

## Paediatric Clinical Research Groups for precision medicine in DEE

June, **1-2**, 2023  
Scuola Superiore di Catania

programma

### 1 giugno 2023

- 09:30 Registrazione dei partecipanti
- 10:00 Welcome and introduction  
**Daniele Malfitana**,  
Presidente Scuola Superiore di Catania  
**Giuseppe Angilella**,  
Coordinatore Classe Scienze Sperimentali  
Scuola Superiore di Catania  
**Raffaele Falsaperla**,  
Responsabile Scientifico  
Modera: **Carlo Minetti**
- 10:15 Welcome address from a founder  
of the Italian Society of Pediatric Neurology  
**Lorenzo Pavone**
- 10:30 A brief history of Pediatric Neurology through  
the centuries and a look to the future of  
the Italian Society of Pediatric Neurology  
**Martino Ruggieri**
- 11:15 NF1 Gene Mutational Spectrum Analysis  
and Epilepsy  
**Manuela Lo Bianco**
- 11:45 TSC1 Vs TSC2 Genes Mutational Spectrum Analysis  
and Epilepsy  
**Roberta Leonardi**
- 12:15 Discussione
- 13:00 *Light Lunch*  
Modera: **Piero Pavone**
- 14:00 Precision Medicine in DEE  
**Pasquale Striano**
- 14:20 Network for Therapy in Rare Epilepsies (NETRE):  
Lessons From the Past 18 Years  
**Gerhard Kluger**
- 14:40 KCNQ2: genotype and anti seizure medicine  
**Roberta Criscione**
- 15:00 Special diet for special neonates  
**Laura Mauceri**
- 15:20 Chetogenic Diet in Neonatology: Useful or Not?  
**Raffaele Falsaperla**
- 16:00 Grand rounds (3-4 clinical cases)
- 18:00 Chiusura dei lavori della giornata

### 2 giugno 2023

- Modera: **Raffaele Falsaperla**
- 09:00 Clinical trial readiness for STXBP1-related disorders  
**Ganna Balagura**
- 10:00 Refractory epilepsy in patients  
with SLC35A2 brain somatic variants  
**Till Hartlieb**
- 11:00 Structural brain anomalies in patients  
with FOXG1 syndrome and Foxg1+/- mice  
**Milka Pringsheim**
- 12:00 Patch-clamp studies in KCNB1 and KCNQ2 DEE  
**Antonella Riva**
- 13:00 *Light Lunch*  
Modera: **Martino Ruggieri**
- 14:00 SYNGAP 1 from discovery to syndromology  
and an overlook to possible treatment  
**Celina von Stülpnagel**
- 15:00 Genetic neonatal screening in DEE  
**Federico Zara**
- 16:00 Grand rounds (3-4 clinical cases)
- 18:00 Meeting closure and greetings  
**Raffaele Falsaperla, Martino Ruggieri**