

## Network Coordinator

Professor H el ene Dollfus  
H opitaux Universitaires de  
Strasbourg, France  
E-mail: [ern.eyeproject@chru-  
strasbourg.fr](mailto:ern.eyeproject@chru-strasbourg.fr)



**European  
Reference  
Network**

for rare or low prevalence  
complex diseases



**Network**

Eye Diseases (ERN-EYE)

# ERN-EYE Ontology Workshop short report

9th to 11th October 2017,  
Mont Ste-Odile, France

LEROUX Doroth ee, Project Manager

Co-funded by  
the European Union



# ERN-EYE Ontology Workshop short report

ERN-EYE Project Management Team

13/10/2017

## Introduction

### *ODILE Ontology Workshop from October 9th – October 11th, 2017*

The first ERN-EYE workshop was held at Mont Ste-Odile in Alsace from 9 to 11 October 2017. It has been a full success with up to 60 participants from all over Europe and the United States. Opened by the coordinator H el ene Dollfus, this workshop was dedicated to the revision of the Orphanet

on four major clinical themes, including retina pediatrics, neuro-ophthalmology or anterior segment, the groups aim to unite the capacities of its members according to their specialties. This curation work is crucial to build all e-Health tools needed in such a network. The "Odile" workshop

*The Ontology Workshop was a great opportunity to present the activities of the network and for the members to advance on their respective projects.*

classification revision and HPO terms revision. ERN-EYE is the first ERN to do such an exercise at European level in one shot. This work was possible thanks a great cooperation of all key actors i.e.: Orphanet, HPO and RD-Connect.

The rest of the meeting was mainly devoted to working groups. Based

was ERN-EYE first achievement. As a matter of fact, to build common tools we had to agree on a common language. With this classification and a well-designed ontology, the building of an efficient IT platform and an EU-wide registry is now possible.



## *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 specialised 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 will be supported by European cross-border telemedicine tools, and can benefit from a range of EU funding mechanisms such as the "Health Programme", the "Connecting Europe Facility" and the EU research programme "Horizon 2020".

## Plenary Opening Session

### Welcome address – ERN-EYE news and purpose of the workshop

Hélène Dollfus, ERN-EYE coordinator



*Hélène Dollfus*

Opened by the coordinator Hélène Dollfus, the workshop was a great opportunity to present the activities of the network and for the members to advance on their respective projects. After thanking the members and experts who made themselves available for the occasion and got involved in this

work, she put the goals of this workshop into perspective in the context of the first year of the network's existence. She also presented the history of the highly symbolic place.

### ERN-EYE patient perspective

Russel Wheeler, ERN-EYE ePAG representative

The patient associations represented by Christina Fasser and Russel Wheeler opened the first part of the meeting with the patient perspective regarding ERN-EYE. Russel Wheeler spoke by emphasizing the original and essential character of patient integration into ERNs. In particular,

he illustrated his remarks with a very timely and humorous video. The short film is a parable that illustrates the benefits of co-production between professionals and patients in a close partnership as a different and efficient way to solve problems.



*Russel Wheeler*

### RdConnect : why and how ontologies in rare diseases ?

Rachel Thompson, RD-Connect communications and data platform manager, Newcastle, UK



*Rachel Thompson*

RD connect is a joint European action funded for 5 years whose goal is the development of a platform connecting databases, registers and biobanks in the field of rare diseases. For this purpose, RD-connect has developed many

skills and bioinformatics tools. Rachel Thompson presented the work done by RD-Connect on data-linkage, classifications and principles of FAIR data (Findable, Accessible, Interoperable, and Reusable).

## *ERN's and new communication tools –*

EC – Jean-Marie Misztela, Team Leader - IT project portfolio management ERN, eHDSI and EU-CEG, and Marta Terron-Cuadrado, eHDSI and semantic transcription - by teleconference

The European Commission, represented by Jean-Marie Misztela, and Marta Terron-Cuadrado, was invited to present the tools made available to the ERNs to facilitate their daily work:

the EC collaborative platform, the webex tool and the CPMS. This intervention could only be done in videoconference. Unfortunately a technical incident shortened the intervention of the commission. For

this reason and because tools are evolving very quickly, the commission will be invited to present to all our members during our next annual General meeting in Tallinn in March 2018.

## *ERN-EYE AND ORPHANET REVISION*

### *ORPHANET session opening: general presentation*

Emmanuel Maxime, ORPHANET, Paris, France

Orphanet is a resource that aims to gather and improve knowledge on rare diseases so as to improve the diagnosis, care and treatment of patients with rare diseases. Orphanet aims to provide high-quality information on rare diseases, and ensure equal access to knowledge for all stakeholders. Orphanet also maintains the Orphanet rare disease nomenclature (ORPHAcodes), essential in improving the visibility of rare diseases in health and research information systems. Orphanet was established in France by the

INSERM (French National Institute for Health and Medical Research) in 1997. This initiative became a European endeavour from 2000, supported by grants from the European Commission: Orphanet has gradually grown to a Consortium of 40 countries, within Europe and across the globe. Emmanuel Maxime presented the goals of this workshop, the preliminary work done before the meeting, what is expected on site and what will remain to be done after the meeting. The preliminary work conducted the month before by

both Orphanet and the ERN-EYE project management team led to the preparation of a reference document which was subsequently used as the basis for the work of each WG. In the same way, a webinar was organized before the summer in order to allow the WG leaders to conduct their work in a similar way from July to September.



*Emmanuel Maxime*

## Plenary: Short presentation from WG1 to WG4 (5-7 minutes each) of work on ORPHANET and general discussion

### WGs leaders and representatives

Each WG leader had the opportunity to present to all members of ERN-EYE and international experts invited for the occasion the work done by his group during the previous months in order to open the discussion regarding the modifications to be done to the current classification with regard to the literature and

the clinical knowledge and experience.



Bart Leroy

Bart Leroy presented the work of the WG dedicated to Retinal RED, Axel Petzold for the Neuro-ophthalmological RED, Panos Sergiounotis for Paediatric RED and Daniel Böhringer for anterior segment RED. They pointed out crucial points to discuss and thank the experts invited for their involvement.

Table 1 : Leaders of each ERN-EYE WGs and Transversal WG5

WG number	Theme	Chairs
WG1	Retinal Rare Eye Diseases	B.Leroy (UG) and M. Larsen (Rigshospitalet)
WG2	Neuro-ophthalmology Rare Diseases	A.Petzold and S. Wong (MEH)
WG3	Pediatric Ophthalmology Rare Diseases	B. Lorenz (UKGM) and J.Ashworth (MREH-MAHSC)
WG4	Anterior Segment Rare Eye Diseases	D. Böhringer (UMCF) and P. Liskova (HPH)
TWG5	Low Vision and Daily Life Patient Group	S. Mohand-Said (CHNO-XV-XX) and D. Keegan (MMUH – pending)

## Parallel: WG meetings to adjust ORPHANET revision

### WGs

All the four clinical working groups, animated by WG leaders and Invited experts, met to continue their work on the roadmaps allocated to them by the coordination of the project. They

discussed about the work on Orphanet classification of rare diseases they've already done during the summer period and discuss all important points pointed out during this step.



Table 2 : Invited experts in each WG

WG number	Theme	Invited experts
WG1	Retinal Rare Eye Diseases	<b>Eyal Banin</b> , Center for Retinal and Macular Degenerations, Hadassah-Hebrew University Medical Center, Israel <b>Kerry Goetz</b> , eyeGENE Coordinator, Health Science Program Manager, Bethesda, USA <b>Veronika Vaclavik</b> , Hôpital ophtalmique Jules-Gonin, Unité d'oculogénétique, Lausanne, Swiss
WG2	Neuro-ophthalmology Rare Diseases	<b>Aki Kawasaki</b> , Hôpital ophtalmique Jules-Gonin, Unité d'oculogénétique, Lausanne, Swiss <b>Alain Vighetto</b> , Hôpital Pierre Wertheimer, Hospices Civils de Lyon, Bron, France
WG3	Pediatric Ophthalmology Rare Diseases	<b>Elias Traboulsi</b> , Pediatric Ophthalmology & Strabismus, Center for Genetic Eye Diseases, Cleveland Clinic, Cleveland, USA <b>Elise Héon</b> , Eye Genetics Program, Hospital for Sick Children, Toronto, Canada <b>Alain Verloes</b> , Département de Génétique - Hôpital Universitaire Robert DEBRE, APHP, Paris, France
WG4	Anterior Segment Rare Eye Diseases	<b>Stefen Tuft</b> , Moorfield Eye Hospital, London, UK <b>Walter Lish</b> , Augenlinik und Polyclinik, Johannes Gutenberg Universität –Mainz, Germany
TWG5	Low Vision and Daily Life Patient Group	<b>Gislin Dagnelie</b> , JHU Lions Vision Research & Rehab Center, Johns Hopkins Hospital, Baltimore, USA

## *Plenary - Invited lecture: National Eye Institute and rare diseases databases and ontologies*

Kerry Goetz, NEI Common Data Element Coordinator, National Eye Institute, National Institute of Health, Bethesda, USA

The National Eye Institute (NEI) was established by Congress in 1968 to protect and prolong the vision of the American people. NEI research leads to sight-saving treatments, reduce visual impairment and blindness, and improve the quality of life for people of all ages. NEI-supported research has also advanced our knowledge of how the visual system—from the eyes to the brain—works in health and disease. NEI supports vision research through approximately 1600 research grants and training awards made to scientists at more

than 250 medical centers, universities, and other institutions across the country and around the world. NEI also conducts laboratory and patient-oriented research at its own facilities located on the NIH campus in Bethesda, Maryland. Kerry Goetz presented us what's up regarding databases and ontologies in the National Eye Institute where initiatives are currently running. They noticeably will set up a Special Interest Group on "Ontology and Common Data Elements for Collaborative Research in Ophthalmology" in

ARVO and organize a meeting regarding these matters and invite ERN-EYE to actively participate to this important event.



*Kerry Goetz*

## *ERN-EYE and HPO REVISION*

### *Plenary: HPO Session general presentation and ERN-EYE Ontology & HPO*

Peter Robinson, HPO, The Jackson Laboratory for Genomic Medicine, Farmington, USA



*Peter Robinson*

Peter Robinson, the inventor of the Human Phenotype Ontology (HPO) presented us the history of classification for computational uses and his work. HPO aims to provide a standardized

vocabulary of phenotypic abnormalities encountered in human disease. Each term in the HPO describes a phenotypic abnormality, such as Macular hypoplasia. The HPO is currently being developed using the medical literature, Orphanet, DECIPHER, and OMIM. HPO currently contains approximately 11,000 terms (still growing) and over 115,000 annotations to hereditary diseases. The HPO also provides a large set of HPO

annotations to approximately 4000 common diseases. Peter Robinson presented and illustrated the principles that govern ontologies in general and HPO in particular. He also stressed how experts' input is needed to enrich and evolve HPO. He was very enthusiastic to participate to this work with all experts working together on HPO terms.

## *Plenary: Each WG (WG1 to WG4 plus TWG5) present their work on HPO & general discussion*

### WGs leaders and representatives



*Panagiotis Sergouniotis*

Each WG leader of the four clinical WG and the transversal WG5 (low vision group) had the opportunity to present to all members of ERN-EYE and international experts the work done this time regarding HPO terms by his group during the previous months in order to open

the discussion regarding the annotations to be done and phenotypic abnormalities to add to the current HPO with regard to the literature and the clinical knowledge and experience. They pointed out crucial points to discuss.

## *Parallel: Work groups to meet for HPO adjustments*

### WGs



All WG and transversal WG5 meet again this time to work on the HPO part of their work. The same strategy was applied: they worked during the summer period. They thus annotated existing

phenotypic abnormalities terms and add/suppress or modify a lot of them. All this work will be integrated by the HPO team in their system.

## **CLINICAL PATIENT MANAGEMENT SYSTEM SESSION**

### *Plenary: Presentation of the CPMS – Presentation, demonstration and general discussion*

Hany Mina, Open App Project Manager for ERN CPMS

OpenApp is a software development company focused on the Healthcare sector. Its specialties lie in utilizing open source tools to deliver applications, systems & solutions.

They were appointed by the European Commission to build up a full solution for creation of virtual clinics for ERNs. This system is called Clinical Patient Management System. Hany Mina

was assigned by the commission to present us the CPMS system, its architecture and demonstrate the functionalities available with this tool.

## *ERN-EYE general BUSINESS MEETING & Roadmap for 2017/2018*

Hélène Dollfus, ERN-EYE coordinator



*Hélène Dollfus*

Hélène Dollfus, the coordinator of ERN-EYE took advantage of the workshop to hold a business meeting of the network. The progresses of the year were presented. The session closed with a discussion about the next steps

for ERN-EYE, roadmap for the next months (inventory missions, guidelines, genetic testing TWG6, research and “think tank”, Guidelines and teaching, registries), communication tools, next meetings and agenda for 2018.

### *Plenary: Wrap-up session & general conclusions*

WG and TWG leader

Before closing the meeting, leaders of each WG & TWG5 or their representative presented a wrap-up of the work to all

participants. Thus participants from all WGs gave their opinion and appreciated the work done during these intensive days. The

publication of the work done by ERN-EYE in a specialized journal is currently running.



## List of Meeting participants

Name	First Name	Working group
Ashworth	Jane	WG3
Audo	Isabelle	WG1
Balciuniene	Vilma Jurate	WG1
Banin	Eyal	WG1
Black	Graeme	WG1
Böhringer	Daniel	WG4
Boon	Camiel	WG1
Bremont-Gignac	Dominique	WG3
Calvas	Patrick	WG3
Castela	Guilherme	WG3
Dagnelie	Gislin	TWG5 Low vision
Dollfus	Hélène	WG3
Downes	Susan	WG1
Fasolo	Adriano	WG4
Fasser	Christina	TWG5 Low vision
Gelzinis	Arvydas	WG3
Goetz	Kerry	WG1
Hamann	Steffen	WG2
Heon	Elise	WG3
Iarossi	Giancarlo	WG1
Kawasaki	Aki	WG2
Keegan	David	TWG5 Low vision
Kessel	Line	WG3
Khan	Kamron	WG1
Klett	Artur	WG4
Leroux	Dorothee	
Leroy	Bart Peter	WG1
Lisch	Prof.Dr.med.Walter	WG4
Liskova	Petra	WG4
Lorenz	Birgit	WG3
Maggi	Riccardo	
Maxime	Emmanuel	
Meunier	Isabelle	WG1
Mina	Hany	
Mohand-Said	Saddek	TWG5 Low vision
Nowomiejska	Katarzyna	WG1
Perdomo	Yaumara	WG1
Petzold	Axel	WG2
Preisling	Markus	WG3
Robinson	Peter	
Scholl	Hendrik	
Sergouniotis	Panagiotis	WG3
Sodi	Andrea	WG1
Stingl	Katarina	WG1

<i>Studer</i>	<i>Fouzia</i>	<i>TWG5 Low vision</i>
<i>Suppiej</i>	<i>Agnese</i>	<i>WG3</i>
<i>Thompson</i>	<i>Rachel</i>	
<i>Touitou</i>	<i>Valerie</i>	<i>WG2</i>
<i>Traboulsi</i>	<i>Elias</i>	
<i>Trumpaitis</i>	<i>Jurgis</i>	<i>WG4</i>
<i>Tuft</i>	<i>Stephen</i>	<i>WG4</i>
<i>Vaclavic</i>	<i>Veronika</i>	<i>WG1</i>
<i>Valeina</i>	<i>Sandra</i>	<i>WG3</i>
<i>Van Cauwenbergh</i>	<i>Caroline</i>	<i>WG1</i>
<i>Verloes</i>	<i>Alain</i>	<i>WG3</i>
<i>Vighetto</i>	<i>Alain</i>	<i>WG2</i>
<i>Wheeler</i>	<i>Russell</i>	<i>WG2</i>
<i>Wheeler-Schilling</i>	<i>Thomas</i>	
<i>Yu-Wai-Man</i>	<i>Patrick</i>	<i>WG2</i>
<i>Zobor</i>	<i>Ditta</i>	<i>WG1</i>
<i>Zrenner</i>	<i>Eberhart</i>	<i>WG1</i>

## Contacts

ERN -EYE

Hôpitaux Universitaires de Strasbourg

1, place de l'hôpital - Bâtiment 2

67091 STRASBOURG CEDEX

FRANCE

+33 (0)3 88 11 67 53 (reception)

[ern.eye.project@chru-strasbourg.fr](mailto:ern.eye.project@chru-strasbourg.fr)