The impact of N-Glycosylation in NeuroDevelopmental Disorders (NDDs): from diagnosis to target therapies

January 16-17, 2026

Catania - Italy

Rationale

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 implementation of the NGLYNEU project will expand current knowledge on the functional impact of genetic variants involved in glycosylation in neurodevelopmental diseases. This will lead to the identification of new potential disease-causing genes and to 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.

PROGRAM

Opening Ceremony: **Friday – 16th Jan 2026**

Venue: Aula Magna – Monastero dei Benedettini - **Piazza Dante 32 - 95124 Catania**

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 Jan 2026

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

08.00 **Participants' registration**

Chairs: *Renata Rizzo (University of Catania, Italy); Corrado Romano (University of Catania, Italy)*

Session – **Next-Generation Sequencing (NGS) AND Multi-OMIC ANALYSES IN UNDIAGNOSED PATIENTS WITH NDDs**

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)

09:50 **MAN2A2 mutation in cognitive disability**

Simone Treccarichi (IRCCS Oasi Research Institute, Troina,Italy)

Keynotes

10:00 **Bi-allelic versus mono-allelic mutation of glycogenes in patients with NDDs**

Rita Barone (University of Catania,Italy)

10.30 **Glycomic signature of N-glycosylation disorders**

Luisella Sturiale (CNR Catania,Italy)

Q&A

11.10 Coffee-Break

Chairs: *R Barone (University of Catania, Italy); C. Spezia (CDG-Italia family association)*

Session – **CDG: NOVEL PHENOTYPES, MODELS AND THERAPY DEVELOPMENT**

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

Chairs Cettina Meli *(University of Catania, Italy);* Marco Fichera *(University of Catania, Italy)*

Session — **EFFECTS OF GLYCO-GENES MUTATION AND INACTIVATION ON CELLULAR PROTEIN GLYCOSYLATION**

15.00 **Molecular characterization of VPS13B gene: assessing the impact of Cohen syndrome mutations on protein function**

Fabiana Fanelli (University of Urbino,Italy)

15.20 **Novel perspectives in the treatment of Cohen syndrome**

Martina Randazzo (University of Catania,Italy)

15.40 **Serum IgG glycosylation in patients with Cohen syndrome**

Angelo Palmigiano (*CNR Catania,Italy*), *Angela Messina* (*CNR Catania,Italy*)

Q&A

16:15 Concluding Remarks

18:30 -19:00 **Welcome Meeting with CDG-Italia family association**

Novel clinical trials in CDG

Eva Morava and Tamas Kozicz (Mount Sinai Hospital New York)

19.45 **Stay together and Dinner**

Commentato [DG1]: taglierei qualche spazio per far stare tutto nella pagina

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