ERN-EYE

PROGRAM of the Workshop Genetic Testing in Rare Eye Diseases (October 10th – October 12th, 2018)

MEETING LOCATION:

Careggi University Hospital
Largo Brambilla
3 50134 Firenze, Italy

WEDNESDAY – 10th OCTOBER 2018

ERN-EYE WORKING GROUPS CAN MEET ALREADY IN THE MORNING IN MEETING ROOMS ON SITE

13.00	Registration of participants
14:00-14:20	**Welcome** meeting opening (Chair HD) Hélène Dollfus, ERN-EYE Coordinator, HUS, Strasbourg, France Andrea Sodi, ERN-EYE member, Florence, Italy, host of the meeting Maria Teresa Mechi, Responsabile Qualità dei servizi e reti cliniche, Regione Toscana
14: 20-14:40	**Presentation of present pathway to genetic diagnostic from the Patient perspective** Christina Fasser, ePAG representative Avril Daly EURORDIS vice-president & CEO of Retina International, Dublin, Ireland

INTRODUCTION OF THE WORKSHOP

14:40-15:00	**Setting the scene & objectives of the workshop** Hélène Dollfus, ERN-EYE Coordinator, HUS, Strasbourg, France Graeme Black, Chair of TWG6 for genetic testing, Manchester, UK
15:00-15:10	**Results of the SURVEY: Overview genomic testing across ERN-EYE members** Dorothée Leroux, ERN Project manager, Strasbourg, France

SESSION 1: SITUATION FOR GENETIC TESTING ACROSS THE MEMBER STATES PARTNERS OF ERN-EYE (Chair: Hélène Dollfus)

15:10 – 16:00 **PART 1 Situation of each member state for ERN –EYE (by one delegate for each member state)**

Member State	SPEAKER
Belgium	Bart Leroy
Czech Republic	Petra Liskova
Denmark	Michael Larsen
Estonia	Artur Klett
France	Hélène Dollfus

16:00- 16:30 COFFEE BREAK

16:30 – 18:00 **PART 2 Situation of each member state for ERN –EYE by one delegate for each**

(TBD = To Be Determined for MS where there are more than one HCP)

Member State	SPEAKER
Germany	TBD
Italy	TBD
Latvia	Sandra Valeina
Lithuania	Arvydas Gelzinis
Netherlands	Lonneke Haer-Wigman
Poland	Katarzyna Nowomiejska
Portugal	TBD
United Kingdom	TBD

18:15-18:30 **Challenges in setting up IRD diagnostics in Poland**
Anna Tracewska, Wroclaw Research Centre EIT, Wroclaw, Poland

18:30-18:45 **Challenges in Genetic testing in Romania**
Adela Chirita-Emandi, Department of Genetics of the University of Medicine Timisoara, Romania

19:00 APERITIF with each participant bringing a solid or liquid "speciality"

THURSDAY - 11th OCTOBER 2018

09:00-09:10 **Short** opening and welcome address
Rocco Damone, Direttore Generale AOU Careggi

SESSION 1: GENOMIC TESTING IN EVERY DAY CLINICAL PRACTICE: HOW IS IT ORGANIZED TODAY AND HOW DOES THIS HELP PATIENTS AND FAMILIES? (Chair: Graeme Black)

09:10- 09:30 **The concept of clinical utility in genomics**
Panagiotis Sergouniotis, Manchester, UK

09:30-09:50 **The role of the clinical laboratory**
Caroline Van Cauwenbergh, Ghent, Belgium

09:50–10:10 **The role of the genetic counsellor and the importance of multidisciplinary working**
Georgina Hall, Manchester, UK

10:10–10:30 **Genomics in everyday IRD practice in France PANELS WES WGS**
Hélène Dollfus, France

10:30–10:50 **A transatlantic perspective of genomic in every day practice**
Elise Héon, Hospital for Sick Children, Toronto, Canada

10:50-11:20 COFFEE BREAK

SESSION 2: GENOMIC DIAGNOSTIC: STATE OF THE ART AND NEEDS FOR THE ERN-EYE WORKING GROUPS

ERN-EYE covers various fields of rare eye diseases and the needs and challenges can be quite different accordingly. Each workgroup will follow a general canvas and cover 2 or 3 main specific topics about genetic testing specificities

WG1 - RETINAL RARE EYE DISEASES (Chair: Michael Larsen)

11:20–11:35 **Retinal dystrophies and genetic testing, the current status and the current needs for genomic diagnostic everyday IRD practice**
Bart Leroy, Ghent, Belgium

11:35-11:50 **Targeted (gene panel) RED diagnostics using WES; from syndromes to isolated RP. Comparison with other rare diseases and phenotype-genotype correlations**
Lonneke Haer-Wigman, Nijmegen, Netherland

11:50-12:05 **Achromatopsia and related disorders: what genetic testing has taught us?**
Susanne Kohl, Tübingen, Germany

12:05-12:20 **CSNB : what has genetic testing told us?**
Christina Zeitz, Paris, France

WG2 - NEURO- OPHTHALMOLOGY RARE DISEASES (Chair: Steffen Hamann)

12:20- 12:35 **What are the clinican's need for RED in neuro-ophthalmology?**
Axel Petzold, London TBC

12:35-12:50 **Optic neuropathies in the genetic laboratories**
Patrick Yu Wai Man, Moorfields Eye Hospital, London, UK

12:50-14:00 LUNCH

WG3 - PAEDIATRIC OPHTHALMOLOGY (Chair: Birgit Lorenz)

14:00-14:15 **Genomic testing in a busy pediatric setting**
Jane Ashworth, Chair of WG3, Manchester

14:15-14:30 **Microphthalmia, aniridia**
Patrick Calvas, CHU Toulouse, France

14:30-14:45 **LCA: how the clinical background enlightens the genetic testing and vice-versa**
Jean-Michel Rozet, Paris, France

WG4 - ANTERIOR SEGMENT (Chair: Daniel Bohringer)

14:45-15:00 **The clinician's need in genetic anterior segment diseases: from cornea to lens**
Petra Liskova , Prague

15:00- 15:15 **Anterior segment and genetic diagnosis**
Jane Sowden, University College, London, UK

15:15-16:00 **General discussion**

16:00-16:30 COFFEE BREAK

SESSION 3: Challenges and Future issues for the genetic testing group in the ERN-EYE (Chair: Hélène Dollfus)

16:30-17:10 **Different rules and practices in EU member states for genetic testing: cross border and economical aspects**
Helena Kaariainen, National Institute for Health and Welfare, Helsinki, Finland

17:10-17:40 **Ethical EU aspects on genetic testing**
Anne Cambon Thomsen, Toulouse, France

17:40-18:10 **Ethical EU aspects on genetic testing and consent**
Davit Chokoshvili, Leuven, Belgium

18:10-18:30 **The patient perspective: what does genetic testing mean?**
Christina Fasser, ePag representative

ADDITIONAL INFORMATION

18:30-18:50 **New proposed legislation for harmonised Health Technology Assessement in Europe**
Russel Wheeler, ePag representative

19:00 Joint diner to be confirmed

FRIDAY - 12th OCTOBER 2018

SESSION 1: CONCLUSIONS NEEDS AND HOPES – PREPARATION OF REPORT and EDITORIAL

9:00 – 9:30 **General conclusions- Wrap up – preparation of the EDITORIAL FOR OPHTHALMIC GENETICS** (Chairs: Graeme Black – Frans Cremers)

SESSION 2: Development and research around genetic testing in the EU (Chair: Frans Cremers)

9:30-10:00 **Follow-up research explorations ('open-the-exome') of WES data sets for REDs and first results WGS of 'WES-negative' samples**
Susanne Roosing, Radboud University Medical Center, Nijmegen, Netherlands

10:00-10:30 **Results from large cohort WGS studies in IRD: promises and challenges**
Gavin Arno, University College, London, UK

10:30-11:00 COFFEE BREAK

11:00- 12:00 **Roundtable discussion: Is there a need to organize RED centers of expertise in Europe and how can they be established?**
Chair: Frans Cremers, Nijmegen, NL

- RPGR-ORF15, OPN1LW/OPN1MW, ABCA4 non-coding, CNV analysis, etc
- How can centers provide training to starting professionals?
- Are their mobility funds available?

12.00-12.30 **GENERAL CONCLUSION**

12:30 END OF THE MEETING

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