Thursday 15th September 2022, 13.00 – 17.00.
Magenta B room in the Bologna Congressi S.r.l

EHDN Joint Meeting of the Genetic Counseling & Testing Working Group, Genetic Modifiers Working Group and Incidental Findings Task Force

Application of genetic modifier of Huntington's disease data in the clinical setting: promise, pitfalls & development of a framework.

12.00: A buffet lunch will be served in the foyer, in the shared catering area, which participants of this meeting are invited to attend.

13.00: Introduction to the afternoon
Dr Nayana Lahiri, Dr Tom Massey, Dr Rhona MacLeod, Dr Davina Hensman Moss

13.10- Talk 1: The clinical dilemma: a genetic counselling perspective (15 mins)
Jill Goldman MS, MPhil, CGC; Genetic Counselor at The Taub Institute at Columbia University Medical Center

13.25- Talk 2: Overview of genetic variants which have been found to modify Huntington's disease onset and progression. (10 mins)
Dr Tom Massey, Clinical Lecturer, University of Cardiff & Dr Davina Hensman Moss, Clinical Lecturer, St George's University of London

13.35- Talk 3: The role of the CAG repeat and CAA effects in Huntington's disease: considering the importance of *HTT* sequence data for diagnostics. (10 mins)
Prof Darren Monckton, Professor of Human Genetics, University of Glasgow

13.45- Talk 4: Polygenic risk scores in Huntington's disease (10 mins)
Prof Peter Holmans, Professor of Biostatistics and Genetic Epidemiology, Division of Psychological Medicine and Clinical Neurosciences, University of Cardiff

13.55- Talk 5: Implementation of Polygenic Risk Scores in Clinical Practice for Breast Cancer: a UK perspective (20 mins)
Prof D Gareth Evans MB BS MD FRCP FLSW FRCOG ad eundem, University of Manchester

14.15- Talk 6: The perspective of the person at risk (15 mins)
Jenna Heilman, Executive Director, HDYO

14.30 COFFEE BREAK (30 mins)

15.00 Talk 5: An ethical perspective (20 mins)
Prof Robert Klitzman, Bioethicist and Professor of Clinical Psychiatry, Columbia University Medical Center

15.20: Questions to presenters

15.45: General discussion among all participants: topics to discuss include:
Can we identify gaps in the current knowledge that need to be addressed?
Are effect sizes big enough to be clinically relevant?
Do we know what people at risk want to know?

What are the implications of genetic modifier data & polygenic risk scores for clinical trials, and implications of any use back to the clinic?

In this session we will aim to construct a framework for the application of genetic modifier of HD data in the clinical setting.

17.00: Session concludes.

Please note that the meeting organisers are not presuming that these data *should* be applied clinically: at this stage we merely advocate that this is an important topic for open discussion in the community.

Organisers:

Dr Davina J Hensman Moss, Genetic Modifiers Working Group
Dr Tom Massey, Genetic Modifiers Working Group
Dr Rhona MacLeod, Genetic Counselling & Testing Working Group
Dr Nayana Lahiri, Genetic Counselling & Testing Working Group and Incidental Findings Task Force