# TUBEROUS SCLEROSIS COMPLEX AS A MODEL DISORDER IN DEE: ADVANCING PATHOPHYSIOLOGICAL INSIGHTS AND THERAPEUTIC BREAKTHROUGHS IN PEDIATRIC NEUROLOGY AND PSICHIATRY

## Rome, November 6th - 7th, 2025

## Forma Spazi


**PROVIDER ECM N. 1293**
**ORGANIZING SECRETARIAT**

PTS Meetings & Events

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## UNDER THE PATRONAGE OF

Bambino Gesù
OSPEDALE PEDIATRICO

TOR VERGATA
UNIVERSITÀ DEGLI STUDI DI ROMA

ASSOCIAZIONE SCLEROSI TUBEROSA

ETSC European Tuberous Sclerosis Complex Association
*Making TSC matter*

LICE Lega Italiana Contro l'Epilessia

Sin
SOCIETÀ ITALIANA DI NEUROLOGIA

SINPIA
Società Italiana di Neuropsichiatria dell'Infanzia e dell'Adolescenza

## CONFERENCE VENUE

Forma Spazi
Via Cavour, 181
00184 Roma
www.formaspazi.it

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## ORGANIZING SECRETARIAT

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## REGISTRATION

Registration (free of charge) is required.
Please scan the QR code to register

or click **HERE**

## RATIONALE

Developmental and epileptic encephalopathies (DEEs) are a group of severe neurodevelopmental disorders characterized by early-onset epilepsy, cognitive and behavioral impairments, and complex treatment challenges. Among these, Tuberous Sclerosis Complex (TSC) stands out as a paradigmatic example. The central nervous system involvement in TSC includes a high prevalence of drug-resistant epilepsy, autism spectrum disorder (ASD), intellectual disability, and a range of neuropsychiatric comorbidities collectively termed TAND (TSC-associated neuropsychiatric disorders).

Over the past decade, TSC has emerged as a powerful model for understanding the pathophysiology of DEEs. This is largely due to the identification of the mTOR signaling pathway as a central molecular mechanism underlying the disease. Importantly, TSC is now one of the very few monogenic DEEs for which disease-specific treatments have been approved. This marks a significant shift from a time when no specific pharmacological options were available. Moreover, TSC has pioneered the concept of preventive treatment in epilepsy, with growing evidence supporting early intervention before the onset of clinical seizures—a strategy increasingly explored in other epileptic syndromes.

In addition, TSC represents a valuable model for syndromic autism, offering unique opportunities to investigate the neurobiological mechanisms of ASD and to explore targeted interventions in well-characterized genetic conditions.
Recent advances in neuroimaging, early EEG biomarkers, and genetic and molecular profiling have also enabled earlier diagnosis and offered promising perspectives for precision medicine approaches.

This two-day international meeting aims to bring together leading experts in pediatric neurology and psychiatry to share the latest insights on TSC as a model disorder in the broader field of DEEs.

By showcasing the most up-to-date clinical and research developments, the meeting intends to foster collaboration across disciplines and translate cutting-edge scientific discoveries into meaningful therapeutic strategies for TSC and related developmental epileptic encephalopathies.

## SCIENTIFIC COMMITTEE

*Romina Moavero* - Chair (Rome, IT)
*Pasquale Striano* (Genua, IT)
*Luigi Mazzone* (Rome, IT)
*Alessandra Voci* (Rome, IT)
*Giorgia Sforza* (Rome, IT)
*Massimiliano Valeriani* (Rome, IT)

## FACULTY

*Sam Amin* (Bristol, UK)
*Andrea Carai* (Rome, IT)
*Maria Roberta Cilio* (Bruxelles, BE)
*Raffaella Cusmai* (Rome, IT)
*Valentina De Giorgis* (Pavia, IT)
*Carlo Di Bonaventura* (Rome, IT)
*Carla Fladrowski* (Rome, IT)
*Stefano Francione* (Genoa, IT)
*Ann Jansen* (Bruxelles, BE)
*Sergiusz Jóźwiak* (Warsaw, PL)
*Katarzyna Kotulska* (Warsaw, PL)
*Pavel Kršek* (Prague, CZ)
*Francesca La Briola* (Milan, IT)
*Lieven Lagae* (Leuven, BE)
*Sara Matricardi* (Chieti, IT)
*Luigi Mazzone* (Rome, IT)
*Romina Moavero* (Rome, IT)
*Alessandra Morano* (Rome, IT)
*Rima Nabbout* (Paris, FR)
*Francesca Felicia Operto* (Catanzaro, IT)
*Angelo Russo* (Bologna, IT)
*Emilio Russo* (Catanzaro, IT)
*Susanne Schubert-Bast* (Frankfurt, DE)
*Pasquale Striano* (Genoa, IT)
*Irene Toldo* (Padua, IT)
*Federico Vigevano* (Rome, IT)
*Aglaia Vignoli* (Milan, IT)
*Alessandra Voci* (Rome, IT)

## ECM

**ID EVENTO N. 1293 - 461151** - Tipologia Formativa: RESIDENZIALE - RES.
All'evento sono stati assegnati n. 9 crediti ECM (9 ore formative).
L'evento è stato accreditato presso la Commissione Nazionale ECM ed è rivolto a:

Medici Specialisti in:

- **Neurofisiopatologia**
- **Neurologia**
- **Neurochirurgia**
- **Neuropsichiatria infantile**
- **Pediatria e Pediatri di libera scelta**
- **Psichiatria**

- **Tecnici di Neurofisiopatologia**
- **Dietisti**
- **Terapisti della neuro e psicomotricità dell'età evolutiva**
- **Logopedisti**
- **Psicoterapeuti e Psicologi**

Massimo 100 partecipanti

Al fine dell'attribuzione dei crediti formativi, i partecipanti dovranno garantire il 90% della propria presenza nonché compilare la scheda di valutazione e il questionario per la verifica dell'apprendimento.
Il percorso formativo dovrà essere completato entro 72 ore dal termine dell'evento (10 Novembre 2025). Completato il percorso e superato il test di apprendimento, i partecipanti potranno scaricare il certificato relativo al conseguimento dei crediti.
I partecipanti dovranno accedere alla user area con le stesse credenziali (user e password) utilizzate per effettuare l'iscrizione (il link di accesso alla user area sarà inviato a mezzo e-mail).

## OBIETTIVO FORMATIVO

Documentazione clinica. Percorsi clinico-assistenziali diagnostici e riabilitativi, profili di assistenza - profili di cura

## TIPOLOGIA DELL'EVENTO

Congresso/Simposio/Conferenza/Seminario

## PROGRAM - NOVEMBER 6th

| | |
|---|---|
| **13:15 - 15:25** | **SESSION 1**<br>Chairs: *C. Di Bonaventura - S. Jozwiak* |
| 13:15 - 13:30 | Welcome and introduction<br>*R. Moavero* |
| 13:30 - 13:55 | Neonatal EEG monitoring in TSC: catching the very first seizures<br>*M.R. Cilio* |
| 13:55 - 14:20 | The role of preventive treatment in the management of TSC<br>*K. Kotulska* |
| 14:20 - 14:45 | The challenge of transition in TSC and other rare epilepsies<br>*R. Nabbout* |
| 14:45 - 15:05 | Biomarkers of disease severity in TSC<br>*A. Morano* |
| 15:05 - 15:25 | Discussion |
| 15:25 - 15:45 | Coffee Break |
| **15:45 - 17:30** | **SESSION 2**<br>Chairs: *F. Vigevano - A. Vignoli* |
| 15:45 - 16:10 | The development of new targeted therapies for TSC<br>*E. Russo* |
| 16:10 - 16:30 | Cannabidiol in TSC related epilepsy<br>*I. Toldo* |
| 16:30 - 16:50 | Everolimus in TSC related epilepsy<br>*S. Jozwiak* |
| 16:50 - 17:10 | Is there room for alternative pharmacological treatments for LGS and other DEE in TSC?<br>*Susanne Schubert-Bast* |
| 17:10 - 17:30 | Discussion |
| **17:30 - 19:10** | **SESSION 3**<br>Chairs: *A. Carai - F. La Briola* |
| 17:30 - 17:50 | The role of epilepsy surgery in TSC related epilepsy<br>*P. Kršek* |
| 17:50 - 18:10 | Epilepsy surgery in “non focal” epilepsy<br>*S. Francione* |
| 18:10 - 18:50 | Other non-pharmacological treatments for refractory epilepsy in TSC<br>- Ketogenic diet<br>*V. De Giorgis*<br>- VNS<br>*A. Russo* |
| 18:50 - 19:10 | Discussion and end of the first day |

## PROGRAM - NOVEMBER 7th

| | |
|---|---|
| 09:00 - 11:00 | **SESSION 4**<br>Chairs: *L. Mazzone - S. Matricardi* |
| 09:00 - 09:25 | Early developmental evaluations as a marker of neurodevelopmental disorders<br>R. Moavero (Rome, Italy) |
| 09:25 - 09:50 | Early EEG findings as a biomarker of future neurodevelopment in TSC<br>*L. Lagae* |
| 09:50 - 10:15 | Modulation of Gut-brain axis as a potential therapeutic target in epilepsy and neurodevelopmental disorders<br>*P. Striano* |
| 10:15 - 10:40 | Targeted therapies in ASD and neuroinflammation in neurodevelopmental disorders<br>*L. Mazzone* |
| 10:40 - 11:00 | Discussion |
| 11:00 - 11:15 | Coffee Break |
| 11:15 - 13:00 | **SESSION 5**<br>Chairs: *R. Cusmai - F.F. Operto* |
| 11:15 - 11:30 | Italian Association - network<br>*C. Fladrowski - F. La Briola* |
| 11:30 - 11:55 | Quality of life in TSC<br>*S. Amin* |
| 11:55 - 12:20 | The development of questionnaires to explore patient related outcome measures in TSC<br>*A. Jansen* |
| 12:20 - 12:40 | Sleep in TSC<br>*A. Voci* |
| 12:40 - 13:00 | Discussion and closing remarks |

# NOTES