Maria Monticelli

Associate Professor · Biochemistry

Saint Camillus International University of Health Sciences

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Education

University of Napoli Federico II — *Napoli, Italy*

PhD Biology — *Apr 2022*

- Thesis: *The role of missense mutations in diseases: two sides of a coin*
- Supervisor: Prof. Maria Vittoria Cubellis

University of Napoli Federico II — *Napoli, Italy*

MSc Biology, *cum laude* — *Mar 2018*

- Thesis: *A look inside the molecular pathways of PMM2-CDG: comparison between phosphomannomutases 1 and 2*
- Research laboratory: Institute of Biomolecular Chemistry, CNR
- Supervisor: Prof. Maria Vittoria Cubellis; Co-supervisor: Dr. Giusepina Andreotti

University of Napoli Federico II — *Napoli, Italy*

BSc Biological Sciences — *Oct 2014*

- Thesis: *Use of Thermal Shift Assay to identify ligands for proteins of medical relevance*
- Research laboratory: Institute of Biomolecular Chemistry, CNR
- Supervisor: Prof. Maria Vittoria Cubellis; Co-supervisor: Dr. Giusepina Andreotti

Professional Experience

Jul 2026 - current	**Associate Professor of Biochemistry**, Saint Camillus International University of Health Sciences, Rome, Italy
Nov 2024 - Apr 2026	**Postdoctoral Research Fellowship**, University of Napoli Federico II, Italy
Oct 2023 - Oct 2024	**Postdoctoral Research Fellowship**, Institute of Biomolecular Chemistry, Pozzuoli, Italy
Apr 2022 - Mar 2023	**Research Fellowship**, University of Campania L. Vanvitelli, Caserta, Italy
Jun 2016 - Oct 2016	**Telethon Scholarship**, University of Napoli Federico II, Italy
Aug 2015 - Dec 2015	**1st Liliana Initiation Scholarship**, CEDOC, University NOVA of Lisboa, Portugal

Publications

31 publications, h-index 16, 720 citations – source: Scopus

Prominent authorship position – first, last or corresponding: 17 publications

(* corresponding author; † co-first authorship)

Gallego D., Andreotti G., **Monticelli M.**, Paris D., Martín-Martínez A., Gámez A., Serrano M., Córdoba-Caballero J., Seoane P., Ranea JAG., Pérez B. Proteo-metabolomic profiling of PMM2-CDG reveals dysregulation of retinoic acid synthesis, myo-Inositol, and the hexosamine pathway. J. Inherit. Metab. Dis. Accepted for publication.

Sodano F., **Monticelli M.***, Hay Mele B., Rolando B., Lazzarato L., De Simone A., Andrisano V., Paris D., Rimoli M.G., Cubellis M.V., Andreotti G.* Lipo-Glc-1,6-P_2: a bioprecursor prodrug for phosphomannomutase-2 Congenital Disorder of Glycosylation. IUBMB Life. 2026 Apr;78(4):e70101.

De Filippis G.M.†, **Monticelli M.†**, Hay Mele B., Calabrò V. Missense variants in nutrition-related genes: a computational study. Int. J. Mol. Sci. 2025, 26, 9619.

Hay Mele B., Rossetti F., Andreotti G., Cubellis M.V., Guerriero S., **Monticelli M.** Bioinformatics driven multi-factorial insight into α-galactosidase mutations. Int. J. Mol. Sci. 2025, 26, 5802.

Hay Mele B., Bovenzi J., Andreotti G., Cubellis M.V., **Monticelli M.** In silico analysis of phosphomannomutase-2 dimer interface stability and heterodimerization with phosphomannomutase-1. Molecules 2025, 30, 2599.

De Filippis G.M., **Monticelli M.**, Pollice A., Angrisano T., Hay Mele B., Calabrò V. Computational strategies in nutrigenetics: constructing a reference dataset of nutrition-associated genetic polymorphisms. J Biomed Inform. 2025;2025.104845.

Monticelli M., Paris D., Monti M.C., Morretta E., Pakanova Z., Nemcovic M., Kodrikova R., Cubellis M.V., Andreotti G. Beneficial effects of Glc-1,6-P_2 modulation on mutant phosphomannomutase-2. Biochim Biophys Acta Mol Cell Res. 2025;1872(5):119948.

Monticelli M., Hay Mele B., Wright D.M., Guerriero S., Andreotti G., Cubellis M.V. Exploring ligand interactions with human phosphomannomutases using recombinant bacterial thermal shift assay and biochemical validation. Biochimie. 2024 Jul;222:123-131.

Hay Mele B., Rossetti F., Cubellis M.V., **Monticelli M.***, Andreotti G. Drug Repurposing and Lysosomal Storage Disorders: A Trick to Treat. Genes (Basel). 2024 Feb 25;15(3):290.

Monticelli M., Wright M.D., Cubellis M.V., Andreotti G. ReBaTSA: A simplified CeTSA protocol for studying recombinant mutant proteins in bacterial extracts. Biochim Biophys Acta Gen Subj. 2024 Feb;1868(2):130526

Monticelli M., D'Onofrio T., Jaeken J., Morava E., Andreotti G., Cubellis M.V. Congenital disorders of glycosylation: narration of a story through its patents. Orphanet J Rare Dis. 2023 Aug 29;18(1):247.

Hay Mele B., **Monticelli M.**, Leone S., Bastoni D., Barosa B., Cascone M., Migliaccio F. Montemagno F., Ricciardelli A., Tonietti L., Rotundi A., Cordone A., Giovannelli D. Oxidoreductases and metal cofactors in the functioning of Earth. Essays Biochem. 2023 Aug 11;67(4):653-670.

Iacobucci I., Hay Mele B., Cozzolino F., Monaco V., Cimmaruta C., Monti M., Andreotti G., **Monticelli M.** Enzyme replacement therapy for Fabry disease: possible strategies to improve its efficacy. Int J Mol Sci. 2023 Feb 25;24(5):4548.

Monticelli M., Hay Mele B., Allocca M., Liguori L., Lukas J., Monti M.C., Morretta E., Cubellis M.V., Andreotti G. Curcumin has beneficial effects on lysosomal alpha-galactosidase: potential implications for the cure of Fabry Disease. Int J Mol Sci. 2023 Jan 6;24(2):1095.

Vignogna R.C., Allocca M., **Monticelli M.**, Norris J.W., Steet R., Perlstein E.O., Andreotti G., Lang G.I. Evolutionary rescue of phosphomannomutase deficiency in yeast models of human disease. Elife. 2022 Oct 10;11:e79346.

Monticelli M., Francisco R., Brasil S., Marques-da-Silva D., Rijoff T., Pascoal C., Jaeken J., Videira P.A., dos Reis Ferreira V. Stakeholders' views on drug development: the congenital disorders of glycosylation community perspective. Orphanet J Rare Dis. 2022 Jul 30;17(1):303.

Pignata L., Cecere F., Verma A., Hay Mele B., **Monticelli M.**, Acurzio B., Giaccari C., Sparago A., Hernandez Mora J.R., Monteagudo-Sánchez A., Esteller M., Pereda A., Tenorio-Castano J., Palumbo O., Carella M., Prontera P., Piscopo C., Accadia M., Lapunzina P., Cubellis M.V., de Nanclares G.P., Monk D., Riccio A., Cerrato F. Novel genetic variants of KHDC3L and other members of the subcortical maternal complex associated with Beckwith-Wiedemann syndrome or Pseudohypoparathyroidism 1B and multi-locus imprinting disturbances. Clin Epigenetics. 2022 May 28;14(1):71.

Monticelli M., Liguori L., Allocca M., Bosso A., Andreotti G., Lukas J., Monti M.C., Morretta E., Cubellis M.V., Hay Mele B. Drug repositioning for Fabry Disease: acetylsalicylic acid potentiates the stabilization of lysosomal alpha-galactosidase by pharmacological chaperones. Int J Mol Sci. 2022 May 4;23(9):5105.

Pantoom S., Hules L., Schöll C., Petrosyan A., **Monticelli M.**, Pospech J., Cubellis M.V., Hermann A., Lukas J. Mechanistic insight into the mode of action of acid-glucosidase enhancer ambroxol. Int J Mol Sci. 2022 Mar 24;23(7):3536.

Bosso A., Gaglione R., Di Girolamo R., Veldhuizen E. J.A., García-Vello P., Fusco S., Cafaro V., **Monticelli M.**, Culurciello R., Notomista E., Arciello A., Pizzo E. Human cryptic host defence peptide GVF27 exhibits anti-infective properties against biofilm forming members of the Burkholderia cepacia complex. Pharmaceuticals (Basel). 2022 Feb 21;15(2):260.

Monticelli M., Hay Mele B., Andreotti G., Cubellis M.V., Riccio G. Why does SARS-CoV-2 hit in different ways? Host genetic factors can influence the acquisition or the course of COVID-19. Eur J Med Genet 2021 Jun;64(6):104227.

Monticelli M., Hay Mele B., Benetti E., Fallerini C., Baldassarri M., Furini S., Frullanti E., Mari F., GEN-COVID Multicenter Study, Andreotti G., Cubellis M.V., Renieri A.. Protective Role of a TMPRSS2 Variant on Severe COVID-19 Outcome in Young Males and Elderly Women. Genes 2021, 12(4), 596.

Cubellis M. V., Pignata L., Verma A., Sparago A., Del Prete R., **Monticelli M.**, Calzari L., Antona V., Melis, D., Tenconi R., Russo S., Cerrato F., Riccio A. Loss-of-function maternal-effect mutations of PADI6 are associated with familial and sporadic Beckwith-Wiedemann syndrome with multi-locus imprinting disturbance. Clin Epigenetics. 2020 Sep 14;12(1):139.

Liguori L., **Monticelli M.**, Allocca M., Hay Mele B., Lukas J., Cubellis M.V., Andreotti G. Pharmacological Chaperones: A Therapeutic Approach for Diseases Caused by Destabilizing Missense Mutations. Int J Mol Sci. 2020 Jan 13;21(2):489.

Liguori L., **Monticelli M.**, Allocca M., Cubellis M.V., Hay Mele B. Bioinformatics tools for marine biotechnology: a practical tutorial with a metagenomic approach. BMC Bioinformatics. 2020 Aug 21;21(Suppl 10):348.

Monticelli M., Liguori L., Allocca M., Andreotti G., Cubellis M.V. β-Glucose-1,6-bisphosphate stabilizes pathological phophomannomutase2 mutants in vitro and represents a lead compound to develop pharmacological chaperones for the most common disorder of glycosylation, PMM2-CDG. Int J Mol Sci. 2019 Aug 26;20(17):4164.

Citro V., Cimmaruta C, **Monticelli M.**, Riccio G., Hay Mele B., Cubellis M.V., Andreotti G. The analysis of variants in the general population reveals that PMM2 is extremely tolerant to missense mutations and that diagnosis of PMM2 -CDG can benefit from the identification of modifiers. Int J Mol Sci. 2018 Jul 30;19(8):2218.

Cimmaruta C., Liguori L., **Monticelli M.**, Andreotti G., Citro V. E-Learning for rare diseases: an example using Fabry Disease. Int J Mol Sci. 2017 Sep 24;18(10):2049.

Marques-da-Silva D., Dos Reis Ferreira V., **Monticelli M.**, Janeiro P., Videira P.A., Witters P., Jaeken J., Cassiman D. Liver involvement in congenital disorders of glycosylation (CDG). A systematic review of the literature. J Inherit Metab Dis. 2017 Mar;40(2):195-207.

Monticelli M., Ferro T., Dos Res Ferreira V., Jaeken J., Videira P.A. Immunological aspects of congenital disorders of glycosylation (CDG): a review. J Inherit Metab Dis. 2016 Nov;39(6):765-780.

Andreotti G., **Monticelli M.**, Cubellis M.V. Looking for protein stabilising drugs with thermal shift assay. Drug Test Anal. 2015 Sep;7(9):831-4.

Awards, Fellowships, and Grants

2026	**First call for proposals for FEBS Supported Society education events in 2026**, FEBS **Lead Applicant**, on behalf of the Italian Society of Biochemistry and Molecular Biology (SIB). Funding request to support the 1st SIB Young Meeting.	*€2,000*
2025	**Travel Grant, 63rd Italian SIB Meeting**, Italian Society of Biochemistry and Molecular Biology	
2024	**Travel Grant, 1st Anglo-Italian Chemical Biology Bilateral Meeting**, Italian Chemical Society	
2023	**Young Researcher Award**, Institute of Biomolecular Chemistry	

Presentations

Invited Talks

Apr 2024. *Biochemical features of phosphomannomutase-2 associated congenital disorder of glycosylation, PMM2-CDG*. Joint IBBC - EMBL seminars, Monterotondo, Italy.

May 2021. *The challenges and solutions faced throughout CDG research and drug development: results from an electronic questionnaire made possible by CDG families and professionals*. Invited talk: 5th World Conference on CDG, Virtual Conference.

Contributed Presentations

Dec 2025. *PROPHECY-GlycoRARE: small-molecule UGGT modulators as a therapeutic approach for misfolding diseases*. 8th Institute of Biomolecular Chemistry Meeting, Napoli, Italy.

Oct 2025. *Phosphomannomutases in the mirror: from structural and biochemical divergence to small molecule therapies in PMM2-CDG*. Giornate Scientifiche di Dipartimento, DSCTM-CNR, Bologna, Italy.

Dec 2024. *Beneficial effects of Glc-1,6-P_2 modulation on mutant PMM2*. 1st Anglo Italian Chemical Biology Bilateral Meeting, Perugia, Italy.

Dec 2023. *Small molecules and missense mutations*. 7th Institute of Biomolecular Chemistry Meeting, Padova, Italy.

Nov 2023. *Raw protein extracts to determine the protein-ligand binding: a tool for drug discovery*. 4th Memorial Workshop Maria Ciaramella, Napoli, Italy.

Nov 2020. *Analysis of whole exome sequencing and protein modelling: the lesson from cyclin B*. Bioinformatics and Computational Biology Conference, Virtual Conference.

Nov 2018. *Passenger mutations as a target for the personalised therapy of cancer*. Bioinformatics and Computational Biology Conference, Napoli, Italy.

Poster Presentations

Sep 2025. *From Bystander to Target: Phosphomannomutase-1 as a Therapeutic Node in Phosphomannomutase-2 Congenital Disorder of Glycosylation*. 63rd Congress of the Italian Society of Biochemistry and Molecular Biology, Palermo, Italy.

Dec 2024. *Characterising a-GAL Mutants: From Misfolding to Rescue*. 1st Anglo Italian Chemical Biology Bilateral Meeting, Perugia, Italy.

May 2023. *Raw protein extracts to determine the protein-ligand binding: a tool for drug discovery*. BioProSys Joint Meeting, Napoli, Italy.

Sep 2020. *^{31}P-NMR: high field characterisation of biological molecules and potential applications to the study of phosphorylated sugars fluxes*. Magnetic Resonance meets the future of biophysics, Biophysical Society Virtual Networking Event.

Jul 2019. *Biochemistry And Bioinformatics In The Service Of PMM2-CDG: Finding Pharmacological Chaperones And Modifier Genes*. 4th World Conference on CDG, Lisbon, Portugal.

Teaching Experience

2025-2026	***Cultore della Materia* in Biochemistry**, Department for the Promotion of Human Science and Quality of Life San Raffaele Open University, Rome
May 2026	***From protein folding to glycosylation: the role of sugars in the functional fate of proteins***, Seminar in the Nutrition Biochemistry course, Department for the Promotion of Human Science and Quality of Life San Raffaele Open University, Rome
2021-2023	**Teaching in the PhD Program in Molecular Life Sciences**, 36th, 37th, 38th cycles, DiSTABiF,University of Campania "Luigi Vanvitelli"
Dec 2018	***Congenital disorders of glycosylation: an example of very rare genetic diseases***, Alternanza Scuola-Lavoro – Lecture for high-school students, Institute of Biomolecular Chemistry (CNR)
May 2018	**Open Educational Farms**, Istituto Zooprofilattico Sperimentale del Mezzogiorno

Participation in Funded Research Projects

Studying rare diseases to understand common diseases: congenital disorders of glycosylation and neurodevelopmental disorders
PI: Dr. G. Andreotti – Institute of Biomolecular Chemistry - CNR
- Accordo per la coesione della Regione Campania - Promozione di progetti di ricerca, sviluppo sperimentale e innovazione collaborativi nel campo delle malattie rare - Intervento n. 28

Analysis of genetic variants associated with Beckwith-Wiedemann syndrome
PI: Prof. A Riccio – DiSTABiF, University of Campania L. Vanvitelli
- Italian MInistry of University and Research (MUR) PRIN 2022 - B2N2BY

Rescuing the function of Trem2 variants associated with Alzheimer's Disease via a novel class of small molecules
PI: Prof. A. Fontana - Institute of Biomolecular Chemistry - CNR
- Italian Ministry of Health - PNRR-MAD-2022-12376849

Characterisation of engineered PMM2-CDG fibroblasts as tools for deciphering the molecular basis of the disease and developing therapeutic approaches
PI: Dr. G. Andreotti – Institute of Biomolecular Chemistry - CNR
- National Research Council, CNR – Joint bilateral agreement CNR –SAS (Slovak Academy of Sciences) – 2023- 2024

Causes and mechanisms of genomic imprinting disturbances in Beckwith-Wiedemann syndrome and Wilms Tumor
PI: Prof. A Riccio – DiSTABiF, University of Campania L. Vanvitelli
- Italian Association for Cancer Research (AIRC) – IG24405–2021-2026

Molecular genetics and new directions for clinical management of growth disorders associated with genomic imprinting
PI: Prof. A Riccio – DiSTABiF, University of Campania L. Vanvitelli
- Italian Ministry of Education, University and Research (MIUR) PRIN 2015 - 2015JHLY35$_0$02

Pharmacological chaperones for the cure of rare diseases
PIs: Prof. M.V. Cubellis – Dept. Biology, University of Napoli Federico II and PI: Dr. G. Andreotti – Institute of Biomolecular Chemistry – CNR
- Telethon Foundation - GGP12108–2012-2016

Scientific Activities

Scientific Service and Leadership

2026	**FEBS Open Bio**, Junior Ambassador
2025-2026	**Coordinator of the Young Section and Representative for the Scientific Group *Proteins* Young Section**, Italian Society of Biochemistry and Molecular Biology

Scientific Meetings Organization

Structural and functional characterization of proteins and their modulation. Meeting of *Proteins* Group in SIB, 20th-22nd May 2026, University of Salerno. Member of the Scientific Committee - Young section.

1st SIB Young Meeting – an international FEBS-supported education event. 25th-27th Mar 2026, Abano Terme (PD). Member of the Scientific and Organizing Committee. Led the application for financial support submitted to the FEBS Education and Training Committee.

La Chimica tra Tradizione e Innovazione. Italian Chemical Society Meeting, 20th Feb 2026, CNR Area della Ricerca NA3 Pozzuoli (NA). Member of the Organizing Committee.

WebPro Proteins on the Web 2021.Meeting of Proteins Group in SIB, 20th-21st May 2021, Virtual Conference. Member of the Scientific and Organizing Committee.

Peer Review

Reviewed several scientific papers (Springer Nature, MDPI, Frontiers journals).

Professional Memberships

Italian Society of Biochemistry and Molecular Biology, since 2019
Italian Chemical Society, since 2024
Italian National Order of Biologists (ONB), since 2020

Other activities

Member of the Evaluation Committee. High-school students ISSA Competition – XIV and XV editions. Held under the patronage of the Institute of Biomolecular Chemistry – CNR of the Institute of Biomolecular Chemistry – CNR.

English-Italian Translation. Immunology and CDG Questionnaire, in collaboration with the CDG Allies – Professionals and Patients Associations International Network.