

Managing rare and complex epilepsies – part II

30 Ottobre 2026

Luogo: Genova, Villa Quartara - Provider: Gaslini Academy

Segreteria scientifica – Nobili, Vignoli, Mancardi, Prato

Session I – Opening

8.30-9.00 – participant registration

9.00-9.10 – Introduction and event opening

9.10-9.15 - welcome from the authorities

09.15- 09.45 Lecture: “Early differential impact of gene mutations on functional networks in Rare Disease”,
Michela Fagiolini

Session II – Epilepsy in Rare Diseases

Chairs: Mancardi , Bagnasco Vittorini

09.45 - 10.00 - The complexity of outcomes evaluations in Rare Diseases with epilepsy - Valentina De Giorgis

10.00 - 10.15 - Assessment of multidimensional outcomes beyond seizures: health-Related Quality of Life – Emanuele Bartolini

10.15 - 10.30 - Neurophysiological markers in Rare Disease with epilepsy – Matteo Cataldi

10.30 – 10.45 - Biochemical markers in Rare Disease with epilepsy – Enrico Tongiorgi

10.45-11.00 Discussion

Coffee break 11.00-11.30

Session III – Comorbidities in Rare Diseases

Chairs: De Grandis, Canavese

11.30-11.45 - Movement disorders in Rare Disease with epilepsy: overview and treatment – Gaetano Cantalupo

11.45 – 12.00 – Sleep disorders in Rare Disease with epilepsy – Ramona Cordani

12.15-12.30 – Psychiatric disorders in Rare Disease with epilepsy – Laura Siri

12.30-13.00 Discussion

13.00-14.00 Lunch

Gaslini Academy

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Sessione IV –Treatment

Chairs: Villani – Zara – Viri

14.00 – 14.15- Pharmacological treatment and the complexity of drug interactions-Emilio Russo

14.15- 14.30 - Emerging therapies: new frontiers– Aglaia Vignoli

14.30-14.45- Digital twins and Rare Disease – Pasquale Striano

14.45-16.00 clinical cases (uno genova, uno torino, uno Milano)

16.00-16.15 - Discussion

Closing remarks and take home message Nobili, Prato, Mancardi

16.15-16.30 - Discussion and summary of the meeting: proposal for a study on neurophysiological biomarkers

Corso Accreditato ECM per: MEDICO CHIRURGO: NEUROLOGIA; NEUROPSICHIATRIA INFANTILE; PEDIATRIA; NEUROFISIOPATOLOGIA; PEDIATRIA (PEDIATRI DI LIBERA SCELTA) - INFERMIERE PEDIATRICO - INFERMIERE - TECNICO DI NEUROFISIOPATOLOGIA

Faculty

Irene Bagnasco, Torino

Emanuele Bartolini, Pisa

Gaetano Cantalupo, Verona

Ramona Cordani, Genova

Matteo Cataldi, Genova

Valentina De Giorgis, Pavia

Elisa De Grandis, Genova

Michela Fagiolini, Pisa

Maria Margherita Mancardi, Genova

Emilio Russo, Catanzaro

Pasquale Striano, Genova

Laura Siri, Genova

Enrico Tongiorgi, Trieste

Roberta Vittorini, Torino

Aglaia Vignoli, Milano

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Scientific Rationale

Rare and complex epilepsies represent a significant challenge in pediatrics. These forms of epilepsy are often associated with genetic syndromes and rare diseases, with an incidence of fewer than 5 cases per 10,000 people. In children, these conditions often manifest with drug-resistant seizures and complex neurological and cognitive disabilities, significantly impacting the quality of life for both patients and their families.

In recent years, advances in medical genetics have allowed for a better understanding of the molecular bases of many forms of pediatric epilepsy. The identification of mutations in specific genes has led to new diagnostic and therapeutic approaches, paving the way for precision medicine. This approach enables clinical management to be directed not only based on the clinical and electroencephalographic characteristics of the patient but also according to the specific genetic profile, thereby improving the effectiveness of therapies and optimizing the child's quality of life.

The objective of the conference is to deepen knowledge about rare and complex epilepsies in childhood, with a specific focus on genetic forms and new targeted treatments. Real clinical cases will be discussed, and diagnostic pathways will be analyzed, including next-generation sequencing (NGS) technologies, the interpretation of genetic data, and their impact on therapeutic decisions. Additionally, the multidisciplinary approach necessary to manage the psychiatric, cognitive, and behavioral comorbidities that often accompany these forms of epilepsy will be evaluated.

Through this event, participants will receive scientific and practical updates to guide them in the integrated and personalized management of rare and complex epilepsies in pediatric patients. The ultimate goal is to contribute to comprehensive patient care and enhance future therapeutic prospects.

