin, Republic of Ireland**

Provide overview of course, explain purpose and takeaway learnings as they relate to:

- Recognizing, describing and discussing what patient data is
- Evaluating and interpreting patient data
- Illustrating the importance of Patient Reported Outcome Measures
- Incorporating and practicing patient engagement in research

#### 1:15 – 1:45pm

#### Patient engagement in benchtop research

**Laura Brady, PhD, Head of Research, Fighting Blindness, Dublin, Republic of Ireland**

Patient and public involvement (PPI) can be viewed as a means of increasing the impact, quality and relevance of research studies. In what can often be a departure from conventional approaches, PPI describes research that is carried out 'with' or 'by' people living with sight loss rather than 'to', 'about' or 'for' them. This lived experience and perspective can be embedded at any stage of the research cycle, for example, by setting research priorities, refining project design, advising on study recruitment as well as contributing towards the dissemination of project outputs. This presentation will provide practical guidance on why and how people living with sight loss can have a role in benchtop research studies.

**1:45 – 2:15pm    How the patient can drive research for inherited retinal disease**

***Todd Durham, PhD, Vice President, Clinical and Outcomes Research, Fighting Blindness, Raleigh, NC and Silvia Cerolini, Founder, Through Vicky's Eyes, London, England, United Kingdom***

Industry sponsors and the FDA have identified many ways to incorporate patient perspectives into product development. What unique role can patients play in prioritizing a research agenda – before and during clinical development? And how can patients and their caregivers bring stakeholders together to address gaps in research?

We will address these questions by presenting the recent experiences of representatives of the *RDH12* Families who, for many years, have advocated and funded a research agenda. We will highlight the lessons learned from a recent scientific symposium we co-sponsored on clinical trial readiness for *RDH12* gene therapy. We will also share insights and recommendations on how researchers can best partner with patient groups.

**2:15 – 2:45pm    The important of gene-specific clinical phenotypes**

***Elise Heon, MD, FRCS, Ophthalmology & Vision Sciences, Hospital for Sick Children, Toronto, Canada***

Inherited retinal diseases (IRD) are a leading cause of blindness. Though these conditions are very genetically heterogenous, we have entered the era of gene specific treatments and over 40 gene specific clinical trials are ongoing. We have learned that the selection of outcome measures most representative of the gene specific changes optimizes the possibility to show evidence of efficacy. These outcome measures are best identified with knowledge of disease mechanisms and with natural history information on phenotype measures. Deep phenotyping allows to not only identify gene-specific outcome measure but also the identification of a therapeutic window. Deep phenotyping also allows selection of outcome measures optimal for specific age groups, e.g. children vs adult. These points will be discussed.

**2:45 – 3pm    Afternoon Break**

**3 – 3:30pm    How to engage patients about participation and use of patient data in clinical trial design and outcome**

***Michel Michaelides, BSc, MB BS, MD, FRCOphth, FACS, Professor of Ophthalmology, Moorfields Eye Hospital, London, England, United Kingdom***

The presentation will include ways to effectively and fully engage patients about participation in clinical trials (with focus on inherited retinal disease trials) and also discussion of the ways patient data are valuable in the design of clinical trials and outcome development. This will include discussion of the language used when engaging patients.

**3:30 – 4pm    Patient rare eye diseases data at the European level with ERN-EYE**

***Helene Dollfus, MD, PhD, Professor, Universite Louis Pasteur, Strasbourg, France***

European Reference Networks (ERN) have been created by the European commission to boost care of patients with rare diseases in the European Union. In conjunction with patient representatives and experts in the field of rare eye diseases ERN-EYE missions (29 EU hospitals in 13 member states) are to enhance the mutualization of expertise, collecting patient data with

semantic curation, boost research (especially for clinical trials) and guideline development. The Case Patient Management System (CPMS) enables sharing of patient data between expert centers especially for challenging cases. The Registry program aims to collect sound basic clinical data for each ERN-EYE patient referred to an ERN-EYE center. Patient representative are fully engaged in the process.

**4 – 4:30pm**

## **The impact of inherited retinal diseases in the Republic of Ireland and the United Kingdom**

***Orla Galvin, PhD, Director of Stakeholder Engagement, Retina International, Dublin, Republic of Ireland***

There is a lack of accurate data regarding the prevalence of the range of conditions which fall under the Inherited Retinal Diseases (IRD) classification, the impact on the individuals and families affected, and the cost burden to the Republic of Ireland (ROI) and the United Kingdom (UK) economies. This incomplete knowledge of burden and impact of IRDs hinders research and development, commissioning of clinical services, treatments, and the planning and implementation of clinical trials. Thus, there is a need for a stronger evidence base to support value for money to regulatory bodies for treatments recently approved, and treatments progressing through clinical trials towards market.

To ensure a strategic approach to future research and service provision it was necessary to learn more about the IRD landscape. The socioeconomic burden of IRDs in the ROI and UK was estimated using a cost of illness methodology applying a prevalence approach. The analysis was based on a targeted literature review and primary data collection. IRD Patient Representatives from each region provided feedback on the research brief. This feedback was incorporated to the design of the primary data collection tool (survey).

IRD Patients from the ROI and the UK with various levels of sight loss reviewed the surveys. This involved reviewing the content in terms of language, understanding and relevance; and reviewing the survey using different devices (PCs, laptops, phones), with different operating systems, screen readers, and magnification tools, and the time taken using each method. This case study highlights two recent cost-of-illness reports on the socio-economic impact of IRDs in the ROI and the UK, which demonstrate the true impact of IRDs on individuals affected, their families, friends and society as a whole.

Total costs attributable to IRDs in the ROI were estimated to be €49.5 million in 2019, comprising both economic (€33.5 million) and wellbeing costs (€16.0 million) which reflected 32.7% of total costs. In the UK, the wellbeing costs attributable to IRDs were £196.1 million, which represented 37.5% of the £523.3 million total costs in 2019, the remainder being economic costs (£327.2 million). Undoubtedly a greater emphasis needs to be placed on the wellbeing of those affected by IRDs, and this burden should be factored into reimbursement processes for therapies and care pathways.

**4:30 – 4:45pm** **Power of the patient**

***Christina Fasser, President, Retina International, Dublin, Republic of Ireland***

Patients are indispensable to drive research in inherited retinal degenerative diseases in many aspects: personal participation in genetic research, clinical trials, donating tissues and participating in long term natural history studies. It is not only the patient, but also his or her

family members that have to participate in all these activities. Furthermore, patients are best qualified to judge patient reported outcome measures. Inclusion of patients in the early stages of designing clinical trials is key for the successful enrolment. patients were and are still extremely important in creating public awareness, in education and last but not least in fundraising to support research.

**4:45 – 5pm**

**Discussion**