Reverse Phenotyping and Pattern Recognition in Genetic Malformations of the Brain

COST Action CA16118 Neuro-MIG

Satellite symposium to the EPNS 2019 in Athens

September 17th

Room MC2 Hall

Congress Venue – Megaron Athens International Convention Centre (MAICC)

The initiative for this satellite symposium was born within the European Network on Brain malformations, Neuro-MIG. This consortium is supported by the European Cooperation in Science and Technology (COST Action CA16118).

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 clinical practice. This is a complex task when approaching the field of brain malformations, which involves multidisciplinary expertise and requires the clinician to gather diverse information, e.g. from neurology, molecular genetics, dysmorphology, brain imaging, neurophysiology, and pathology.

The goal of this educational session is to discuss the diagnostic approach for MCD and show the distinctive patterns that are highly associated with specific genes and 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 physical examination.

All presenters are members of the Neuro-MIG. We welcome both clinicians and research scientists from different medical and pre-medical? disciplines and different career stages who are involved in diagnosis and care of people affected by 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 daily practice.

Participation is free of charge, but due to limited space at the venue, participants are requested to register in advance by email to: neuro-mig@cardiff.ac.uk
CME accreditation will be applied for

Further information on the website: https://www.neuro-mig.org/events

On behalf of Neuro-MIG, we hope to welcome you in Athens!

Grazia Mancini, Anna Jansen, Nadia Bahi-Buisson, Daniela Pilz and Dina Amrom

Preliminary Program
Tuesday September 17th, M2, Theatre -1

Registration 8.45-9.00

9.00-9.05 Welcome by the Action Chair Grazia Mancini

Approach to MCD

09.05 – 9.30: Approach to prenatally diagnosed MCD - Tally Lerman-Sagie, Tel Aviv, Israel

09.30 – 10.00: Approach to postnatally diagnosed MCD - Renske Oegema, Utrecht, the Netherlands

10.00 – 10.30: Diagnostic clues from the neurological examination - Anna Jansen, Brussels, Belgium

10.30 – 11.00: Pitfalls in the MCD diagnostic work-up - Nataliya Di Donato, Dresden, Germany

11.00 – 11.30: coffee break

MCD phenotyping

11.30 – 12.00: Microcephaly – Ghayda Mirzaa, University of Washington, Seattle, USA

12.00 – 12.30: Polymicrogyria - Andrew Fry, Cardiff, UK

12.30 – 13.00: Tubulinopathies and dyneinopathies - Nadia Bahi-Buisson, Paris, France

13.00 – 14.00: lunch

14.00 – 14.30: Lissencephaly - Nataliya Di Donato, Dresden, Germany

14.30 – 15.00: Heterotopia - Renske Oegema, Utrecht, the Netherlands

From the clinic to the lab and back

15.00 – 15.30: Multidisciplinary interaction and MCD gene discovery - Grazia Mancini, Rotterdam, the Netherlands

15.30 – 16.00: Models to study brain development - Carlos Cardoso, Marseille, France