Reverse Phenotyping and Pattern Recognition in Genetic Malformations of the Brain

COST Action CA16118, Ancillary meeting to the ESHG 2018, Milano, June 19th. Room Yellow-3

Congress Venue – MiCo Milano Congressi
Piazzale Carlo Magno, 1
Viale Eginardo – GATE 2 – 20149 Milan, Italy

The initiative for this ancillary meeting raised within the European Network on Brain malformations, Neuro-MIG. This consortium is supported by the European Committee for Science and Technology (COST Action CA16118). The spirit is that by giving an overview of the clinical clues to recognize and diagnose malformation of cortical development (MCD), Neuro-MIG stimulates discussion and broad participation to most relevant unsolved issues in genetics.

With the advances of "unbiased" genome wide analysis, often leading to detection of unclassified variants, there is an increasing urge to develop skills in reverse phenotyping, essential in the clinical genetics practice. This is a complex task when approaching the field of brain malformations, which involves multidisciplinary expertise and requires the geneticist to gather information of very different kind, e.g. from neurology, pathology, neurophysiology, morphology, ranging from brain imaging to clinical dysmorphology.

The goal is to illustrate the state-of-the-art distinctive patterns that can be recognized with a high degree of certainty as related to specific genetic causes and MCD syndromes.

Distinctive anomalies can be recognized ex-vivo at the anatomical and pathological level, but also in vivo by dedicated MRI imaging, by electrophysiological characteristics or dysmorphology clues. Also the main types of genomic techniques and the advantages of custom-designed NGS panels versus whole genome/exome analysis will be reviewed. All presenters are members of the Neuro-MIG.

Stakeholders are clinicians and research scientists from different medical and pre-medical disciplines and different career status who are involved in diagnosis and care of people suffering from brain malformations or have brain malformations as scope of their research. We hope that participants involved in patient care will be able to make use of the information in the daily practice.

NB: Participation is free of charge, but due to limited space at the venue, participants are requested to register in advance mailing to: neuro-mig@cardiff.ac.uk

Further information on the website: www.Neuro-MIG.org

On behalf of the Neuro-MIG, I hope to welcome you in Milano!

Grazia M.S. Mancini, Chair of the Action

Preliminary Programme

June 19th, Morning 8.45-13.30

Registration 8.45-9.00

Anatomic/Pathology clues

9.00-9.20 Brain development and abnormalities — tbc

Clinical Syndromes associated with brain malformation

9.20-9.40 Dysmorphology — Renske Oegema, Utrecht

9.40-10.00 Epilepsy syndromes — Renzo Guerrini, Firenze

10.00-10.20 Neurologic syndromes (PNP, myopathy, Mov. Dis.) Anna Jansen, Brussels

10.20-10.40 Coffee break

MRI clues

10.40-11.00 Cortex: lissencephaly+Tubulin vs non-genetic — Bill Dobyns, Seattle

11.00-11.20 Megalencephaly and polymicrogyria — Ghayda Mirzaa, Seattle

11.20-11.40 Cerebral with cerebellar patterns — Mariasavina Severino, Genova

11.40-12.00 Microcephaly — Nadia Bahi-Buisson, Paris

Reverse phenotyping: MCD Detection without clinical diagnosis

12.00-12.20 Diagnostic NGS panels versus WES — Martina Wilke, Rotterdam

12.20-12.40 Animal models of cortical malformations: from function to pathophysiology
Carlos Cardoso, Marseille

12.40-13.00 Brain organoids in MCD — Orly Reiner, Rehovot

13.00-13.20 WES vs WGS and the future — Enza Maria Valente, Pavia