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*Networking event on Progressive Myoclonus Ataxias*

*Groningen, the Netherlands*

*1-2 December 2022*

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**Movement disorders**  
GRONINGEN



**Organizing committee**

Prof. Marina de Koning-Tijssen

Prof. Tom de Koning

Sterre van der Veen

Sjoukje Polet

## Day 1 (Thursday December 1<sup>st</sup> 2022)

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

**09:45-09:50:** Welcome and introduction to the PMA Networking Event

### **FIRST SESSION: Phenotyping PMA**

<b>Time</b>	<b>Title</b>
09:50-10:20	Introduction into the progressive myoclonus ataxia syndromes
10:20-10:50	Phenotypes of <i>GOSR2</i> , <i>SCARB2</i> , <i>KCTD7</i> , <i>NUS1</i> , <i>DHDDS</i>
10:50-11:20	Clinical neurophysiology of PMA
11:20-11:50	Neuroimaging and neurotransmitters
11:50-12:20	Discussion: PMA classification and diagnostics
12:20-13:20	Lunch

### **SECOND SESSION: Genetics and underlying mechanisms of PMA**

<b>Time</b>	<b>Title</b>
13:20-13:50	Strategies for gene discovery
13:50-14:50	Animal studies i. Mouse model of <i>CSTB</i> ii. Drosophila model of <i>GOSR2</i>
14:50-15:50	Cellular studies i. Autophagy and <i>GOSR2</i> ii. Glycosylation defects in PMA
15:50 – 16:20	Coffee break
16:20– 16:50	Discussion: Ongoing and future research: gene discovery and fundamental research
16:50 – 17:20	Unsolved cases

**18:30:** PMA Dinner at Florentin restaurant

## Day 2 (Friday December 2<sup>nd</sup> 2022)

### **THIRD SESSION: Therapies for PMA**

<b>Time</b>	<b>Title</b>
9:00 – 9:30	Symptomatic therapies for PMA
9:30-10:00	Disease-specific therapies
10:00-10:30	Outcome measurements for PMA
10:30 – 11:00	Trial design in rare diseases
11:00-11:30	Coffee break
11:30 – 12:00	Discussion: Ongoing and future research: new therapies and clinical trials

### **FOURTH SESSION: Consortium**

<b>Time</b>	<b>Title</b>
12:00 – 12:20	A story from a patient with PMA
12:20 – 12:40	Funding options for the consortium
12:40 – 13:10	The role of the PMA consortium
13:10 – 14:10	Lunch