ERN-EYE first year, set the scene for e-Health within the network: a common ontology for all Rare Eye Diseases and first achievements of the year

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

www.ern-eye.eu

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Acknowledgement

Summary

European Reference Networks (ERNs) are unique and innovative networks between healthcare specialists for the diagnosis and treatment of rare/low prevalence complex diseases. **ERN-EYE is dedicated to Rare Eye Diseases (RED)** and is organized to cover **all RED conditions in four thematic groups** and six transversal working groups and had fully integrated patient representatives (e-PAG noticeably). Launched in March 2017, ERN-EYE conducted during its first year an inventory of available resources and started to deal with corner stones for an efficient use of all e-health tools. The most important ones were the work on a standardized ontology for Rare Eye Diseases and the settlement of a virtual clinic.

ERN-EYE, a one-year old network dedicated to care

ERN-EYE, European Reference Network dedicated to Rare Eye Diseases (RED), is structured in four thematic working & transversal working groups as described in Figure 1. ERN-EYE had **fully integrated patient representatives** (European Patient Advocacy Groups (e-PAG) noticeably) organized in a specific council with **2 elected representatives** with voting rights in the governance board. **One e-PAG representative is co-chairing a transversal working group**, and besides, **at least one e-PAG representative attend each ERN-EYE meetings or workshops**.
Launched in March 2017, ERN-EYE set all necessary bases to develop activities. The main activities are described in Figure 2.
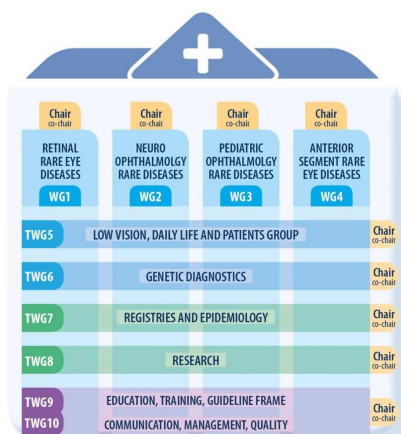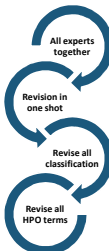
ERN-EYE it's:
- 29 Hospitals
- 24 000 patients in our centers
- 13 countries
- 900 Rare Eye Diseases

Figure 1: ERN-EYE Working groups and transversal Working Groups

Chair / co-chair — WG1 RETINAL RARE EYE DISEASES | WG2 NEURO OPHTHALMOLGY RARE DISEASES | WG3 PEDIATRIC OPHTHALMOLGY RARE DISEASES | WG4 ANTERIOR SEGMENT RARE EYE DISEASES
TWG5 LOW VISION, DAILY LIFE AND PATIENTS GROUP — Chair co-chair
TWG6 GENETIC DIAGNOSTICS — Chair co-chair
TWG7 REGISTRIES AND EPIDEMIOLOGY — Chair co-chair
TWG8 RESEARCH — Chair co-chair
TWG9 EDUCATION, TRAINING, GUIDELINE FRAME
TWG10 COMMUNICATION, MANAGEMENT, QUALITY — Chair co-chair

Figure 2: ERN-EYE first year main activities

- Standardized ontology
- Website creation & Communication plan
- Resources inventory
- Guidelines program initiation
- Research & clinical trial inventory
- Management of the network & ERNs Workshops
- Education & Training program initiation
- Initiation of the virtual clinic: tests and Eye dataset

The most important achievement: agree on the classification of REDs and ontology

▶ **IMPORTANT To launch the ERN-EYE general activities**
 ▶ agree on a common tools ⇨ enable ERN members to exchange between each other and beyond.
 ▶ consensual curation & revisiting of the existing rare eye diseases ontologies/coding with ORPHANET and HPO
  ➢ Save time by doing a physical meeting
  ➢ One shot action on the year
▶ Close collaboration with
 ▶ ORPHANET (at the fore front of rare diseases nomenclature),
 ▶ HPO (the Human Phenotype Ontology leader in phenotypic classifications of rare diseases)

All experts together → Revision in one shot → Revise all classification → Revise all HPO terms

Up to 60 participants (including clinicians, patient representatives and ontology developers) | 17 countries represented

Agreement on :
1390 terms related to RED on the Orphanet Rare Diseases Ontology (ORDO)
& including **1089 terms related to ocular phenotypes in HPO**

Development of the virtual clinic EyeClin

The creation of a virtual Clinic, EyeClin, is the heart of ERN-EYE, to bring expertise to RED-affected EU citizens. Based on the generic version provided by the European Commission we developed a dataset dedicated to the EYE (Figure 3) in order to use specifically the virtual clinic for REDs (Figure 4).
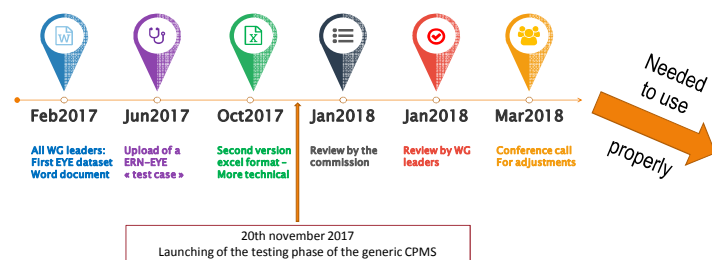

Feb2017 — All WG leaders: First EYE dataset Word document
Jun2017 — Upload of a ERN–EYE « test case »
Oct2017 — Second version excel format – More technical
Jan2018 — Review by the commission
Jan2018 — Review by WG leaders
Mar2018 — Conference call For adjustments

20th november 2017 Launching of the testing phase of the generic CPMS

Figure 3: Timeline to establish the EYE-specific dataset of the virtual clinic

Needed to use properly

PATIENTS — CLINICIAN — HEALTHCARE PROVIDERS — clinical data
RECOMMAN-DATIONS — EXPERTS PANEL
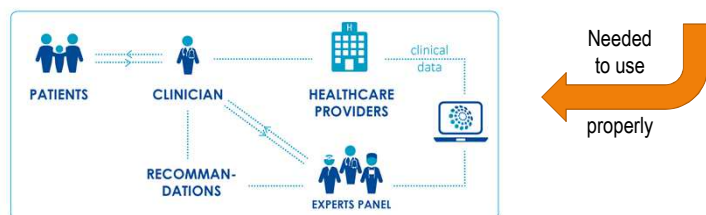
Needed to use properly

Figure 4: Chart of the Virtual clinic

Conclusion

ERN-EYE, has already shown great achievements in establishing a **deep inventory of resources**, **agreeing on a common ontology** and **developing an Eye-specific dataset** to ensure the launch of a **virtual clinic dedicated to RED, EyeClin**. This inventory year was an excellent base to build on and the common ontology and the EyeClin are **building blocks** of our next activities **for the following years**.

Les Hôpitaux Universitaires de STRASBOURG

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