



**fame**  
bafme

INTERNATIONAL WORKSHOP on  
**FAMILIAL ADULT  
MYOCLONIC EPILEPSY**

**NAPLES ITALY**  
ROYAL CONTINENTAL HOTEL  
**27TH-28TH MAY 2022**



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Awarded to Antonietta Coppola

## ENDORSEMENT

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Dear colleagues and friends, we are very glad to welcome you to the International Workshop on Familial Adult Myoclonic Epilepsy (FAME/BAFME)

We wish you a great meeting and a delightful time in Naples  
Antonietta and Pasquale

### **COURSE DIRECTORS**



Antonietta Coppola



Pasquale Striano

# FACULTY

**Betul Baykan** *Istanbul*  
**Mark Bennett** *Melbourne*  
**Samuel Berkovic** *Melbourne*  
**Leonilda Bilo** *Naples*  
**Francesco Brancati** *Rome*  
**Laura Canafoglia** *Milan*  
**Mar Carreno** *Barcelona*  
**Gianpiero Cavalleri** *Dublin*  
**Sirio Cocozza** *Naples*  
**Antonietta Coppola** *Naples*  
**Mark Corbett** *Adelaide*  
**Christel Depienne** *Essen*  
**Raffaele Dubbioso** *Naples*  
**Silvana Franceschetti** *Milan*  
**Beatriz Gonzalez Giraldez** *Madrid*  
**Edouard Hirsch** *Strasbourg*  
**Akio Ikeda** *Kyoto*  
**Karl Martin Klein** *Calgary*  
**Laura Licchetta** *Bologna*  
**Fiore Manganelli** *Naples*  
**Roberto Michelucci** *Bologna*  
**Christopher Pearson** *Toronto*  
**Ingrid Scheffer** *Melbourne*  
**Josè Serratosa** *Madrid*  
**Isabel Silveira** *Porto*  
**Pasquale Striano** *Genoa*  
**Antonio Suppa** *Rome*  
**Marina Tijssen** *Groningen*  
**Shoji Tsuji** *Tokyo*  
**Arn van den Maagdenberg** *Leiden*  
**Anne-Fleur van Rootselaar** *Amsterdam*  
**Liana Veneziano** *Rome*  
**Federico Zara** *Genoa*

INTERNATIONAL WORKSHOP  
on FAMILIAL ADULT MYOCLONIC EPILEPSY



**FRIDAY 27TH MAY**

09.00 Registration of the participants

10.00 Welcome and introduction to the Workshop:

**Antonietta Coppola**

**Leonilda Bilo**

**Pasquale Striano**

**Fiore Manganelli**

**FIRST SESSION:**

**Myoclonic epilepsies**

Chairs: **Samuel Berkovic, Pasquale Striano**

10.30 Overview on myoclonic epilepsies

**Ingrid Scheffer**

11.00 Classification, clinical and neurophysiological  
features, prognosis

**Silvana Franceschetti**

11.30 Drug treatment of myoclonus

**Mar Carreno**

12.00 Lunch

## **SECOND SESSION:**

### **Electroclinical features and history of FAME/BAFME**

Chairs: **Ingrid Scheffer, Antonio Suppa**

13.30 From BAFME to FAME: history of the disease across the world  
**Samuel Berkovic**

13.50 Clinical findings, disease course and comorbidities  
**Akio Ikeda**

14.10 Neurophysiological findings  
**Marina Tijssen**

14.30 Discussion

15.00 Coffee break

## **THIRD SESSION:**

### **Diagnostic investigations**

Chairs: **Josè Serratosa, Karl Martin Klein**

15.30 Genetics of FAME: the history  
**Shoji Tsuji**

15.50 Genetics of FAME: the state of the art  
**Mark Corbett**

16.10 Neuropathology and Neuroimaging  
**Anne-Fleur van Rootselaar**

16.30 Treatment options  
**Edouard Hirsch**

16.50 Animal and cellular studies in (myoclonic) epilepsies  
**Arn van den Maagdenberg**

17.10 Discussion

17.30 **Round table: Patients meet the Experts**

## **SATURDAY 28TH MAY**

### **FOURTH SESSION:**

#### **FAME and Beyond: Pentanucleotide Related Disorders**

Chairs: **Christel Depienne, Federico Zara**

09.00 Repeat expansion disorders: an overview  
**Christopher Pearson**

09.20 Emerging pentanucleotide related disorder: the SCA37 model  
**Isabel Silveira**

09.40 Bioinformatic analysis for repeat expansion  
in short read sequencing data  
**Mark Bennett**

10.00 Coffee break

10.30 **Research projects**

Chairs: **Gianpiero Cavalleri, Roberto Michelucci**

FAME: structural and functional implications  
of aberrant sensory - motor excitability  
**Raffaele Dubbioso, Sirio Cocozza**

Perampanel as adjunctive treatment in FAME 2 patients  
**Antonietta Coppola**

Epidemiology of FAME in Italy. Unusual pentanucleotide repeat  
expansions: disease or polymorphism?  
**Laura Licchetta, Liana Veneziano, Francesco Brancati**

FAME 3 repeat Expansion structure revealed  
by complementary long - read technologies  
**Christel Depienne**

SAMD12 repeat expansion in a non - asian family  
with negative repeat - primed PCR  
**Karl Martin Klein**

Comparing progression and phenotypic severity  
in FAME vs. PME  
**Laura Canafoglia**

Long range PCR and targeted adaptive sampling  
of FAME loci with nanopore sequencing  
**Mark Corbett**

Previous misdiagnoses of patients with FAME  
**Betul Baykan**

Refining the phenotype in FAME 3:  
novel seizure types and clues to minimize misdiagnosis  
**Beatriz Gonzalez Giraldez**

13.00 Closing remarks

13.30 Light lunch





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### **General Information**

Congress venue  
HOTEL ROYAL CONTINENTAL NAPOLI  
VIA PARTENOPE, 38/44  
80121 Naples  
Meeting Room Mirabilis - First floor

### **Organizing Secretariat**



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