

Network Coordinator

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**European
Reference
Network**

for rare or low prevalence
complex diseases

 **Network**
Eye Diseases (ERN-EYE)

ERN-EYE Kick-Off Meeting brief report

3rd to 5th April 2017,
T ubingen, Germany

LEROUX Doroth e, Project Manager

ERN-EYE Kick-Off Meeting brief report

ERN-EYE Project Management Team

29/06/2017

Introduction

Kick-Off Meeting in Tübingen, Germany from April, 3rd to April, 5th, 2017

Professor Hélène Dollfus opened the first meeting of the European Reference Network ERN-EYE, which officially launched the work of 29 health care providers from 13 European member states.

Rare Eye Diseases (RED) are the leading cause of visual impairment and blindness for children and young adults in Europe. More than

neuro-ophthalmology rare diseases, paediatric ophthalmology rare diseases, and rare anterior segment conditions. In addition, six transversal working groups are addressing issues common to the four main themes. Additional working groups focus on specific areas, including genetic testing,

The Kick-Off Meeting officially launched the European Reference Network ERN-EYE, which is composed of 29 health care providers from 13 European member states.

900 REDs are listed in the portal for rare diseases and orphan drugs (ORPHANET). These include more prevalent diseases such as retinitis pigmentosa which has an estimated prevalence of 1 in 5 000, as well as some very rare entities described only once or twice in medical literature. ERN EYE addresses these conditions in four thematic groups: rare diseases of the retina,

registries, research, education, communication and patients.

The network's main aim is the development of a virtual clinic — known as EyeClin — to guarantee the best coverage of REDs and facilitate cross-border dissemination of expertise.

ERNs in brief

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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 will be 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".



Opening Session

Various speakers

The conference opened with inaugural welcome speeches of H el ene Dollfus, the ERN-EYE coordinator, Eberhart Zrenner, the host of this meeting in the beautiful new Eye Clinic in T ubingen,



Christina Fasser and H el ene Dollfus

Christina Fasser, Funding member and Patient representative in the Coordinating committee) and finally a video message from Christophe Gautier, CEO of H opitaux Universitaires de Strasbourg, the



Eberhart Zrenner

ERN-EYE coordinator's HealthCare Provider.

*"The knowledge travels,
not the patient"*



Enrique Terol

The ERNs in the Rare Diseases environment in Europe

Enrique Terol, Seconded National Expert, Policy Officer, European Reference Networks, European Commission, DG SANTE, Brussels Belgium. *By teleconference*

On the theme of "The ERNs in the Rare Diseases in Europe", Enrique Terol spoke about the importance of ERNs in the European Union and the possibilities offered by this

network for the care of patients their countries and the pooling of expertise. He welcomed the establishment of ERN-EYE and its extension to 13 countries.



Enrique Terol

EURORDIS point of view of ERN objectives

Matt Johnson, Healthcare and Research Director, EURORDIS, Brussels, Belgium



Matt Johnson

As a key partner in the European Joint Action on Rare Diseases, EURORDIS works with the Commission Expert Group on Rare Diseases, clinical leads and patient representatives to help shape the scope of rare disease ERN, potential services provided by

successful ERN and to ensure patient representation in potential ERN boards and sub-clinical committees. EURORDIS is working to ensure that every rare disease patient has a home within the ERN system.

The ERN-EYE project: Brief summary of the overall project, main goals and involvement of each member. Working strategy and plan for 2017

Hélène Dollfus, ERN-EYE Coordinator, HUS, Strasbourg, France

In her presentation Prof. Hélène Dollfus recalled the history of the ERN-EYE project and its launch, shortly before the Kick-Off Meeting. She named the 29 hospitals involved and the workshops developed. Finally, she cited all the objectives: improve patient trajectory in the EU and reduce inequalities of



patient care in the EU, reduce the

time to diagnosis and care, bring diagnosis to the undiagnosed, ensure molecular diagnosis to the largest number of patients in the EU, facilitate involvement of patients in clinical trials and give access to innovation.

Communication rules for ERNs

Dorothee Leroux, ERN-EYE Project Manager, HUS, Strasbourg, France

Dorothee Leroux explained all the rules in the field of communication for ERN-EYE. Thus, the logo, the writing, the graphics can only be

used according to a specific graphic charter that meets many criteria. This is intended to unify

the communication of each member of the network.



Avril Daly



David Keagan

The ERN-EYE Members across the EU – A brief overview

Various speakers

A representative of each member states (13 MS) presented the MS HCPs participants, the general organization in their country for rare

diseases and more specifically rare eye diseases, expectations and involvement in ERN-EYE for the next years.



Francesco Testa

Member State	SPEAKER	HCP
Belgium	Bart Leroy	Ghent University Hospital, Ghent, Belgium
Czech Republic	Petra Liskova	General University Hospital in Prague, Czech Republic
Denmark	Michael Larsen	Rigshospitalet, Glostrup, Denmark
Estonia & Latvia	Sandra Valeina (in the name of Artur Klett for Estonia)	Children's Clinical University Hospital, Riga, Latvia
France	Hélène Dollfus	CARGO, Hôpital Universitaire de Strasbourg, Strasbourg, France
Germany	Daniel Boehringer	Eye Centre, University Hospital Freiburg, Freiburg, Germany
Italy	Francesco Testa	Azienda Ospedaliera Universitaria, Second University of Naples
Lithuania	Vytautas Jasinskas	Hospital of Lithuanian University of Health Science, Kauno Klinikos, Kaunas, Lithuania
Netherlands	Camiel Boon	Leiden University Medical Center/Academic Medical Center Amsterdam, Netherlands
Poland	Katarzyna Nowomiejska	Medical University in Lublin, Lublin, Poland
Portugal	Jao Pedro Marques	Centro Hospitalar e Universitário de Coimbra, Coimbra, Portugal
United Kingdom	Kamron Khan	Central Manchester University Hospitals NHS Foundation Trust, Manchester Academic Health Sciences Centre, Manchester, UK

EVICRnet

Cecilia MARTINHO, CEO of AIBILI, Coimbra, Portugal

Cecilia Martinho explained the role of European Vision Institute Clinical Research Network. It is a network of European Ophthalmological Clinical Research Sites, dedicated to perform clinical research in ophthalmology with the highest standards of quality, following the

European and International Directives for Clinical Research according to harmonized SOPs in order to strengthen the capacity of the European Union to study the determinants of ophthalmic diseases and to develop and optimize the use of diagnostic,

prevention and treatment strategies in ophthalmology.



Cecilia Martinho

Ontology & DataBases, classification of diseases, FAIR data approach, Preparation of October Meeting

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

Annie Olry, Orphanet, Plateforme Maladies Rares, Paris, France, by teleconference



Rachel Thompson

This session was a first approach to organize the ERN-EYE future ontology work. Rachel Thompson presented the Rare Disease Data

Base environment, bottlenecks, principles of data sharing. Annie Olry presented the Orphanet classification principles and all tools developed by Orphanet to deal with ontologies. As ontology work, as a corner stone of all IT tools, is the first goal of newly created ERN, the debate with the assembly was rich and constructive. This session was a good start to prepare the future

ontology meeting that will take place in Mont Ste-Odile from 9th to 11th October 2017.



Annie Olry

Presentation of each TWG by leaders, key objectives for 2017 and the 5 next years

TWG leaders



Graeme Black

In ERN-EYE, 6 transversal working groups: Low vision, daily life and patient group, Genetic diagnostics, Registries & epidemiology, Research,

Education, training & guidelines, Communication, management & quality. Each TWG presented their key objectives for 2017 and for the 5 next years.

TWG number	Theme	Chairs
TWG5	Low Vision and Daily Life Patient Group	S. Mohand-Said (CHNO-XV-XX) and D. Keegan (MMUH – pending)
TWG6	Genetic Diagnostics	G. Black (MREH- MAHSC) and F. Cremers (RUMC)
TWG7	Registries and Epidemiology	C. Boon (LUMC/AMC) and K.Khan (JUH)
TWG8	Research	D. Fisher (EKUT) and C.Boon (LUMC/AMC)
TWG9 & TWG10	Education, Training & Guidelines	T. Wheeler-Schilling (EKUT) and H. Dollfus (HUS)
	Quality, Communication & Management	H. Dollfus and D. Leroux (HUS)

PARALLEL SESSIONS: First parallel Working session for all TWGs (5 to 10)



Bart Leroy

The transversal working groups met to begin work on the roadmaps allocated to them by the coordination of the project. The progress of each group will then be

presented during the workshop at Mont Ste-Odile from 9th to 11th October 2017.

How to work on Guidelines in Europe

Sheela Upadhyaya, Associate Director Highly Specialized Technologies, Centre for Health Technology Evaluation, National Institute for Health and Care Excellence, London, UK

Sheela Upadhyaya presented the key principles of guidelines development process and a rich discussion with the audience followed her talk. Guidelines are crucial for all ERNs in general and

for ERN-EYE to ensure the best possible care for patients and the production of guidelines is scheduled for the next years within the network.



Sheela Upadhyaya and Hélène Dollfus

Presentation of each WG by leaders, key objectives for 2017 and the 5 next years

WG leaders



Daniel Böhringer

ERN-EYE is composed of 4 clinical thematic working groups: Retinal rare eye diseases, Neuro-ophthalmology rare diseases, Pediatric ophthalmology rare

diseases and anterior segment rare eye diseases. Each WG presented their key objectives for 2017 and for the 5 next years.

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	T. Reinhard & D. Boehringer (UMCF) and P. Liskova (HPH)

PARALLEL SESSIONS: First parallel Working session for all TWGs (5 to 10)

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Mont Ste-Odile from 9th to 11th October 2017.

Wrap-up from Tuesday's sessions: each leader presents the 2017 workplan for WG/TWG

WG and TWG leaders

A restitution of WG and TWG working session to the whole assembly by WG&TWG leaders

allowed all participants to have an overview of the project.



Specific meetings



Aside the general program, other meetings took place:

- The Patient & Families Advisory council Meeting with ePAG representatives
- The advisory Board Meeting
- The Governance Board

Meeting allowed to vote and officialize all important decisions of the network. This governance Board is constituted by all HCP representatives (1 per HCP) and 2 elected patient representatives among the PaFaC.

Contacts

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