

## GIOVANNI MEOLA CV

Giovanni Meola, MD, PhD  
Professor and Chair of Neurology  
University of Milan  
IRCCS Policlinico San Donato  
Department of Neurology and Stroke-Unit  
Piazza E. Malan, 1  
20097 San Donato Mil. (MI) – Italy  
Phone: 39-02-52774480  
Fax: 39-02-5274717  
e-mail: [giovanni.meola@unimi.it](mailto:giovanni.meola@unimi.it)



### WORK EXPERIENCE

- Assistant Professor of Neurology (1975-85) and Associate Professor of Neurology (since 1986), Institute of Clinical Neurology, Università degli Studi di Milano, Milano, Italy.
- Director of Division of Neurology II (since 1992), Director of Division of Neurology I and Chairman of Department of Neurology (since 1994), San Donato Hospital, San Donato Milanese, Milano, Italy.
- Director of Stroke-Unit (since 2004), San Donato Hospital, San Donato Milanese, Milano, Italy.
- Director Neurology Residency Program – University of Milan (since 2013).
- Full Professor of Neurology, University of Milan (since 2001).
- Director of Department of Neurology – Stroke Unit and SM Center (ongoing).
- Director of Residency Programm in Neurology – University of Milan since 2013 (ongoing).
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### EDUCATION AND TRAINING

- Degree in Medicine and Surgery cum laude (1973), Università degli Studi di Milano, Milano, Italy;
- Residency in Neurology (1977),
- Neuromuscular Laboratory Newcastle General Hospital (Prof. JN Walton, 1974-75, 1978), Newcastle, England;
- Neuromuscular Laboratory Hammersmith Hospital (Prof. V Dubowitz, 1975), London, England;
- Houston Merritt Neuromuscular Laboratory Department of Neurology, Columbia University (Prof. S Di Mauro e AF Miranda, 1980, 1981), New York, USA;
- Neuromuscular Laboratory Montreal Neurological Institute (Prof. G Karpati, 1991), Hopital de l'Enfant Jesus (Prof. JP Tremblay, 1991), Quebec, Canada;
- External Examiner of Claude Lebreque's PhD Neurobiological Thesis in 1993, for Laval University (1993);
- Visiting Professor University of Rochester (NY) since 1995;
- Nominated by University of Göteborg (Sweden) as senior Faculty opponent for the PhD thesis on myotonic dystrophy type 1 (DM1) written by Stephan Winblad (2006).
- Visiting Professor of Neurology, University of Beograd (Serbia) since 2009.
- Member of Ethical Committee (San Raffaele Institute) since 2013.
- Nominated by University of Paris Descartes as Discussant for PhD thesis on "DM1 neuropathogenesis in transgenic mouse models" by Geraldine Sicot, 2013.
- Organizer of Italian Neurology Residence Day (from 2013 to 2018).

## **GRANTS**

I conducted clinical research according to GCP

- 1975-present: From Public Ministry of Instruction (MURST 60%)
- 1991: From Telethon
- 1998, 1999: From MURST 40%
- 1999: Special Project about PROMM, University of Milan
- 1999-2001: Italian Ministry of University and Research Grant
- 2001: University of Milan (Joint Project with the European Community)
- 2000-2005: FIRST
- 2001-2007: COFIN
- 2002-present: Clinical Trial with WMS
- 2004-2005: Co-investigator Trial (University of Rochester)
- 2005: Co-investigator in Telethon Grant in QoL (quality of life) in adult neuromuscular disease
- 2008-2013: Co-PI by AFM
- 2013-2016: Co-PI by Telethon
- 2016-2017: By MDF on "Insulin resistance in DM"
- 2013-2016: By Welfare Ministry
- 2018-2019: Co-PI by Welfare Italian Ministry on "PEARL study":

## **MEMBERSHIP**

- American Neurological Association
- American Academy of Neurology;
- European Academy of Neurology;
- American Tissue Culture Association;
- World Muscle Society;
- Società Italiana di Neurologia;
- Società Italiana di Neuropatologia;
- Società Italiana di Istochimica;
- Centro "Dino Ferrari" per le Malattie Neuromuscolari.

## **EDITORIAL BOARD**

- Editor in Chief – Frontiers in Neurology, Neuromuscular disease section- since 2017
- Neuromuscular Disorders (Associate Editor);
- European Journal of Histochemistry;
- Neurological Sciences;
- Acta Myologica;
- Journal of Neuromuscular Disease.

## **REFEREE**

- Acta Neurologica Scandinavica
- American Journal of Medical Genetics Part B: Neuropsychiatric Genetics
- Annals of Neurology
- European Journal of Neurology
- Journal of the Neurological Sciences
- Journal of Neurology
- Journal of Neurology, Neurosurgery and Psychiatry
- Molecular Basic of Disease

- Muscle & Nerve
- Nature Clinical Practice Neurology
- Nature Scientific Report
- Nature communications
- Neurology

Author of more than 800 publications, including full papers in peer-reviewed Journals (254), Chapters (41), Abstracts and Invited Lectures and Seminars (720). IF: 822,229.  
H-index (Scopus): 37 (5122 citations on 233 papers – from 2003 to 2018).

## **ORGANIZATION OF NATIONAL AND INTERNATIONAL CONFERENCES**

### **National**

- 1992-present: “ Martedì di San Donato” Neurological Seminars
- 2010: Italian Association of Myology (AIM)

### **International**

- 1997: Co-organizer 54th ENMC International Workshop: PROMM and other Proximal Myotonic Syndrome
- 2000: Co-organizer 84th ENMC International Workshop: PROMM and other Proximal Myotonic Syndrome
- 2003: Co-organizer 115th ENMC International Workshop: DM2/PROMM and other myotonic dystrophies
- 2006: Co-organizer 140th ENMC International Workshop: Myotonic dystrophy DM2/PROMM and other Myotonic dystrophies with guidelines on management
- 2007: Organizer and Chairman IDMC-6 (International Myotonic Dystrophy Consortium Meeting – 6); Milan September 12-15, 2007
- 2010: Organizer and Chairman 1st Workshop “Cerebral Involvement in Myotonic Dystrophy type I and II”; Paris, Institute de Myologie – Hôpital la Salpêtrière;
- 2011: Organizer and Chairman 2nd Workshop “Cerebral Involvement in Myotonic Dystrophy type I and II”; Paris, Institute de Myologie – Hôpital la Salpêtrière;
- 2011: Founder of FMM (Fondazione Malattie Miotoniche)
- 2013: Organizer and Chairman of 4th Workshop Myotonic Dyustrophy cerebral Involvement, Ferrere (AT)-Italy
- 2014: Organizer and Chairman of DM-CNS 5th Workshop, University of Milan, Italy

### **OTHER LANGUAGES**

- English excellent
- French good

## SELECTED PEER-REVIEW PUBLICATIONS (in chronological order)

(Publications selected from 254 peer-review publications)

1. Statland JM, Bundy BN, Wang Y, Rayan DR, Trivedi JR, Sansone V, Salajegheh MK, Venance SL, Ciafaloni E, Matthews E, **Meola G**, Herbelin L, Griggs RC, Barohn RJ, Hanna MG, for the Consortium for Clinical Investigation of Neurologic Channelopathies. Mexiletine for Symptoms and signs of Myotonia in Nondystrophic myotonia. *JAMA* 2012, October 3; Vol 308(13).
2. Cardani R, Giagnacovo M, Botta A, Rinaldi F, Morgante A, Udd B, Raheem OR, Penttila S, Suominen T, Renna LV, Sansone V, Bugiardini E, Novelli G, **Meola G**. Co-segregation of DM2 with a recessive CLCN1 mutation in juvenile onset of myotonic dystrophy type 2. *J Neurol*. Published online: 10 March 2102
3. Ulzi G, Lecchi M, Sansone V, Redaelli E, Corti E, Saccomanno D, Pagliarani S, Corti S, Magri F, Raimondi M, D'Angelo G, Modoni A, Bresolin N, **Meola G**, Wanke E, Comi GP, Lucchiari S. Myotonia congenita: novel mutations in CLCN1 gene and functional characterizations in Italian patients. *J Neurol Sci* 2012;318:65-71.
4. Jones K, Wei C, Iakova P, Bugiardini E, Schneider-Gold C, **Meola G**, Woodgett J, Killian J, Timchenko NA, Timchenko LT. GSK3 $\beta$  mediates muscle pathology in myotonic dystrophy. *J Clin Invest* 2012;122:4461-72.
5. Gagnon C, **Meola G**, Hébert LJ, Puymirat J, Laberge L, Leone M. Report of the first Outcome Measures in Myotonic Dystrophy type 1 (OMMYD-1) international workshop: Clearwater, Florida, November 30, 2011. *Neuromuscul Disord*. 2013 Dec;23(12):1056-68.
6. Cardani R, Bugiardini E, Renna LV, Rossi G, Colombo G, Valaperta R, Novelli G, Botta A, **Meola G**. Overexpression of CUGBP1 in Skeletal Muscle from Adult Classic Myotonic Dystrophy Type 1 but Not from Myotonic Dystrophy Type 2. *PLoS One*. 2013;8(12):e83777.
7. Bugiardini E, **Meola G**, on behalf of the DM-CNS Group. Consensus on cerebral involvement in myotonic dystrophy Workshop report: May 24-27, 2013, Ferrere (AT), Italy. *Neuromuscular Disord* 2014 [Epub ahead of print]
8. **Meola G**, Cardani R. Myotonic dystrophies: an update on clinical aspects, genetic, pathology, and molecular pathomechanisms. *Biochim Biophys Acta* 2014;May 29 pii: S0925-4439(14)00147-1. doi: 10.1016/j.bbadis.2014.05.019. [Epub ahead of print]
9. Perfetti A, Greco S, Bugiardini E, Cardani R, Gaia P, Gaetano C, **Meola G**, Martelli F. Plasma microRNAs as biomarkers for myotonic dystrophy type 1. *Neuromuscul Disord*. 2014;24:509-15.
10. Perfetti A, Greco S, Fasanaro P, Bugiardini E, Cardani R, Manteiga JM, Riba M, Cittaro D, Stupka E, **Meola G**, Martelli Genome wide identification of aberrant alternative splicing events in myotonic dystrophy type 2. *PLoS One*. 2014 Apr 10;9(4):e93983.
11. Cardani R, Giagnacovo M, Rossi G, Renna LV, Bugiardini E, Pizzamiglio C, Botta A, **Meola G**. Progression of muscle histopathology but not of spliceopathy in myotonic dystrophy type 2. *Neuromuscul Disord*. 2014 Jun 25. pii: S0960-8966(14)00601-4. doi: 10.1016/j.nmd.2014.06.435. [Epub ahead of print].
12. Klinck R, Fourrier A, Thibault P, Toutant J, Durand M, Lapointe E, Caillet-Boudin ML, Sergeant N, Gourdon G, **Meola G**, Furling D, Puymirat J, Chabot B. RBFOX1 Cooperates with MBNL1 to Control Splicing in Muscle, Including Events Altered in Myotonic Dystrophy Type 1. *PLoS One*. 2014 Sep 11;9(9):e107324. doi: 10.1371/journal.pone.0107324. eCollection 2014. PMID: 25211016 [PubMed - as supplied by publisher].
13. Bugiardini E, Rivolta I, Binda A, Soriano Caminero A, Cirillo F, Cinti A, Giovannoni R, Botta A, Cardani R, Wicklund MP, **Meola G**. SCN4A mutation as modifying factor of myotonic dystrophy type 2 phenotype. *Neuromuscul Disord*. 2015 Apr;25(4):301-7.
14. Gagnon C, **Meola G**, Hébert LJ, Laberge L, Leone M, Heatwole C. Report of the second

- Outcome Measures in Myotonic Dystrophy type 1 (OMMYD-2) international workshop San Sebastian, Spain, October 16, 2013. *Neuromuscul Disord*. 2015 Jul;25(7):603-16.
15. Passeri E, Bugiardini E, Sansone VA, Pizzocaro A, Fulceri C, Valaperta R, Borgato S, Costa E, Bandera F, Ambrosi B, **Meola G**, Persani L, Corbetta S. Gonadal failure is associated with visceral adiposity in myotonic dystrophies. *Eur J Clin Invest*. 2015 May 7. doi: 10.1111/eci.12459. [Epub ahead of print].
  16. Jones K, Wei C, Schoser B, **Meola G**, Timchenko N, Timchenko L. Reduction of toxic RNAs in myotonic dystrophies type 1 and type 2 by the RNA helicase p68/DDX5. *Proc Natl Acad Sci U S A*. 2015 Jun 15. pii: 201422273. [Epub ahead of print].
  17. **Meola G** and Cardani R. Myotonic Dystrophy type 2: an update on clinical aspects, genetic and pathomolecular mechanism. *BBA-MOL BASIS DIS*. 2015;1852:594-606.
  18. Bosco G, Diamanti S, **Meola G**; DM-CNS Group. Workshop Report: consensus on biomarkers of cerebral involvement in myotonic dystrophy, 2-3 December 2014, Milan, Italy. *Neuromuscul Disord* 2015;25:813-23.
  19. Esposito F, Cè E, Rampichini S, Limonta E, Venturelli M, Monti E, Bet L, Fossati B, **Meola G**. Electromechanical delay components during skeletal muscle contraction and relaxation in patients with myotonic dystrophy type 1. *Neuromuscul Disord*. 2016;26(1):60-72.
  20. Sansone VA, Burge J, McDermott MP, Smith PC, Herr B, Tawil R, Pandya S, Kissel J, Ciafaloni E, Shieh P, Ralph JW, Amato A, Cannon SC, Trivedi J, Barohn R, Crum B, Mitsumoto H, Pestronk A, **Meola G**, Conwit R, Hanna MG, Griggs RC; Muscle Study Group. Randomized, placebo-controlled trials of dichlorphenamide in periodic paralysis. *Neurology*. 2016;86(15):1408-16.
  21. Guglielmi V, Oosterhof A, Voermans NC, Cardani R, Molenaar JP, van Kuppevelt TH, **Meola G**, van Engelen BG, Tomelleri G, Vattermi G. Characterization of sarcoplasmic reticulum Ca(2+) ATPase pumps in muscle of patients with myotonic dystrophy and with hypothyroid myopathy. *Neuromuscul Disord*. 2016;26(6):378-85.
  22. Serra L, Cercignani M, Bruschini M, Cipolotti L, Mancini M, Silvestri G, Petrucci A, Bucci E, Antonini G, Licchelli L, Spanò B, Giacanelli M, Caltagirone C, **Meola G**, Bozzali M. "I Know that You Know that I Know": Neural Substrates Associated with Social Cognition Deficits in DM1 Patients. *PLoS One*. 2016 Jun 3;11(6):e0156901. doi: 10.1371/journal.pone.0156901. eCollection 2016.
  23. Serra L, Mancini M, Silvestri G, Petrucci A, Masciullo M, Spanò B, Torso M, Mastropasqua C, Giacanelli M, Caltagirone C, Cercignani M, **Meola G**, Bozzali M. Brain Connectomics' Modification to Clarify Motor and Nonmotor Features of Myotonic Dystrophy Type 1. *Neural Plast*. 2016;2016:2696085. doi: 10.1155/2016/2696085. Epub 2016 May 25.
  24. Winblad S, Samuelsson L, Lindberg C, **Meola G**. Cognition in myotonic dystrophy type 1: a 5-year follow-up study. *Eur J Neurol*. 2016 Jun 20. doi: 10.1111/ene.13062. [Epub ahead of print]
  25. Peric S, Rakocevic Stojanovic V, Mandic Stojmenovic G, Ilic V, Kovacevic M, Parojcic A, Pesovic J, Mijajlovic M, Savic-Pavicevic D, **Meola G**. Clusters of cognitive impairment among different phenotypes of myotonic dystrophy type 1 and type 2. *Neurol Sci*. 2016 Nov 28. [Epub ahead of print] PubMed PMID: 27896491
  26. Perfetti A, Greco S, Cardani R, Fossati B, Cuomo G, Valaperta R, Ambrogi F, Cortese A, Botta A, Mignarri A, Santoro M, Gaetano C, Costa E, Dotti MT, Silvestri G, Massa R, **Meola G**, Martelli F. Validation of plasma microRNAs as biomarkers for myotonic dystrophy type 1. *Sci Rep*. 2016 Dec 1;6:38174. doi: 10.1038/srep38174. PubMed PMID: 27905532; PubMed Central PMCID: PMC5131283.
  27. **Meola G**, Cardani R. Myotonic dystrophy type 2 and modifier genes: an update on clinical and pathomolecular aspects. *Neurol Sci*. 2017 Jan 11. doi: 10.1007/s10072-016-2805-5. [Epub ahead of print] Review. PubMed PMID: 28078562
  28. Gourdon G, **Meola G**. Myotonic Dystrophies: State of the Art of New Therapeutic

- Developments for the CNS. *Front Cell Neurosci.* 2017 Apr 20;11:101. doi: 10.3389/fncel.2017.00101. eCollection 2017. Review.
29. **Meola G**, Biasini F, Valaperta R, Costa E, Cardani R. Biomolecular diagnosis of myotonic dystrophy type 2: a challenging approach. *J Neurol.* 2017 May 26. doi: 10.1007/s00415-017-8504-1. [Epub ahead of print] Review.
  30. Renna LV, Bosè F, Iachettini S, Fossati B, Saraceno L, Milani V, Colombo R, **Meola G**, Cardani R. Receptor and post-receptor abnormalities contribute to insulin resistance in myotonic dystrophy type 1 and type 2 skeletal muscle. *PLoS One* 2017 Sep 15;12(9):e0184987. doi: 10.1371/journal.pone.0184987. eCollection 2017. PMID: 28915272.
  31. Dozio E, Passeri E, Cardani R, Benedini S, Aresta C, Valaperta R, Corsi Romanelli M, **Meola G**, Sansone V, Corbetta S. Circulating Irisin Is Reduced in Male Patients with Type 1 and Type 2 Myotonic Dystrophies. *Front Endocrinol (Lausanne).* 2017 Nov 14;8:320. doi: 10.3389/fendo.2017.00320. eCollection 2017.
  32. Provenzano C, Cappella M, Valaperta R, Cardani R, **Meola G**, Martelli F, Cardinali B, Falcone G. CRISPR/Cas9-Mediated Deletion of CTG Expansions Recovers Normal Phenotype in Myogenic Cells Derived from Myotonic Dystrophy 1 Patients. *Mol Ther Nucleic Acids.* 2017 Dec 15;9:337-348. doi: 10.1016/j.omtn.2017.10.006. Epub 2017 Oct 14.
  33. Sellier C, Cerro-Herreros E, Blatter M, Freyermuth F, Gaucherot A, Ruffenach F, Sarkar P, Puymirat J, Udd B, Day JW, **Meola G**, Bassez G, Fujimura H, Takahashi MP, Schoser B, Furling D, Artero R, Allain FHT, Llamusi B, Charlet-Berguerand N. rbFOX1/MBNL1 competition for CCUG RNA repeats binding contributes to myotonic dystrophy type 1/type 2 differences. *Nat Commun.* 2018 May 22;9(1):2009. doi: 10.1038/s41467-018-04370-x. PMID: 29789616.
  34. Cavalli M, Fossati B, Vitale R, Brigonzi E, Ricigliano VAG, Saraceno L, Cardani R, Pappone C, **Meola G**. Flecainide-Induced Brugada Syndrome in a Patient With Skeletal Muscle Sodium Channelopathy: A Case Report With Critical Therapeutical Implications and Review of the Literature. *Front Neurol.* 2018 May 30;9:385. doi: 10.3389/fneur.2018.00385. eCollection 2018. PMID: 29899727.
  35. Altamura C, Lucchiari S, Sahbani D, Ulzi G, Comi GP, D'Ambrosio P, Petillo R, Politano L, Vercelli L, Mongini T, Dotti MT, Cardani R, **Meola G**, Lo Monaco M, Matthews E, Hanna MG, Carratù MR, Conte D, Imbrici P, Desaphy JF. The analysis of myotonia congenita mutations discloses functional clusters of amino acids within CBS2 domain and C-terminal peptide of CIC-1 channel. *Hum Mutat.* 2018 Jun 23. doi: 10.1002/humu.23581. [Epub ahead of print] PMID: 29935101.
  36. Cappella M, Perfetti A, Cardinali B, Garcia-Manteiga JM, Carrara M, Provenzano C, Fuschi P, Cardani R, Renna LV, **Meola G**, Falcone G, Martelli F. High-throughput analysis of the RNA-induced silencing complex in myotonic dystrophy type 1 patients identifies the dysregulation of miR-29c and its target ASB2. *Cell Death Dis.* 2018 Jun 28;9(7):729. doi: 10.1038/s41419-018-0769-5. PMID: 29955039.
  37. Binda A, Renna LV, Bosè F, Brigonzi E, Botta A, Valaperta R, Fossati B, Rivolta I, **Meola G**, Cardani R. SCN4A as modifier gene in patients with myotonic dystrophy type 2. *Sci Rep.* 2018 Jul 23;8(1):11058. doi: 10.1038/s41598-018-29302-z. PMID: 30038349.

I authorize the use of my personal data in compliance with GDPR (Legislative Decree UE 2016/679).

San Donato Mil. April 2019