
Networking event on Progressive Myoclonus Ataxias

Groningen, the Netherlands

1-2 December 2022



Movement disorders
GRONINGEN



Organizing committee

Prof. Marina de Koning-Tijssen

Prof. Tom de Koning

Sterre van der Veen

Sjoukje Polet

Day 1 (Thursday December 1st 2022)

09:00-09:45: Registration of the participants

FIRST SESSION: Phenotyping PMA

Chair: Martje van Egmond

| Time | Title | Speaker |
|-------------|--|---|
| 09:45-09:50 | Welcome and introduction to the PMA Networking Event | Marina de Koning-Tijssen <i>The Netherlands</i> |
| 09:50-10:20 | Introduction into the progressive myoclonus ataxia syndromes | Sterre van der Veen, <i>the Netherlands</i> |
| 10:20-10:50 | Phenotypes of <i>GOSR2</i> , <i>SCARB2</i> , <i>KCTD7</i> , <i>NUS1</i> , <i>DHDDS</i> | Malco Rossi, <i>Argentina</i> |
| 10:50-11:20 | Clinical neurophysiology of PMA | Martje van Egmond, <i>the Netherlands</i> |
| 11:20-11:50 | Neurotransmitters in <i>KCTD7</i> | Farrukh Abbas Chaudhry, <i>Norway</i> |
| 11:50-12:20 | Discussion: PMA classification and diagnostics | Marina de Koning-Tijssen, <i>the Netherlands</i> |
| 12:20-13:20 | Lunch | |

SECOND SESSION: Genetics and underlying mechanisms of PMA

Chair: Dineke Verbeek

| Time | Title | Speaker |
|---------------|--|--|
| 13:20-13:50 | Strategies for gene discovery | Anna-Elina Lehesjoki, <i>Finland</i> |
| 13:50-14:50 | Animal studies | |
| | The <i>CSTB</i> -deficient mouse model for EPM | Saara Tegelberg, <i>Finland</i> |
| | Heat-induced seizures in a <i>GOSR2</i> knockdown model | Roald Lambrechts, <i>the Netherlands</i> |
| 14:50-15:50 | Cellular studies | |
| | Autophagy and <i>GOSR2</i> | Hormos Salimi Dafsari, <i>Germany</i> |
| | Dolichol-dependent glycosylation defects in PMA | Kariona Grabinska, <i>USA</i> |
| 15:50 – 16:20 | Coffee break | |
| 16:20– 16:50 | Discussion: Ongoing and future research: gene discovery and fundamental research | Jenke Gorter <i>the Netherlands</i> |
| 16:50 – 17:20 | Interesting cases | Marina de Koning-Tijssen <i>the Netherlands</i> |

18:30: Dinner at Florentin restaurant (Langestraat 66, 9712 LT Groningen)

Day 2 (Friday December 2nd 2022)

THIRD SESSION: Therapies for PMA

Chair: Serena Galosi

| Time | Title | Speaker |
|---------------|--|---|
| 9:00 – 9:30 | Symptomatic therapies for PMA | David Anderson <i>Scotland</i> |
| 9:30-10:00 | Disease-specific therapies | Sabine Fuchs <i>the Netherlands</i> |
| 10:00-10:30 | Outcome measurements for PMA | Sjoukje Polet <i>the Netherlands</i> |
| 10:30 – 11:00 | Trial design in rare diseases | Joost Raaphorst <i>the Netherlands</i> |
| 11:00-11:30 | Coffee break | |
| 11:30 – 12:00 | Discussion: Ongoing and future research: new therapies and clinical trials | Tom de Koning, <i>the Netherlands</i> |

FOURTH SESSION: Consortium

Chair: Marina de Koning-Tijssen

| Time | Title | Speaker |
|---------------|--|--|
| 12:00 – 12:20 | A story from a patient with PMA | Renske Wassenaar <i>the Netherlands</i> |
| 12:20 – 12:40 | Funding options for the consortium | Paul Bessems <i>the Netherlands</i> |
| 12:40 – 13:10 | Discussion: The role of the PMA consortium | Marina de Koning-Tijssen <i>the Netherlands</i> |
| 13:10 – 14:10 | Lunch | |