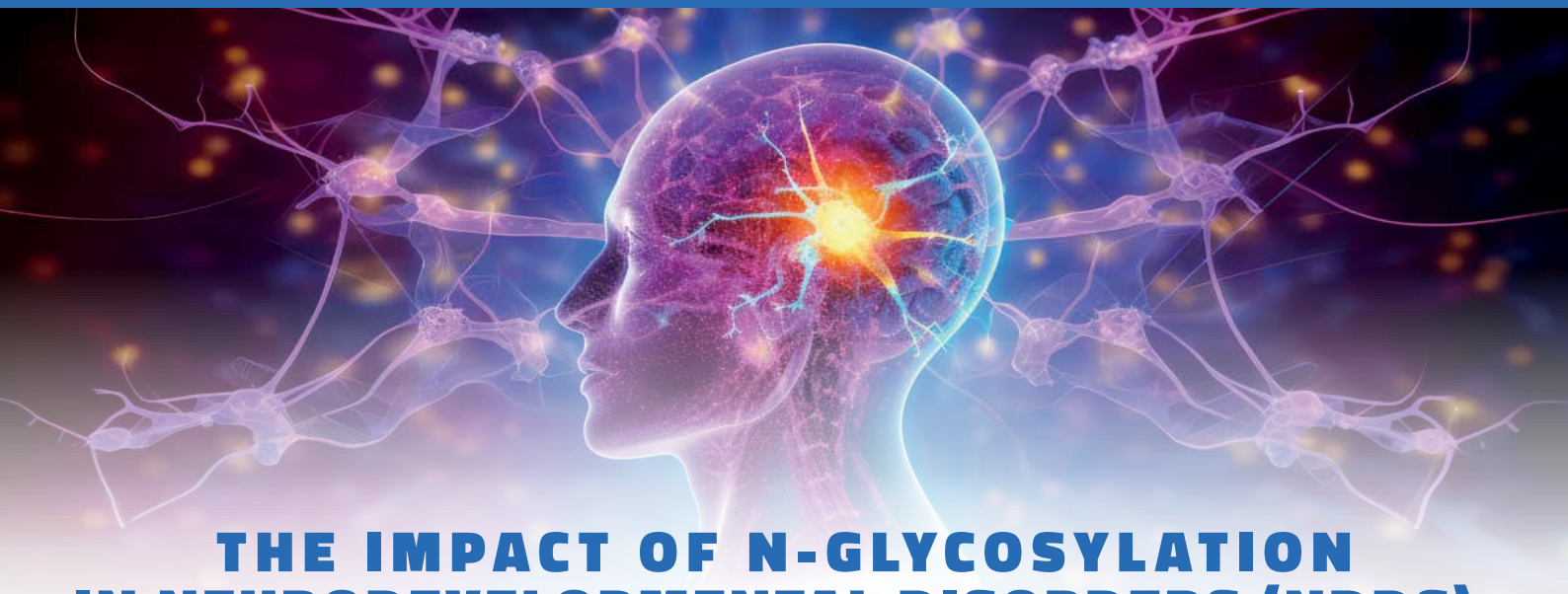
Finanziato dall'Unione europea NextGenerationEU
Ministero dell'Università e della Ricerca
Italiadomani PIANO NAZIONALE DI RIPRESA E RESILIENZA
Università di Catania



# THE IMPACT OF N-GLYCOSYLATION IN NEURODEVELOPMENTAL DISORDERS (NDDS): FROM DIAGNOSIS TO TARGET THERAPIES

## PRIN 2022 RESEARCH PROJECT *NGLYNEU* DISSEMINATION

### JANUARY 16-17, 2026 - CATANIA, ITALY

Glycosylation is the most frequent post-translational modification of proteins and is essential for the development and function of the central nervous system (CNS). Congenital Disorders of Glycosylation (CDG) are a group of genetic CNS diseases caused by defects in the synthesis and attachment of glycans to proteins and lipids.

CDG belong to the broader group of neurodevelopmental disorders (NDDs), whose genetic basis remains largely unknown. The NGLYNEU project aims to improve current knowledge on the impact of genetic variants involved in glycosylation in neurodevelopmental diseases.

This will support identification of new potential disease-causing genes and the characterization of previously unrecognized neurodevelopmental disorders resulting from glycosylation defects. The project's outcomes will contribute to broadening the boundaries of genetic CNS disorders and to identifying new molecular targets for their treatment.

## OPENING CEREMONY - FRIDAY 16TH JANUARY 2026

**Venue: Aula Magna – Monastero dei Benedettini – Piazza Dante 32, Catania**

**14.30** PARTICIPANTS' REGISTRATION

**15.30** WELCOME AND GENERAL INFORMATION ON THE SCIENTIFIC PROGRAM
**Rita BARONE** *University of Catania, Italy*

**15.40** INSTITUTIONAL GREETINGS
Chairs: **Domenico GAROZZO** *CNR Catania, Italy*
**Elena BARBIERI** *University of Urbino, Italy*

**16.00** OPENING KEYNOTE
Congenital Disorders of Glycosylation (CDG): Quo Vadis?
**Eva MORAVA** *Mount Sinai Hospital New York, USA*

KEYNOTES

**16.30** Tourette Syndrome: a paradigm for NDD
**Renata RIZZO** *University of Catania, Italy*

**17.00** IGF1 and IGF1 receptor glycosylation: emerging biomarkers of N-glycosylation defects
**Giosuè ANNIBALINI** *University of Urbino, Italy*

## SATURDAY 17TH JANUARY 2026

Venue: Aula Magna Torre Biologica – Via Santa Sofia 98, Catania

**8.00** PARTICIPANTS' REGISTRATION

### SESSION NEXT-GENERATION SEQUENCING (NGS) AND MULTI-OMIC ANALYSES IN UNDIAGNOSED PATIENTS WITH NDDS

Chairs: **Renata RIZZO** *University of Catania, Italy*
**Corrado ROMANO** *University of Catania, Italy*

**9.00** OPENING KEYNOTE

Cracking the Code of CDG: Multi-Omic Models for Mechanism and Treatment
**Tamas KOZICZ** *Mount Sinai Hospital New York, USA*

**9.30** Targeted NGS analyses in the suspect of N-glycosylation disorders
**Amelia MORRONE** *Meyer Children's Hospital Firenze, University of Florence, Italy*

**9.50** MAN2A2 mutation in cognitive disability
**Simone TRECCARICHI** *IRCCS Oasi Research Institute, Troina, Italy*

KEYNOTES

**10.10** PRIN-2022 NGLYNEU project: preliminary results
**Rita BARONE** *University of Catania, Italy*

**10.40** Glycomic signatures of N-glycosylation disorders
**Luisella STURIALE** *CNR Catania, Italy*

Q&A

**11.20** COFFEE BREAK

### SESSION CDG: NOVEL PHENOTYPES, MODELS AND THERAPY DEVELOPMENT

Chairs: **Rita BARONE** *University of Catania, Italy*
**Caterina SPEZIA** *CDG - Italia Family Association*

**11.45** KEYNOTE

New treatment options in CDG, from bench to bedside
**Eva MORAVA** *Mount Sinai Hospital New York, USA*

**12.15** NDDs phenotypes in N-glycosylation disorders
**Lara CIRNIGLIARO** *University of Catania, Italy*

**12.35** Epileptic phenotypes in N-glycosylation disorders
**Fabio PETTINATO** *University of Catania, Italy*

**12.55** Peripheral Blood Mononuclear Cells (PBMC) analyses for metabolomic evaluation in PMM2-CDG
**Angela Maria AMORINI, Giuseppe LAZZARINO** *University of Catania, Italy*

**13.15** CDG - Italia Family Association
**Caterina SPEZIA** *Trapani, Italy*

Q&A

**13.30** LIGHT LUNCH

## SESSION EFFECTS OF GLYCO-GENES MUTATION AND INACTIVATION ON CELLULAR PROTEIN GLYCOSYLATION

Chairs: **Cettina MELI** *AOUP Catania, Italy*
**Marco FICHERA** *University of Catania, Italy*

**14.30** Molecular characterization of VPS13B gene:
assessing the impact of Cohen syndrome mutations on protein function
**Fabiana FANELLI** *University of Urbino, Italy*

**14.50** Novel perspectives in the treatment of Cohen syndrome
**Martina RANDAZZO** *University of Catania, Italy*

**15.10** Serum N-glycosylation in patients with Cohen syndrome
**Angelo PALMIGIANO** *CNR Catania, Italy*
**Angela MESSINA** *CNR Catania, Italy*

**Q&A**

**15.30 CONCLUDING REMARKS**

**18.30 -19.00 WELCOME MEETING WITH CDG - ITALIA FAMILY ASSOCIATION**

Novel clinical trials in CDG
**Eva MORAVA, Tamas KOZICZ** *Mount Sinai Hospital New York, USA*

**19.45 STAY TOGETHER AND DINNER**

## SCIENTIFIC COMMITTEE

Rita BARONE — Uni ct MEDICINA CLINICA E SPERIMENTALE

Luisella STURIALE — CNR iPCB Institute of Polymers, Composites and Biomaterials

Giosuè ANNIBALINI — 1506 UNIVERSITÀ DEGLI STUDI DI URBINO CARLO BO

## ORGANIZING COMMITTEE

Lara CIRNIGLIARO Fabio PETTINATO Martina RANDAZZO Roberta SALTARELLI Caterina SPEZIA

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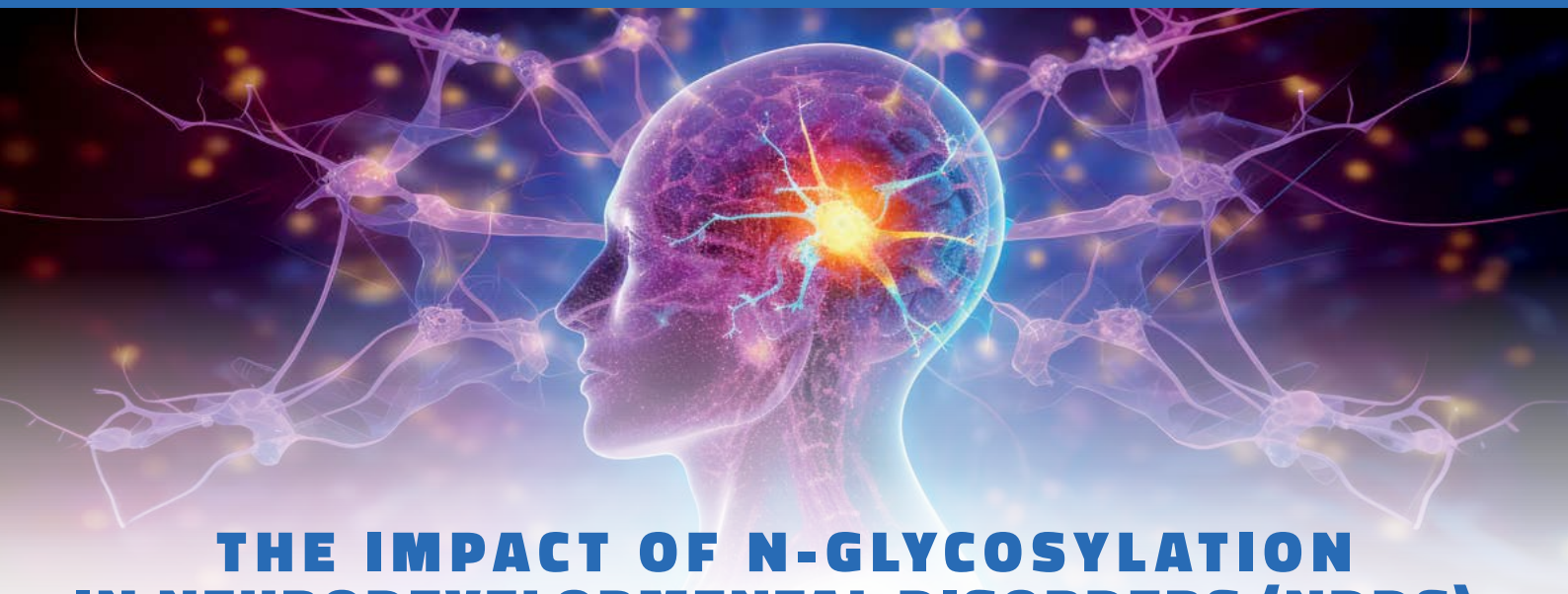

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### JANUARY 16-17, 2026 - CATANIA, ITALY

### FACULTY

| | |
|---|---|
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| **STURIALE Luisella** | *IPCB CNR Catania* |
| **TRECCARICHI Simone** | *IRCCS Oasi Troina (EN)* |