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Short REPORT

ERN-EYE

5th Scientific Workshop

NETWORK COORDINATOR

Professor Hélène Dollfus
Hôpitaux Universitaires de Strasbourg, France

E-mail: contact@ern-eye.eu

LEROUX Dorothée, Project Manager
VACCHI Isabella Anna, Project Manager
GAVARD Amélie, Medical Writer
WERNERT IBERG Caroline, Communication Officer

This document includes :
- *The short report, published on the ERN-EYE website*
- *The results of the satisfaction survey as annex 1*
- *The program of the event as annex 2*

European Reference Network
for rare or low prevalence complex diseases
Network Eye Diseases (ERN-EYE)

1st to 2nd December 2022
Ghent, Belgium

This report is a deliverable of the ERN-EYE project, funded by the 4th Health program, under the Specific Grant agreement n°101085439. The deliverable was created under the Work Package 8, Objective 8 – To give an impulse to ERN-EYE research collaborations by advertising calls for clinical and scientific research, organising workshops and promoting research committees and workgroups outside of this ERN-EYE network.

ERN-EYE WORKSHOP ON CLINICAL TRIALS

ERN-EYE Project Management Team

Introduction

ERN-EYE Workshop– December 1st- 2nd, 2022

Following the four previous scientific ERN-EYE workshops devoted to ontology, genetic testing, novel therapies, and modelling the eye, ERN-EYE has organized a fifth scientific workshop on "Eye Modelling" from 1st to 2nd of December in Ghent, Belgium. This meeting took place in a hybrid format and gathered nearly 90 people, remotely or on site. These included the full members of ERN-EYE and the affiliated partners of the network, ePAG members as well as prestigious invited international speakers. The workshop was opened by the organizing committee composed by Bart Leroy, host of this ERN-EYE workshop and chair of the Working Group 1 on Retina, Elfride de Baere, chair of the Transversal Working Group 6 on Genetic Diagnostics and Hélène Dollfus, ERN-EYE coordinator.

The morning of the first day was dedicated to internal meeting of ERN-EYE working groups.

In the afternoon, the plenary sessions of the workshop were dedicated to Lessons learned from clinical trials in Rare Eye Diseases. Speakers from 6 countries presented clinical trials on going or on hold due to various causes.

In the light of the overarching theme "Meaningful outcomes", ePAGs and patient representatives of the wider patient community presented a 360 degree patients' view and invited all stakeholders, including regulatory bodies, to embrace patient partnership in the development of research priorities and endpoints that are clinically significant AND meaningful for patients, in order to make clinical trials successful.

The second day was entirely dedicated to outcome measures: how to identify and improve them in Rare Eye Diseases. The word was also given to a member of the European Medicines Agency to talk about meaningful outcomes and specificities for eye-related studies.

Over the two days, more than twenty European and international experts presented and discussed the latest advances in clinical trials, their current challenges and their implications for the Rare Eye Diseases patients.

ERNs in brief

European Reference Networks (ERNs) are unique and innovative cross-border cooperation platforms between specialists for the diagnosis and treatment of rare or low prevalence complex diseases.

ERNs are virtual networks bringing together healthcare providers across Europe to tackle complex or rare medical conditions that require highly specialized treatment and a concentration of knowledge and resources. They are being set up under the EU Directive on Patients' Rights in Healthcare (2011/24/EU), which also makes it easier for patients to access information on healthcare and thus increase their treatment options.

The ERNs are supported by European cross-border telemedicine tools, and can benefit from a range of EU funding mechanisms such as the "Health Program", the "Connecting Europe Facility" and the EU research program "Horizon 2020".

DAY 1 -

Opening Session

Hélène Dollfus, ERN-EYE coordinator, Elfride de Baere, and Bart Leroy, hosts of this ERN-EYE workshop.

The workshop was opened by the organizing committee composed by Bart Leroy and Elfride de Baere, hosts of this ERN-EYE workshop and Hélène Dollfus, ERN-EYE coordinator. They welcomed all attendees, and introduced the topic of the meeting, highlighting the importance of rethinking the outcomes in Clinical Trials for REDs.

LESSONS FROM CLINICAL TRIALS IN RARE EYE DISEASES (REDS)

STAR (choroideremia) & Xirius (XLRP) Trials

Robert MacLaren, Professor of Ophthalmology, University of Oxford, Vitreoretinal Surgeon,Oxford University Hospitals NHS Foundation Trust

Pr Maclaren presented two clinical trials. The first one targeted Choroideremia, which, with a 1.9 kb gene, is fairly easy to integrate in a vector, whose components have been validated in other genes. The administration involves a subretinal injection. This gene therapy was tested in a phase three clinical trial, the STAR study. Despite encouraging clinical results, the product failed to reach its primary endpoint. It reached its secondary endpoints, but no approval. Despite being relatively safe, the promoter decided to end the study. The results have been published, and show the importance of choosing endpoints and question whether Best Corrected Visual Acuity (BCVA) is the best measure.

The second trial was the X-Linked retinitis Pigmentosa in the Xirius study, which showed promising results but had to stop due to COVID.

Lessons from Optogenetics: Generically Rebuilding Basic Sight

José Alain Sahel, Distinguished Professor and Chairman Department of Ophthalmology University of Pittsburgh School of Medicine Exceptional Class and Professor at Sorbonne Université, Paris

Optogenetics combines gene therapy with the insertion of a gene that produce rhodopsin, a photopigment sensitive to light, with an apparatus that converts light from outside into the precise wavelength necessary to reactivate remaining cone photoreceptors in the retina and start the transmission of an electric signal. There are currently phase I trials available, with a dose escalation process, however, it was impacted by Covid and so far only one patient has been treated. Nevertheless, results are promising, but again, the outcomes need to be dependent on what is meaningful for the patient.

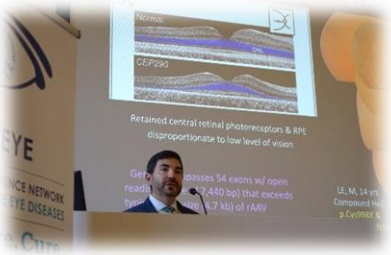
The BRILLANCE Phase ½ Trial: Lessons from CRISPR/Cas9 Technology

Mark E. Pennesi, MD/PhD, Paul H. Casey Ophthalmic Genetics Division

Leber Congenital Amaurosis is an autosomal recessive retinal disease that will cause an early and rapid vision loss caused by mutations in CEP290 gene. It manifests with an early loss of rod photoreceptors, nystagmus, photophobia, poor pupillary responses, and poor visual acuity, but the foveal cone-rich region remains intact until adulthood, providing an opportunity for gene editing. EDIT-101 is a vector designed to specifically edit CEP290 Gene. It uses a photoreceptor-tropic AAV5 vector and is administered subretinally with a single dose.

EDIT-101 was studied in the BRILLIANCE Study, which was a Phase 1/2, Open-label, Single Ascending Dose, to study safety and tolerability of a single sub-retinal dose of EDIT-101.

Endpoints were changes in BCVA, Full-field stimulus threshold, Visual function navigation and Vision-related quality of life.



Overall, EDIT-101 showed a favourable tolerance and safety profile. The best responders were homozygous patients, but the trials were stopped while waiting for an investor to continue.

AONs for CEP290-IRD (Illuminate): The Relativity of Outcomes

Bart Leroy, MD, PhD, Dept of Ghent University Hospital, and Aniz Girach, MD, Chief Medical Officer, ProQR Therapeutics,

Leber Congenital Amaurosis is an autosomal recessive retinal disease that will cause an early and rapid vision loss caused by mutations in CEP290 gene.

In the Phase 1/2 trial, sepofarsen, an antisens oligonucleotide, had demonstrated after 12 months of treatment that it was well tolerated and had a mean iBCVA improvement of more than 25-letters, with improvements in other clinical measures (FST and Mobility Course)

In the Insight extension trial, patients were followed up to 4 years and sepofarsen demonstrated continued good tolerability, sustained improvements in the first treated eye and a similar response in the second treated eye.

Sepofarsen was injected intravitreally and does not have a permanent action. Regular injections are required.

Illuminate was a double-masked, randomized, sham-controlled study, assessing efficacy and safety of sepofarsen in participants aged ≥ 8 years.

Participants were randomly assigned (1:1:1) to receive intravitreal injection of high or low dose or were part of the sham group. The primary endpoint was the mean change from baseline in BCVA, in the treatment eye (worse seeing eye), compared with sham at Month 12. Secondary endpoints included full-field stimulus testing threshold (FST; red, blue, white), a mobility course composite score, and safety.

Unfortunately, the trial failed to reach its primary endpoints despite the encouraging self-reported results in patients and a good safety profile.

This highlighted the need for trial design optimization, in order to reduce variability. Discussions with regulatory bodies will be needed to determine what would be the best control for these diseases.

Another suggestion was to better understand what the different regulatory agencies will or will not accept with respect to trial design and endpoints. Their difference in regulatory preferences on data analysis can be accommodated

through trial design, with for example one sham-controlled trial with two statistical analysis plans to satisfy both FDA and EMA requirements.

Regarding endpoint selection, was BCVA truly the best Primary Endpoint for LCA10, or were there any other alternatives that could be "approvable" in a Phase 3/Pivotal trial for registration?

FUNCTIONAL VISION: WHAT IS ENOUGH?- PATIENTS' & PARENTS' PERSPECTIVES

Meaningful Outcomes

Avril Daly, president of Eurordis

Avril Daly is the president of Eurordis and part of Retina International. In her view, there has been a tremendous improvement from the "go home, go blind" mentality that used to be prevalent.

People are starting to go back to their clinicians, with hopes to participate to clinical trials, but improvements can only come from keeping an open mind and listening to the challenges faced by patients.

Regulatory agencies are now open for discussion and are listening to the needs, though it should be noted that regulatory agencies are only the first step of the process, and that HTA will then have to be convinced

to reimburse those treatments. Studies on the economic burden of disease should be led.

A 360° view from ePAG representatives

Steven Van Cauwenberghe & Patients representative

Patients shared their lived experience through different lens. Dominique Sturz started on the parent experience. Christina Fasser, shared her experience of living with LCA, the slow decline of her vision, and the crucial points where she would have liked a treatment and how here expectations for treatment changed at those key points in life. Petia Stratiateva shared her life experiences, the challenges she is facing and her hopes for treatments. Michael Längsfeld shared his life stories in particular his decision to be included in a clinical trials and the reasons that pushed him to participate but also to refuse the treatment of the second eye.

All the testimonies will later be gathered in a booklet in order to do justice to the stories shared.

Meaningful Outcomes

Todd Durham, PhD, Sr. Vice President, Clinical & Outcomes Research

Dr Durham started by acknowledging that there were many perspectives to defining meaningful outcomes, and that what is meaningful for one may not be for another. Through a series of disease-specific workshops, results of pre-meeting surveys targeted to affected individuals and caregivers were collected and presented. Gathered with an informal and qualitative approach, the surveys highlighted patients and caregivers common Concerns and Impacts, such as progression of vision loss, loss of independence, worries about Safety....

He also presented two Patient questionnaires, developed specifically for patients with rare eye diseases. The first was the Michigan Retinal Degeneration Questionnaire (MRDQ), developed for adults with variety of IRDs and containing 59 items across 7 visual domains (central vision, colour vision, contrast sensitivity, scotopic vision, photopic peripheral vision, mesopic peripheral vision, photosensitivity).

The second one was the ViSIO-PRO (for adults) and an Obs-RO (for children), developed in collaboration with Novartis for individuals with variety of genetic causes of RP / LCA.

DAY 2 -

IDENTIFYING OUTCOMES MEASURES IN REDS- VISUAL FUNCTION VS FUNCTIONAL VISION

USH1C Vision and balance Natural History Studies and approach to sharing Clinical Data

Jennifer Lentz, Associate Professor at LSU

Dr Lentz presented the way she and her team have approached the challenges posed by the need for Natural History Studies in Rare Diseases combined to the small number of patients available. She described, for example, the process for their inclusion and the following study visits, organised over two days to minimize test fatigue. She also gave a demonstration of the functioning of the platform the team developed to share the patients' clinical data across all study sites in a secure and anonymised manner, and the way the data generated could be used for future Natural History studies.

ERN-EYE REDgistry as a database Model for REDs

Hélène Dollfus, ERN-EYE coordinator

Pr Hélène Dollfus, as coordinator of the ERN-EYE, presented the ERN-EYE REDgistry. Rare diseases are, by definition, rare and patient numbers are therefore often limited, which makes sharing data crucial. With REDgistry, ERN-EYE aims to develop a rare disease registry in order to reinforce their research capabilities and the knowledge about Rare Eye Diseases.

REDgistry is composed of two datasets, one common to all RD Registries in ERDRI, ensuring the interoperability of all ERDRI-based registries, the JRC Common Dataset, and a REDgistry Eye Dataset, specific to ophthalmology. REDgistry would be secured by pseudonymisation, GDPR compliant and interoperable, to allow data exchange in ERN-EYE, under *ad-hoc* governance rules. It was developed to follow the FAIR principles of being Findable, Accessible, Interoperable and Re-usable. The onboarding of the first centres is planned for 2023.

Pr Dollfus also presented the Together for Rare Diseases initiative, which is a Multi-stakeholder Alliance, aiming to unlock opportunities for ERN collaboration with the pharmaceutical industry. This initiative aims first, to accelerate research for rare diseases; second, to promote the EU to global industry leaders as a place for ground-breaking rare diseases research, and finally, to help ERNs fulfil their potential in the research field by developing transferable learning that can benefit all ERNs, no matter their

size, resourcing, current intensity of research etc.

This initiative has met several challenges. The first one is that, as it has been stated, industry could not provide funding to ERNs in any capacity. The second is the status of the ERNs, which are not legal entities.

Accelerating Research in REDs through an International Consortium

Allison Ayala, MS, presented on behalf of the FFB consortium investigator group

After asserting the vital need for natural history studies (NHS), Ms Ayala, as the director of the coordinating centre for the Foundation Fighting Blindness Consortium (FFB), presented their international Consortium of clinical centres to conduct IRD research, an initiative started in 2016. Their goal is to accelerate development of treatments for IRDs and is based on three principles, which are the collaboration of ideas, the collection of natural history study data and the sharing of data. To date, the Consortium has launched 4 natural history studies, in the USH2A, EYS, PCDH15, and OAT genes.

Uni-Rare has two components: The first component is a Registry of over 300 rare IRD genes that will collect prospective, standardized, cross-sectional clinical data - including visual acuity, OCT, static perimetry. The second component is the natural history study, which will serve as a platform to organise, gene by gene, a prospective, standardized, longitudinal data collection over four years from annual visits, including functional, structural and patient reported outcome measures.

The next question covered was how to use all the generated data to help design trials that industry can use, and that regulatory bodies will accept. The Regulatory Endpoints and Trial Design for IRDs (REDI) Working Group is currently trying to use NHS data to evaluate functional and structural endpoints, develop a formal endpoints proposal to present to regulatory bodies for input and publish their final recommendations.

A Multi-Luminance Mobility Test – Visual Function vs Functional vision

Daniel Chung, Chief Medical Officer at SparingVision

Visual Acuity is only one component of vision. However, the issue is that developing new outcomes is time consuming, costly and will need to correlate with visual field test.

The Multi-Luminance Mobility Test (MLMT) took two years to be developed. It included a bilateral approach, a high level of standardisation of patterns, which were prepared manually, according to daily situations and tasks encountered. The design also had to be adapted to children, adults and light levels. This is an expensive test, requiring two independent assessors and a dedicated room. However, in the validation studies MLMT showed a high correlation to visual tests. In conclusion, The Multi-Luminance Mobility Test should be made more available to the community.

Outcome Measures in Paediatric Trials

Elise Heon, MD, FRCSC, University of Toronto, Henry Brent Chair in Innovative Pediatric Ophthalmology Research at The Hospital for Sick Children

Pr Héon started by stating that, while MOST IRD are early onset, few kids are included in clinical trials. As an example, less that 10% of participants to RPE65 clinical trials are under 16 years old, and paediatric clinical trials represent 16% of all trials, despite 30% of world population being children. In the IRD field, 12% of CT are paediatric while this group carries 60% of the disease burden and are a leading cause of childhood blindness.

She advocated that outcome measures should be validated and designed for the paediatric age group and that the trial should be designed to include the paediatric age group. While children are not little adults, and will require adaptations, such as, for example, obtaining a child's agreement to

participate in research, which is an endeavour that should be carefully planned and implemented, they have the right to access essential medication.

Visual electrophysiology & objective tests as outcome measures

Katarina Stingl, University of Tübingen

Pr Katarina Stingl explained that retinal functional testing is important, as it tracks an improvement for the patient.

With the use of FST, Luxturna was approved while dark adapted perimetry can, for example, track the improvement on the treated area. Nevertheless, patient independent readouts are still a necessity, especially if results are inconclusive. These are important for understanding "what did not work".

She continued by emphasising the need for objective retinal functional evaluations of the numbers of rods and cones in terms of improvement or worsening in the treated area, and an exploration of whether photoreceptors connect in the retina. If there are no treatment effect, readouts on retina levels will allow to understand why. She concluded that proving that gene therapy is restoring retinal function has not been addressed properly so far.

Current & Future Outcome Measures

Rachel Huckfeldt, MD, PhD, Assistant Professor of Ophthalmology

The question is: how much are we assuming in gene therapies? Further information on the actual mechanism of action is needed. To that end, reporters that allow the identification of targeted cells, which could provide more proximal read-outs of biologic effects as well as more sophisticated immunologic markers, are needed. However, identifying reporters of genetic therapies is a shared challenge. The development of new intermediate outcomes will help us to understand the impact of genetic (and cellular) therapies in more depth.

MEANINGFUL OUTCOMES IN REDS – PERSPECTIVES OF REGULATORY BODIES

An EU regulator's perspective on clinical trials in inherited retinal diseases

Jane Moseley, Senior Scientific Officer, Scientific Advice office, EMA

Please note that the views presented are those of the individual and may not be understood or quoted as being made on behalf of EMA or reflecting the position of EMA, or Scientific committees or working parties.

EMA follows the general methodological guidance on clinical trials of the International Conference harmonisation (ICH). It has published a guidance for clinical trials, which includes specific guidance for early approval or incomplete data. Indeed, for some categories of medicinal products, in order to meet unmet medical needs of patients and in the interest of public health, it may be necessary to grant marketing authorisations on the basis of less complete data than is normally required.

This is applicable when it is intended for the treatment of seriously debilitating or life-threatening diseases, in which case a conditional marketing authorisation may be granted if several conditions are met, for example that the risk-benefit balance is favourable, or that unmet medical needs of patients will be met, etc.

Several suggestions were made, notably on the need for scientific advice on drug development program, and pivotal study design. Population inclusion could be staggered by age group.

Regarding endpoints, it was emphasised that, as the integrity of pivotal clinical trials is essential, so is the pre-specification, meaning that there could be no changes to the primary endpoint during trial, and that *post hoc* analyses are not suitable basis for approval. These should only be used as a hypothesis-generating tool.

However, while they prefer functional endpoints over structural, regulatory agencies are open to discussion regarding endpoints as long as the context is well defined. In conclusion, EMA is open to discussion and proposals for trial and endpoints with their procedures, and are interested in innovative approaches. Academia has a role in developing these into valid endpoints.

Increasing collaboration between outcome measure researchers, clinical trials teams, and regulatory bodies to optimise the outcome measures development

Jasleen K Jolly, Associate Professor, Vision and Eye Research Institute, Anglia Ruskin University

Currently the field is using tests that were designed for diagnosis, with old outcomes measure. Outcome measures are not recognised as a proper field of science.

Clinical trials fail regulatory approval due to poor endpoint choice. In the future, multidisciplinary teams deciding outcomes measures, earlier and better acknowledgment and funding of outcome research, in addition to better trial management and interaction between teams, could make a huge impact in Clinical trial successes.

How to improve Clinical Trials in REDs

Introducing Luxturna to the European Market

Quentin Spillaert, Novartis.

In most of the countries, the Health Authorities were open to dialogue and to make Voretigene Neparvovec accessible to patients, but the reimbursement processes took longer than anticipated. Payers appreciated being proposed a variety of flexible models, but the lack of epidemiological data and real-world data complexified the discussions.

The central onboarding process required unprecedented multidisciplinary collaboration between the centres of expertise and Novartis, and completing the whole onboarding process took on average 6 months.

Gaps were identified in Epidemiological data collections, genetic diagnosis advances and gathering of RWE. Novartis launched 19 research projects. The IRD field was keen to collaborate and has shown a strong investigator initiation. Collaborating with existing research centres proved to be most

effective, but still requires an extensive preliminary work, and those actions should have taken place years before the launch.

Round Table

Hélène Dollfus; Bart Leroy and Sue Lacey

Points of action

Working with patient organisations, which represent a challenge, should happen much earlier. The question is how to get patient perspectives and go beyond a simple advisory position:

(1) Patients should be engaged at the concept and research stage, to learn from them.

(2) Current rules of engagements are stopping engagement. There is a need for better, more efficient rules of engagements. Small focus groups on a specific topic may be a solution.

Even though pharmaceutical companies have patient-relation departments, any relation need to go through the legal department. Sometimes it requires up to 6 months of preparatory work to have meetings, and this creates delays and frustration. Moreover, the patient-relation department usually has no or too little knowledge of CTs, so there is a loss of information. It is important for patients to have access to the scientific and medical departments of pharmaceutical companies.

A solution might be Community Advisory Boards, where the agenda is created by patients. The meeting would include one patient organisation and several companies. Retina International for example could be such a platform.

Annex 1

Satisfaction Survey- Results

ERN-EYE organized a fifth workshop on “Clinical Trials in Rare Eye Diseases - Meaningful Outcomes” from the 1st to 2nd December 2022 in Ghent, Belgium. This meeting took place in hybrid form and gathered nearly 150 people, remotely or on site, from the HCPs representing 17 member states of the network, ePAG members and patients associations as well as prestigious invited speakers.

33 participants answered the survey, almost a fourth of the attendees.

The morning of the first day was dedicated to internal meeting of ERN-EYE working groups. In addition to contributing to the workgroup and transversal workgroup sessions, the ePAGs developed thir goals and a 2 years action plan in two ePAGs working meetings, having a focus on translating achievements and learnings of the very well established IRD space into a patient/clinician partnership in all RED areas in all EU countries. In the afternoon, the plenary sessions of the workshop were dedicated to lessons learned from clinical trials in Rare Eye Diseases. Speakers from 6 countries presented clinical trials, both ongoing or on hold due to various causes.

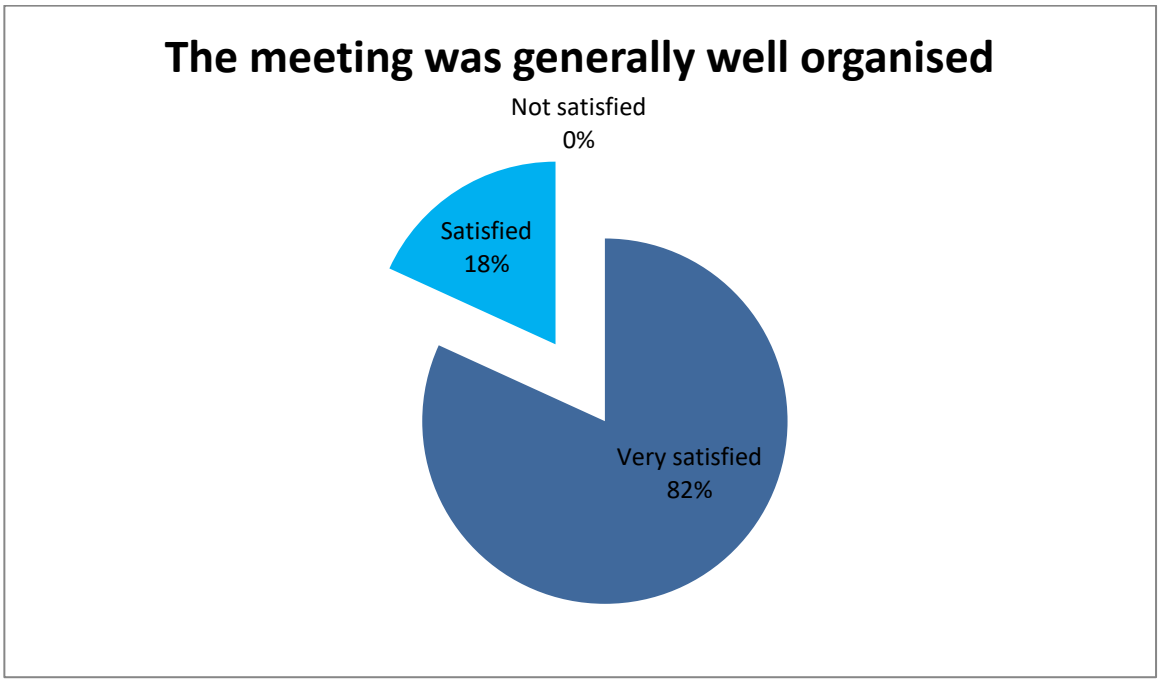
The second day was entirely dedicated to outcome measures: how to identify and improve them in Rare Eye Diseases. The word was also given to a member of the European Medicines Agency about meaningful outcomes and specificities for eye-related studies.

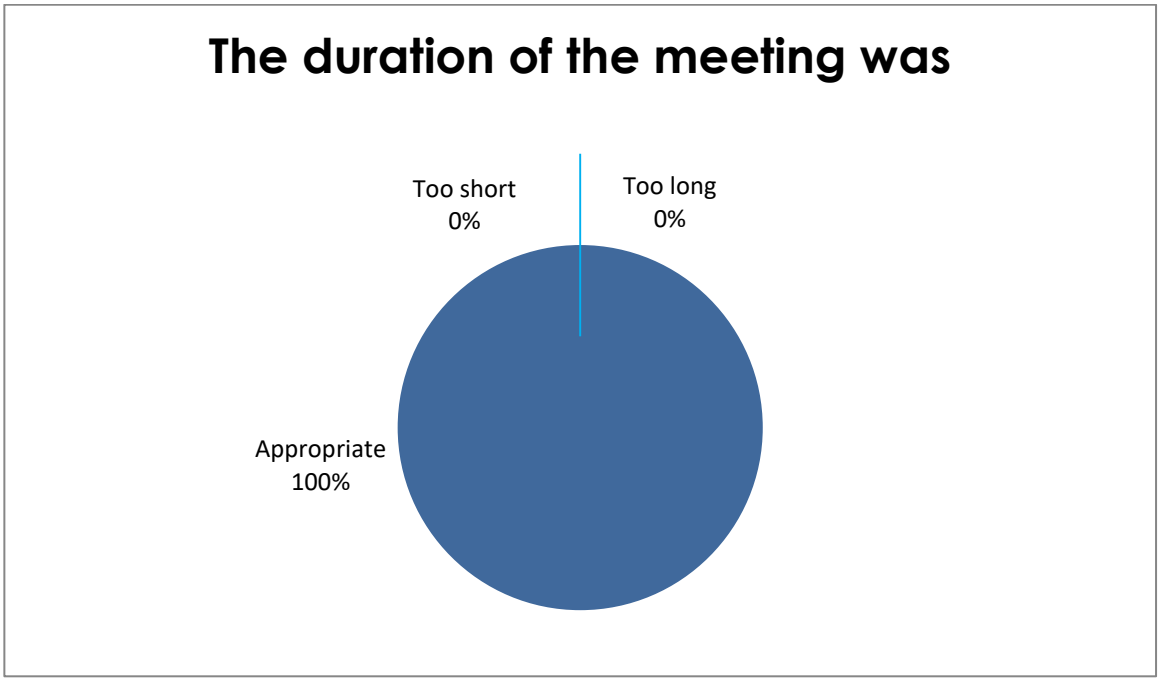
Over the two days, more than twenty European and international experts presented and discussed the latest advances in clinical trials, their current challenges and their implications for the Rare Eye Diseases patients.

When considering the answers of the satisfaction survey, it seems that, overall, this meeting met the expectations of the participants.

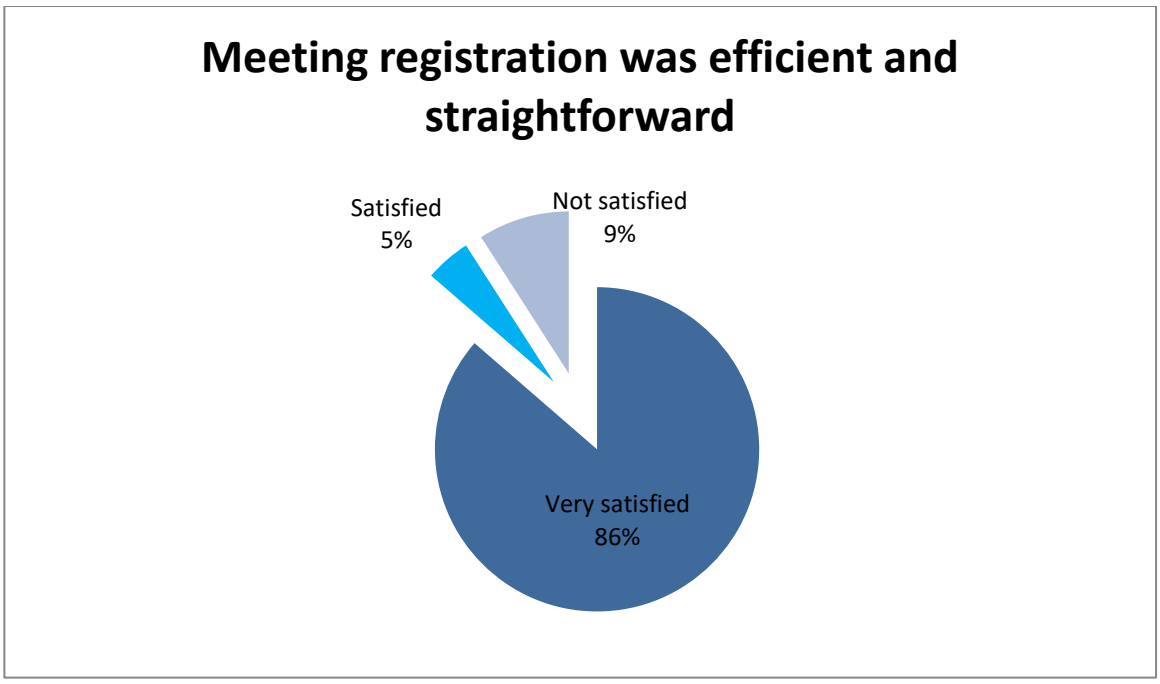
The only weakness in the organisation of this meeting was perhaps that the documentation (programme) was sent out too late. Otherwise, the organization in general suited the participants.

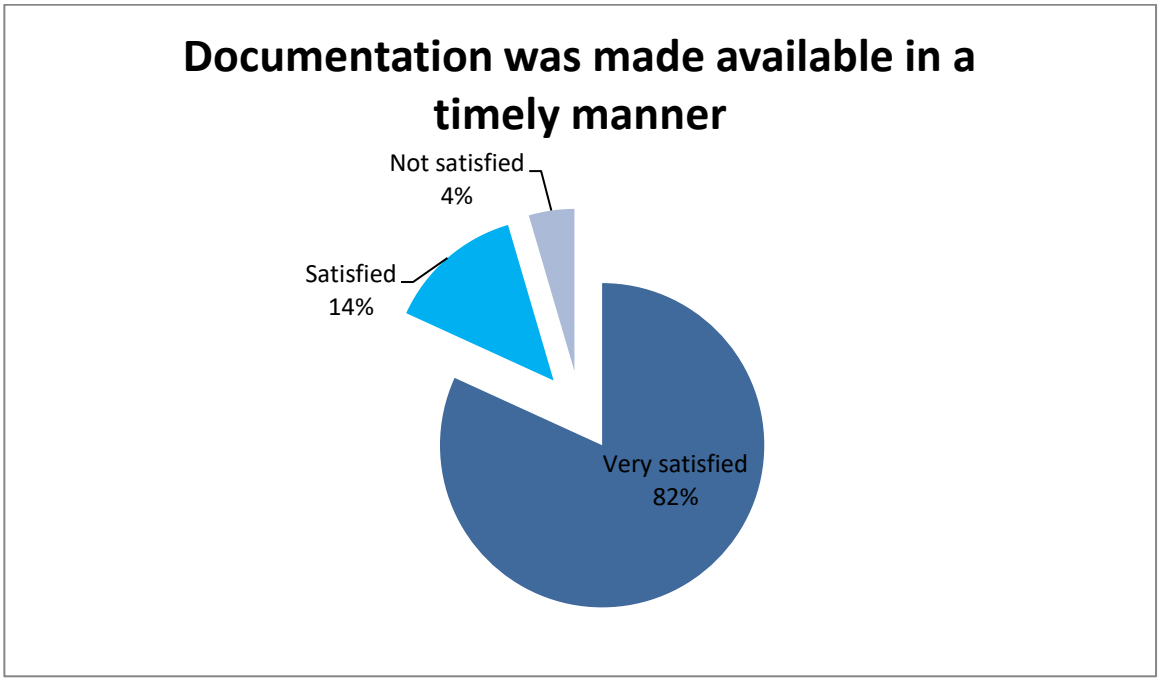
The results are presented below, question by question, with the percentages of responses.



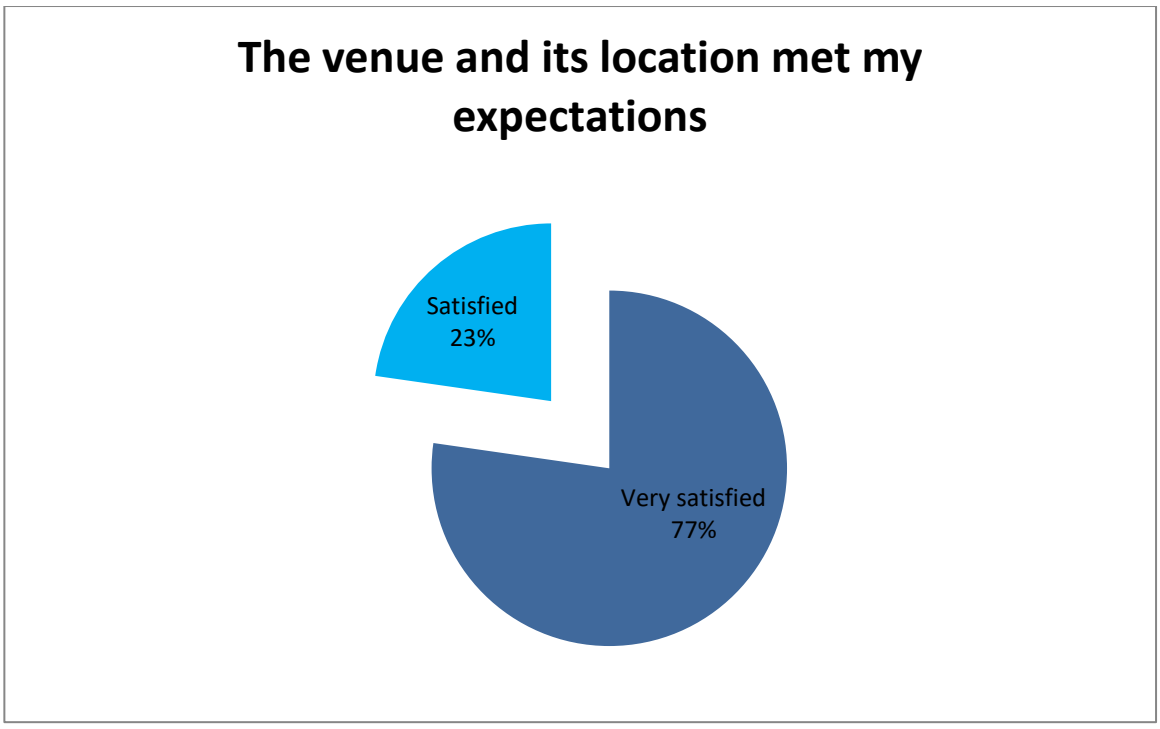


We noted that the participants were generally satisfied with the progress and organization of the meeting. This time, the duration of two days seemed to be perfect.

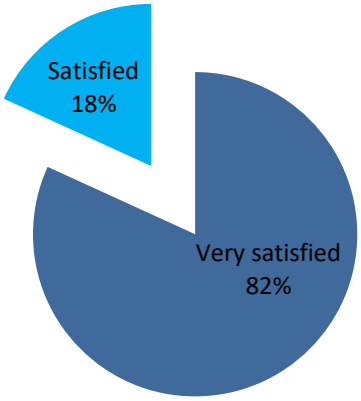




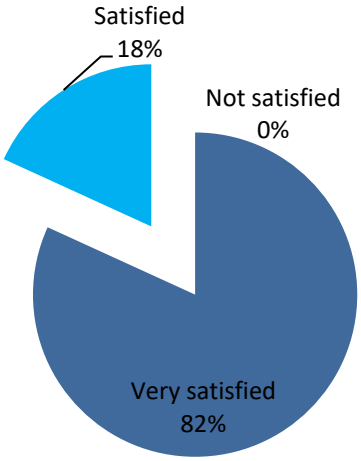
The registration was overall efficient. 4% out of the participants to the survey would have preferred to receive the documentation earlier.

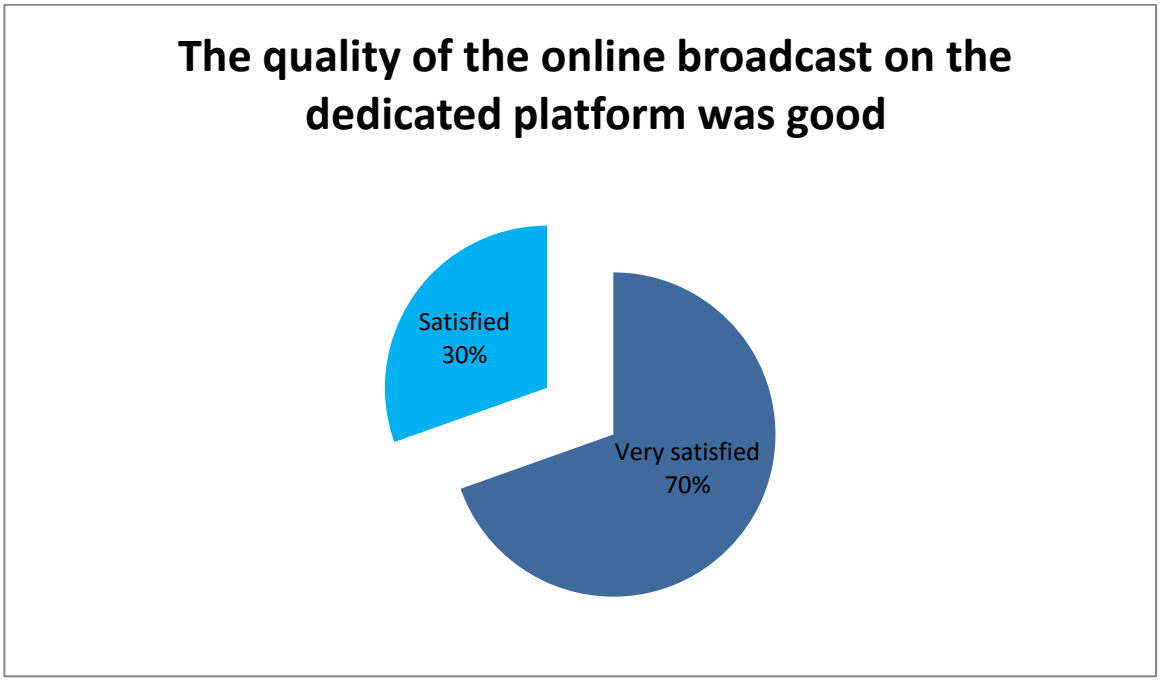


The quality of the sound in the meeting rooms to hear speaker on site was good



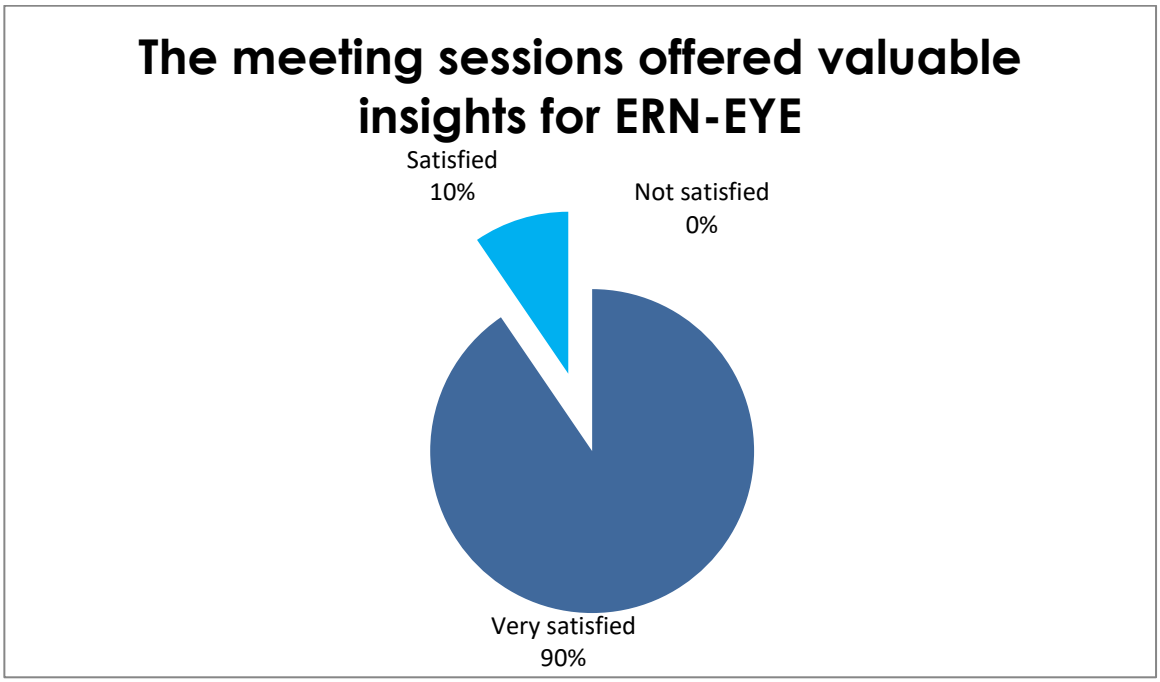
The quality of the sound in the meeting rooms to hear speakers online was good

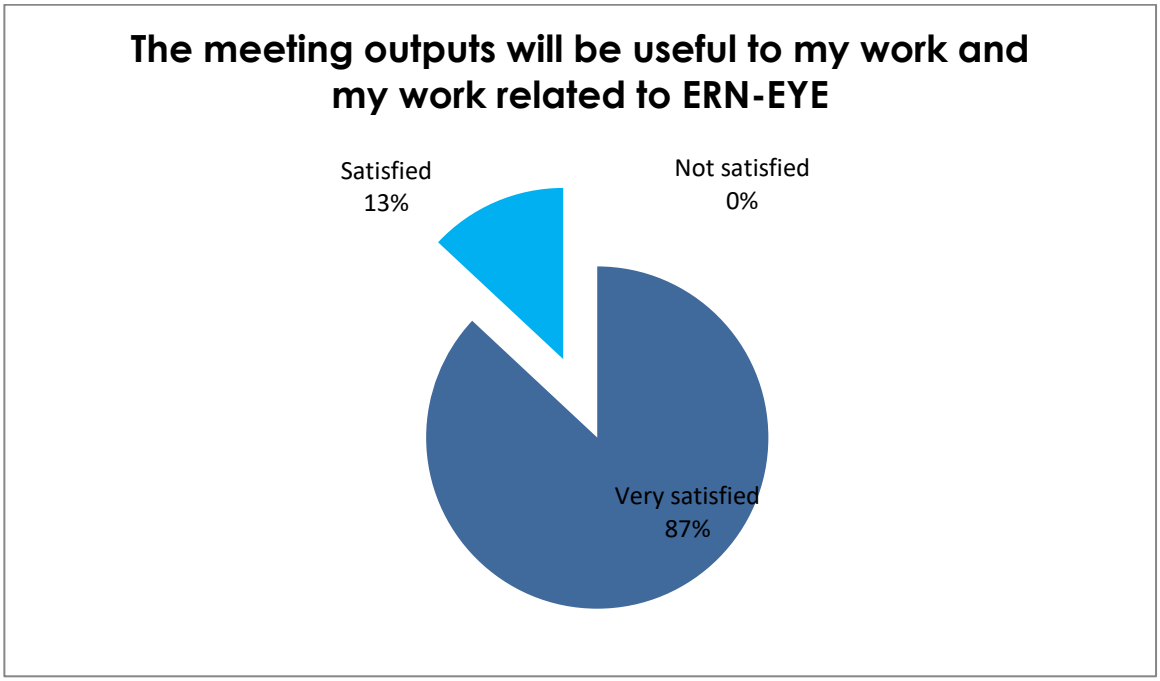




The event's format made it possible to satisfy almost all the participants.

The quality of the sound and internet was good, as well as the online platform to broadcast the meeting.





Thanks to this workshop, the issues, the needs and the actions needed in clinical trials, registries and natural history studies were discussed, as well as the role of ERN-EYE in the coming months on this subject. In view of the results mentioned above, it seems that the objective has been achieved.

Annex 2

5th SCIENTIFIC WORKSHOP

Clinical Trials in Rare Eye Diseases - Meaningful Outcomes

PROGRAM

December 1st - 2nd 2022

Het Pand, Onderbergen 1, 9000 Ghent, Belgium

THURSDAY - 1st OF DECEMBER 2022

11.00 - 13.00	*Individual ERN-EYE sessions of Working Groups (WGs)*
13.00 - 14.30	*LUNCH BREAK*
14.30 - 15.00	*Opening Session* by Organizing Committee - ***Hélène DOLLFUS, Elfride DE BAERE, Bart P LEROY***

LESSONS FROM CLINICAL TRIALS IN RARE EYE DISEASES (REDs)

15.00 - 15.20	STAR (choroideremia) & Xirius (XLRP) Trials - ***Robert MACLAREN***
15.20 - 15.40	Lessons from Optogenetics: Generically Rebuilding Basic Sight - ***José Alain SAHEL***
15.40 - 16.00	The BRILLIANCE Phase 1/2 Trial: Lessons from CRISPR/Cas9 Technology - ***Mark PENNESI***
16.00 - 16.20	AONs for CEP290-IRD (Illuminate): The Relativity of Outcomes - ***Bart P LEROY & Aniz GIRACH***
16.20 - 16.50	Q&A
16.50 - 17.00	Group picture
17.00 - 17.30	*COFFEE BREAK*

FUNCTIONAL VISION: WHAT IS ENOUGH? - PATIENTS' & PARENTS' PERSPECTIVES

17.30 - 17.45	Meaningful Outcomes - ***Avril DALY***
17.45 - 18.00	Meaningful Outcomes - ***Steven VAN CAUWENBERGHE***
18.00 - 18.20	A 360° view from ePAG representatives

- Lived experience - Parents' perspectives on meaningful outcomes and perspectives of children, teenagers, young adults with syndromic or non-syndromic IRDs/REDs - ***Dominique STURZ***
- Personal journey with LCA - Which meaningful outcomes would have changed my life and career? - ***Christina FASSER***
- Lived experience - Personal journey at work and meaningful outcomes - ***Petia STRATIEVA***
- Psycho-social aspects - Expectations from a clinical trial and decision making: What about the second eye? - ***Michael LÄNGSFELD***

18.20 - 18.35	Meaningful Outcomes - ***Todd DURHAM***
18.35 - 19.00	Q&A
19.30	*POTLUCK DINNER*

European Reference Network for rare or low prevalence complex diseases
Network Eye Diseases (ERN-EYE)

Funded by the European Union

FRIDAY - 2nd OF DECEMBER 2022

IDENTIFYING OUTCOME MEASURES in REDs - VISUAL FUNCTION vs FUNCTIONAL VISION

08.30 - 08.50	USH1C Vision and Balance Natural History Studies and Approach to Sharing Clinical Data - ***Jennifer LENTZ***
08.50 - 09.10	ERN-EYE REDgistry as a Database Model for REDs - ***Hélène DOLLFUS***
09.10 - 09.30	Accelerating Research in REDs through an International Consortium - ***Allison AYALA***
09.30 - 09.50	A Multi-Luminance Mobility Test - Visual Function vs Functional Vision - ***Daniel CHUNG***
09.50 - 10.20	Outcome Measures in Paediatric Trials - ***Élise HÉON***
10.20 - 10.40	Visual Electrophysiological Tests as Outcome Measures - ***Katarina STINGL***
10.40 - 11.00	Current & Future Outcome Measures - ***Rachel HUCKFELDT***
11.00 - 11.30	***COFFEE BREAK***

MEANINGFUL OUTCOMES in REDs - PERSPECTIVES OF REGULATORY BODIES

11.30 - 12.00	An EU regulator's perspective on clinical trials in Inherited retinal diseases - ***Jane MOSELEY***
12.00 - 12.30	Increasing collaboration between outcome measure researchers, clinical trials teams, and regulatory bodies to optimise the outcome measures development - ***Jasleen JOLLY***
12.30 - 13.00	Q&A
13.00 - 14.00	***LUNCH BREAK***

HOW TO IMPROVE CLINICAL TRIALS in REDs

14.00 - 14.20	Introducing Luxturna to the European Market, lessons learned - ***Quentin SPILLIAERT***
14.20 - 15.45	Round Table Discussion with All Stakeholders Mediated by ***Hélène DOLLFUS, Sue LACEY & Bart P LEROY***
15.45 - 16.30	Conclusions & End of Meeting