

"Explo...RARE: Bridges between research and therapy"

Bari February 20-21, 2026 Mövenpick Hotel

Scientific Director: Albina Tummolo

Scientific Secretariat: Donatella De Giovanni

FACULTY

Accadia	Maria	Panico Foundation – Tricase
Annicchiarico	Giuseppina	Co.Re.Ma.R. - Puglia Region
Ardissone	Anna	IRCCS C. Besta Neurological Institute – Milan
Aricò	Maurizio	Bari Polyclinic
Barile	Maria	University of Bari Aldo Moro
Beheshti	Afshin	University of Pittsburgh
Belviso	Benny Danilo	CNR – National Research Council
Bianco	Mariana	Stella Rare Association - A.Ma.Re Puglia
Burlina	Alberto	University of Padua
Caggianelli	Gabriele	Sapienza University of Rome
Cardinale	Fabio	Giovanni XXIII Pediatric Hospital - Bari
Carella	Rosa	Giovanni XXIII Pediatric Hospital - Bari
Cavallo	Luciano	Giovanni XXIII Pediatric Hospital - Bari
D'Amore	Simona	University of Bari Aldo Moro
Dardis	Andrea	ASUFC – Central Friuli
De Giovanni	Donatella	Bari Polyclinic
De Grassi	Anna	University of Bari Aldo Moro
Fecarotta	Simona	University of Naples Federico II
Gentile	Mattia	ASL Bari
Manti	Filippo	Sapienza University of Rome
Mazzia	Francesca	University of Bari Aldo Moro
Melpignano	Livio	Giovanni XXIII Pediatric Hospital - Bari
Moliterni	Paola	Bari Polyclinic
Noto	Davide	University of Palermo
Orsini	Paola	ASL Bari
Papa	Riccardo	Gaslini Institute - Genoa
Paterno	Giulia	Giovanni XXIII Pediatric Hospital - Bari
Pession	Andrea	University of Bologna – IRCCS Sant’Orsola
Pierri	Ciro	University of Bari Aldo Moro
Piovani	Paola	ASUFC – Friuli Centrale
Simonetti	Simonetta	Bari Polyclinic
Spada	Marco	University of Turin
Squicciarini	Rosella	ASL Bari
Srinivasulu	Vishnuvardhan Pogunulu	Vizzhy healthcare - Middletown
Tandoi	Francesco	University of Bari Aldo Moro
Trabacca	Antonio	IRCCS Eugenio Medea - Brindisi
Vanzi	Valentina	UniCamillus University – Rome
Zuvadelli	Juri	ASST Santi Paolo e Carlo – Milan

PRE-CONGRESS SESSIONS (Parallel)

February 20, 2026 | 9:00 AM – 1:00 PM

Dietetics

- **Practical applications of the diet for inherited metabolic diseases**
 - *Leads:* Rosa Carella, Giulia Paterno, Juri Zuvadelli
- **Presentation of clinical cases and group activities on diet therapy in:**
 - Phenylketonuria
 - Glycogenosis type IA
 - Inherited metabolic diseases treated with ketogenic diet

Nursing

- **The research nurse: competencies, methodology, and activities**
 - *Speakers:* Gabriele Caggianelli, Paola Piovani, Valentina Vanzi
- **From research to evidence-based practice:** knowledge, skills, and attitudes
- **From evidence to the improvement of care practice:** implementation methods and tools
- **Research and nursing:** Roles and paths for nurses
- **Discussion on presented topics**

CONGRESS PROGRAM

Friday, February 20, 2026

2:00 PM – Welcome and introduction to the congress

I SESSION: BASE AND TRANSLATIONAL RESEARCH *Moderators: Benny Danilo Belviso, Francesca Mazzia*

- **2:20 PM** – The role of B-group vitamins in cellular energy balance and gene expression regulation – *Maria Barile*
- **2:40 PM** – Drug repurposing in rare diseases: a continuously evolving resource – *Ciro Pieri*
- **3:00 PM** – Energy Collapse: Mitochondrial Dysfunction as the Engine of Long COVID Pathophysiology – *Afshin Beheshti*
- **3:20 PM** – Why Two Hearts With the Same Disease Do Not Heal the Same Way? Non-Ergodicity, Biological Memory, and the Limits of Risk-Based Cardiology – *Vishnuvardhan Pogumulu Srinivasulu*
- **3:40 PM** – Transversal procedures for the discovery of new genes associated with mitochondrial pathologies: two case studies – *Anna De Grassi*
- **4:00 PM** – Discussion
- **4:15 PM** – Coffee break

II SESSION: PREVENTION AND EARLY DIAGNOSIS IN RARE DISEASES Moderators: *Maurizio Aricò, Andrea Pession*

- **4:50 PM** – Genomic screening: the experience of the Puglia region – *Mattia Gentile*
- **5:10 PM** – Diagnoses from extended and super-extended screening in nearly 10 years – *Simonetta Simonetti*
- **5:30 PM** – How biochemical and genomic screening coexist in our region – *Donatella De Giovanni, Paola Orsini*
- **6:00 PM** – Can traditional laboratory tests and omic techniques coexist? – *Andrea Gardini*
- **6:20 PM** – Discussion
- **6:45 PM** – Closing of the first day

Saturday, February 21, 2026

III SESSION: CURRENT THERAPIES Moderators: *Fabio Cardinale, Luciano Cavallo*

- **8:30 AM** – Mitochondrial encephalopathies: focus on pediatric phenotypes and potentially treatable forms – *Anna Ardisson*
- **8:50 AM** – Liver transplant in inherited metabolic diseases: the Italian experience – *Francesco Tandoi*
- **9:10 AM** – Anti-inflammatory drugs in the therapy of genetic diseases: Fibrodysplasia Ossificans Progressiva – *Riccardo Papa*
- **9:30 AM** – New therapeutic strategies for lysosomal diseases: from Gaucher disease to alpha-mannosidosis – *Simona D'Amore*
- **9:50 AM** – The use of artificial intelligence for predicting therapeutic response in rare diseases – *Davide Noto*
- **10:10 AM** – Discussion
- **10:30 AM** – Coffee break

IV SESSION: THE FUTURE Moderators: *Maria Accadia, Antonio Trabacca*

- **11:00 AM** – Phenylketonuria: how the combination of different approaches will change the face of the disease – *Alberto Burlina*
- **11:20 AM** – Evolution of phenotypes and biomarkers in Fabry disease: between classic and late-onset phenotype – *Marco Spada*
- **11:40 AM** – New standard of care in Mucopolysaccharidosis: therapeutic perspectives in 2026 – *Simona Fecarotta*
- **12:00 PM** – Autism in the pediatric setting: follow-up protocols for a multidisciplinary approach – *Filippo Manti*

V SESSION: HOT TOPICS IN METABOLIC-GENETIC DISEASE MANAGEMENT *Moderators: Livio Melpignano, Rosella Squicciarini*

- **12:20 PM** – Regional Governance and the importance of the network between centers – *Giuseppina Annicchiarico*
- **12:40 PM** – The thin line between curative and palliative medicine in children with rare diseases – *Paola Moliterni*
- **1:00 PM** – PROMs from the patient's perspective – *Marianna Bianco*
- **1:20 PM** – Discussion and closing