

WG1 and WG3 Meeting Program- COST Action CA16118

Malta 2018

Wednesday February 21st

- 15.00-15.20 start meeting, welcome, introduction participants, goal of the meeting (Renske / Nataliya)
- 15.20- 15.40 Dr. Frans Verheijen –Functional validation of variants (mTOR signaling, ciliopathies, zebrafish..)
- 15.40 - 16.00 Prof. Orly Reiner – Use of on-chip brain organoids to model lissencephaly
- 16.00 - 16.15 questions / experiences / ideas participants
- 16.15 - 16.30 coffee break
- 16.30- 17.00 Dr. Renske Oegema – MCD nomenclature / HPO project
- 17.00-18.00 discussion HPO terms
- 18.00-19.00 All participants – case review session
- 19.00 End meeting
- 19.30 Dinner at hotel

Thursday February 22nd

- 8.30-8.45 Start meeting – welcome (Renske / Nataliya)
- 8.45-9.05 Dr. Edith Said – MCD in Malta, experiences and challenges
- 9.05- 9.25 Prof. Ute Hehr - Clinical management of patients with LIS1-related lissencephaly and FLNA-associated PNH
- 9.25-10:15 Group discussion, MCD diagnosis and management, goals and challenges in Europe, preparation manuscript draft
- 10.15-10.30 Coffee break
- 10.30-10.50 Dr. Nataliya DiDonato- Genetics of lissencephaly: overview
- 10.50-11.05 Dr. Martina Wilke – MCD molecular diagnostics strategy Rotterdam
- 11.05- 11.20 Dr. Valerio Conti / Dr. Elena Parini - MCD molecular diagnostics strategy Florence
- 11.20-11.35 Prof. Ute Hehr - MCD molecular diagnostics strategy Regensburg
- 11.35-11.50 Dr. Katrien Stouffs - - MCD molecular diagnostics strategy Brussels
- 11.50-12.30 MCD genes, coverage across different exome enrichment kits, current diagnostic yield and group discussion MCD diagnostics strategy (Nataliya)
- 12.30-13.30 Lunch

13.30-14.00 Break

14.00-15.00 Group discussion: Consensus on MCD diagnostics strategy

15.00-15.20 Dr. med. Till Hartlieb/Dr. med. Milka Pringsheim: NETRE study group for sharing personalized therapy strategies and exchange clinical data of children suffering from rare genetic diseases

15.20-15.40 Dr. Romina Romaniello – cerebellar dysplasia in tubulinopathies

15.40-16.00 Coffee break

16.00-17.00 All participants – case review session

17.00 end meeting

17.30 leave for dinner at Valetta

Friday February 23rd

8.00 Start meeting

8.00-8.30 Dr. Carlos Cardoso - animal models in variant and candidate gene validation

8.30- 9.00 Prof. Eleanora Aronica – mTOR pathway-related malformations of cortical development (MCD) -From Histology to an “Integrated diagnosis”

9.00-9.45 Group discussion, challenges in variant interpretation, practical recommendations

9.45-10.00 Coffee break

10.00-11.30 Discussion: key points for the manuscript Best practices in MCD diagnostics;
Closing remarks

11.30 End of the meeting