Name MARIA GIUSEPPINA
Surname MIANO
Position CNR Senior Researcher

Education

1985−1990. University of Naples Federico II, Faculty of Biological Science, Degree summa cum laude.
1993−1997. University of Rome La Sapienza, Faculty of Medicine Specialist in Medical Genetics summa cum laude.
1999−2000. University of Pavia Faculty of Medicine. European Schools for Advanced Studies in Molecular Medicine and Genetic Epidemiology. Master of Science in Statistical Genetics.
2000−2003. University of Naples Federico II Faculty of Biological Science, PhD in Systematic Biology.

Professional Experience and Training

1991−1992. International Institute of Genetics and Biophysics−CNR Naples. Post−degree Training.
1992−1993. University of Naples Federico II Unit of Cytogenetic and Prenatal Diagnosis. Visiting Scientist.
1994−1995. National Cancer Institute, Naples. APRO Fellowship.
1995−1996. International Institute of Genetics and Biophysics. CNR Fellowship.
1997. International Institute of Genetics and Biophysics. Telethon Fellowship.
1998−2000. International Institute of Genetics and Biophysics. Employment with Minister of University and Scientific Research – Italy.
1998-1999. Medical Research Council (MRC, UK). Human Genetic Unit−MRC. Edinburgh UK.

Employment and Research Experience

2001−2021. Researcher of the National Research Council (CNR) at the Institute of Genetics and Biophysics, Naples, Italy. Group Leader of Human Molecular Neurogenetics Laboratory.
2021-present. Senior Scientist of the National Research Council (CNR) at the Institute of Genetics and Biophysics, Naples, Italy. Group Leader of Human Molecular Neurogenetics Laboratory

Scientific Visit

1998−1999. Medical Research Council (MRC). Human Genetic Unit−MRC, Edinburgh. Laboratory of Prof. Alan Wright. EMBO fellowship and Employment with MRC.
2013. University of Adelaide. Laboratory of Prof. Jozef Gecz. CNR-Short Term Mobility Program.

Research Grants

2007−2008. Programma Italia−USA Malattie Rare Istituto Superiore di Sanità: X−linked or Autosomal Rare Mental Retardation Syndromes: Phenotypic Analysis in Transgenic Mouse models. Principal Investigator of CNR Unit Dr. MG Miano.
2007. PRIN− Studio degli effetti prodotti dallo stress durante l'adolescenza e loro implicazioni psicopatologiche Coordinator Dr. Riva Investigator CNR, Principal Investigator of CNR Unit Dr. MG Miano.
2012-2014. Foundation Jerome Lejeune. Cell-based drug discovery to compensate epigenetics and morphological defects associated to ARX-PolyAlanine Intellectual Disabilities. Principal Investigator Dr. MG Miano.
2015-2017. Foundation Jerome Lejeune. Towards the identification of potential epi- therapies for XLID/Epilepsy by in vivo treatment of Arx polyalanine mouse model. Principal Investigator Dr. MG Miano.
2014-2018. Telethon Foundation. Dissecting the Aristaless-related Homeobox Epilepsy path to find druggable target molecules. Principal Investigator Dr. MG Miano.
2014-2018. GW Pharmaceuticals. Exploring the effects of Phytocannabinoids on ARX murine models. Principal Investigator Dr. MG Miano.
2019. GW Pharmaceuticals. Exploring the effects of Phytocannabinoids on ARX murine models. Principal Investigator Dr. MG Miano.
2020- 2024. Maria Rosaria Maglione Foundation Onlus. Biomarkers in Glioblastoma. Principal Investigator Dr. MG Miano.
2019-2023. Italian Minister of Economic Development (MISE). Functional Genomics of rare diseases: Development of high resolution diagnostic kits (Coordinator Prof. Simeone). Principal Investigator Dr. MG Miano Of Research Unit on Cortical malformations and pediatric epilepsy.
2022-2023. Maria Rosaria Maglione Foundation Onlus. Analysis of FOXP1 mutations in patients with language impairments. Principal Investigator Dr. MG Miano.
2022-2024. Foundation Jerome Lejeune. Analysis of the proteome and behavioural responses to the HDACi Vorinostat in Arx mouse model of developmental epileptic encephalopathy. Principal Investigator Dr. MG Miano.

Scientific meeting Grant

2017. EMBO. EMBO-IGB Workshop “From epigenome towards epitranscriptome in cell fate choice”.
2019. Foundation Jerome Lejeune. “19th International Workshop on Fragile X and other Neurodevelopmental

disorders".

International and National PhD Committee
2018. Institute of Neuroscience, Alicante, Spain.
2019. Department of Pharmacology and Neurology, University of Naples Federico II, Naples, Italy
2020. Department of health of Women 's and Children's Health, University of Padua, Italy

Teaching Duties
2010. University of Naples, "Federico II, Italy. Human Genetics Lectures.
2011-2014. University of Basilicata, Potenza, Italy. Annual Course of Human Physiology
2013. University of Lecce, Lecce (Italy). High-tech School "Repair Project". Master in Tissue Engineering technologies.
2014. Second University of Naples. "Brain, Know Thy Transcriptome, Know Thyself". PhD course lecture.
2015-2016. University of Sannio (Benevento, Italy), BioGeM, Annual Course of Genomics
2017-2018. University of Campania "Luigi Vanvitelli" _INCIPIT PhD program. PhD course lecture.
2018. University of Sannio (Benevento, Italy) "Lincei for the School". Lectures.
2021. University of Campania "Luigi Vanvitelli" . Biomolecular Science PhD course lecture.
2022. University of Naples. Neuroscience Course. Lecture.

Invited Seminars and selected talks
2013. 16th International Fragile X and Other Early-Onset Disorders Workshop.17-20 September Adelaide, Australia, 2013.
2014. University of Bologna. Department of Medical Genetics. Bologna, 5 February 2014.
2014. University of Padova. Department of Paediatric Neurology. Padova, 20 March 2014.
2014. University of Piemonte Orientale "A. Avogadro" Department of Health Sciences. 21 July 2014.
2014. Galliera Hospital Medical Genetics Department, Genova, 5 December 2014.
2015. 17th International Fragile X and Other Early-Onset Disorders Workshop. 27-30 September 2015. Strasbourgh, France.
2015. Mendel Institute, Rome, 17 December 2015.
2016. University of Torino, Department of Genetics, 11 October 2016, Torino.
2016. Istituto di Chimica molecolare – CNR, 21 April 2016, Pozzuoli,
2017. SIGU Workshop - Chromatin human diseases – Bari, 8-9 May 2017
2019. UMHD9 - 9th International Conference on Unstable Microsatellites and Human Disease – 21-26 April, Capri, Italy.
2021. Department of Biomedical Sciences-CNR Conference - Mechanistic Insights into Neurological Disorders and New Therapeutic Strategies – Rome, July 7th 2021
2021. VI Course of Medical Genetics/SIGU – Naples, 17th December 2021.
2022. SCN2A Awareness Day– Webinar 24th February 2022.
2023. University of Naples "Federico II" – Seminar on Trinucleotide repeats in Neurodevelopmental disorders – 23th March 2023.

Awards
2013. SIGU Award 2013 Best Oral Presentation in Clinical Genetics
Poeta L, Padula A, Fusco F, Shoubridge C, Manganelli G, Filosa S, Collombat P, Friocourt G, Passafaro M, Helin K, Altucci L, Gecz J, Ursini MV, Miano MG. *ARX-KDM5C regulatory path in malignant Epilepsy: how to correct KDM5C-dependent alterations?* XVI Congresso Nazionale Società Italiana di Genetica Umana, SIGU. Settembre 25-27, Roma, 2013.
2014. ESHG Award 2014 Best Poster Presentation in Basic Research
Poeta L, Padula A, Shoubridge C, Zucchelli S, Fusco F, Filosa S, Collombat P,. Helin K, Altucci L, Lioi MB, Gustincich S, Gecz J, Ursini MV, Miano MG. *Functional studies of ARX mutants linked to neurophenotypes and application of rescue strategies targeting KDM5C downregulation* 47th European Society of Human Genetics, ESHG. 31 May-3 June, Milan, 2014

CNR Duties
2009-to 2016. Member of Institute Committee at IGB-CNR
2014- Present. Member of IGB Worshop Committee

Editorial Board & Guest Editor
2019-2020. Genes – MDPI Special Issue "Selected Papers from the International Workshop on Fragile X and Other Neurodevelopmental Disorders". Guest Editor – MG. Miano.
2018-to date. Frontiers in Molecular Neuroscience. Reviewer Editor –MG. Miano
2020 –to date. Frontiers in Molecular Biosciences - Cellular Biochemistry. Reviewer Editor – MG- Miano.
2020- to date. Frontiers in Neuroscience – Neurodegeneration. Reviewer Editor – MG- Miano.

2021- to date. Frontiers in Neuroscience. Research Topic "The Rna worl: Non-coding RNAs and Innovative Therapies in Neurological Disorders_II edition". Guest Editor – MG. Miano.
2022- to date. Biomolecules – MDPI Special Issue "Neurodevelopmental Disorders: Linking Genetics and Epigenetics to Disease-Related Pathways". Guest Editor – MG. Miano.

Organization of Meeting and Symposia

2006. Riva del Garda, Italy VIII FISV 2006. Symposia: Genetics models of infertility from yeast to humans. Symposia Scientific Organizer and Chair.
2011. Milano, Italy. Italian Society of Human Genetics (SIGU) 2011. Symposia: Mouse models to study ID: from the identification of pathways to therapies. Symposia Scientific Organizer and Chair.
2013. Rome, Italy. Italian Society of Human Genetics (SIGU) 2013. Parallel Session: Repeat Expansion Disorders: instability mechanisms, chromatin structure and therapeutic perspectives. Symposia Scientific Organizer and Chair.
2015. Capri, Italy EMBO-IGB Workshop. Stem cell mechanobiology in development and disease. 18-21 October 2015. Meeting Coordinator.
2017. Naples, Italy. Italian Society of Human Genetics (SIGU) 2017. Symposia: RNA editing in health and disease. Scientific Organizer and Chair.
2018. Capri, Italy. 19th International Conference on Unstable Microsatellites and Human Disease (UMHD9). 21-26 April 2018. Meeting Scientific Organizer and Chair.
2018. Capri, Italy EMBO-IGB Workshop. "From epigenome towards epitranscriptome in cell fate choice". 14-17 October 2018. Meeting Coordinator.
2019. Sorrento, Italy. "19th International Workshop on Fragile X and other neurodevelopmental disorders". 18-21 September 2019. Meeting Scientific Organizer and Chair.
2021. Virtual meeting "Targeting the (un)usual suspects in cancer" 29th IGB Workshop. 2-3 December 2021.

Scientific Committee of National and International Meetings

2014-2015. Member of Scientific Committee Board of Italian Society of Human Genetics (SIGU).
2017-2018. Member of Scientific Committee Board of the International Conference on Unstable Microsatellites and Human Disease (UMHD).
2017-2019. Member of Scientific Committee Board of International Fragile X and other Early-Onset Cognitive Disorders Workshop.
2017-to date. Member of the Scientific Committee Board of Neapolitan Brain group (NBG).
2022-to date. Member of the Scientific Committee Board of SCN2A Italia Famiglie in rete.
2022-2023. Member of Scientific Committee Board of Italian Society of Human Genetics (SIGU).
2022-2023. Member of Scientific Committee Board of International Fragile X and other Early-Onset Cognitive Disorders Workshop.
2023-to date. Member of Scientific Committee Board of KARES FOUNDATION-KDM5C, Advocacy, Research, Education & Support.

Main Publications

1. Vervoort R, Lennon A, Bird A, Tulloch B, Axton R, Miano MG, Meindl A, Meitinger T, Ciccodicola A, Wright AF. (2000). Mutational hot spot within a novel RPGR exon in X-linked retinitis pigmentosa. *Nat. Gen*., 25: 462-466.
2. Miano MG, Jacobson SG, Carothers A, Hanson I, Teague P, Lovell J, Cideciyan AV, Stone EM, Sheffield VC, Wright AF. 2000. Pitfalls in homozygosity mapping. *Am. J. Hum. Gen*. 67.
3. Miano MG, Testa F, Filippini F, Trujilio M, Conte I, Lanzara C, Millan JM, De bernardo C, Grammatico B, Mangino M, Torrente I, Cazzozzo R, Rinaldi E, Ventruto V, D'Urso M, Ayuso C, Ciccodicola A. (2001). Identification of novel RP2 mutations in a subset of X-linked Retinitis pigmentosa Families and prediction of new RP2 domains. *Human Mutat*. 18:109-19.
4. Annunziata I, Lanzara C, Conte I, Zullo A, Ventruto V, Rinaldi MM, D'Urso M, Casari G, Ciccodicola A, Miano MG. (2003). Mapping of MRX81 in Xp11.2-Xq12 suggests the presence of a new gene involved in Non Specific X-Linked Mental Retardation. *Am. J. Med Gen*. 118: 76-80.
5. Fusco F, Bardaro T, Fimiani G, Mercadante V, Miano MG, Falco G, Israel A, Courtois G, D'Urso M, Ursini MV. (2004). Molecular analysis of the genetic defect in a large cohort of IP patients and identification of novel NEMO mutations interfering with NF-kappaB activation. *Hum Mol Genet.* 13:1763-1773.
6. Fimiani G, Laperuta C, Falco G, Ventruto V, D'Urso M, Ursini MV, Miano MG. (2006). Heterozygosity mapping by QF-PCR reveals an interstitial deletion in Xq26.2-q28 associated to ovarian dysfunction. *Hum Reprod.*, 21:529-535.
7. Busiello R, Fimiani G, Miano MG, Aricò M, Santoro A, Ursini MV, Pignata C. (2006.) A91V perforin variation in healthy subjects and FHLH patients *Int J Immunogenet*. 33:123-125.
8. Fusco F, Mercadante V, Miano MG, D'Urso M, Ursini MV. (2006). Multiple regulatory regions and tissue specific transcription initiation mediate the expression of NEMO *Gene*, 383:99-107.
9. Abidi F, Miano MG, Murray J, Schwartz CE. (2007). A novel mutation in the PHF8 gene is associated with X linked mental retardation with cleft Lip/cleft palate. *Clin Genet.* 72:19-22.

10. Laperuta C, Spizzichino L, D'Adamo P, Monfregola J, Maiorino A, D'Eustacchio A, Ventruto V, Neri G, D'Urso M, Chiurazzi P, Ursini MV, Miano MG. (2007). MRX87 family with Aristaless dup24bp mutation and implication for polyAlanine expansions. *BMC Medical Genetics*, 4;8:25.
11. Miano MG, Laperuta C, Chiurazzi P, D'Urso M, Ursini MV. (2007.) Ovarian dysfunction and *FMR1* alleles in a large Italian family with POF and FRAXA disorders: Case report. *BMC Medical Genetics* 11;8:18.
12. Fusco F, Pescatore A, Bal E, Ghoul A, Paciolla M, Lioi MB, D'Urso M, Hadj Rabia S, Bodemer C, Bonnefont JP, Munnich A, Miano MG, Smahi A, Ursini MV. (2008). Alterations of *IKBKG/NEMO* locus and diseases: an update and a report of 14 novel mutations. *Hum Mut* 18:595-604.
13. Fusco F. Paciolla M, Pescatore A, Lioi MB, Ayuso C, Faravelli F, Mattia G, Zollino M, D'Urso, Miano MG, MV Ursini. (2009). Microdeletion/duplication at the Xq28 IP locus causes a de novo IKBKG/NEMO/IKKgamma exon4_10 deletion in families with incontinentia pigmenti. *Hum Mut,* 30:1284-1291.
14. Fusco F, D'Urso M, Miano MG, Ursini MV. 2010. The LCR at the IKBKG locus is prone to recombine. *Am J Hum Genet.* 86:650-652.
15. Paciolla M, Boni R, Fusco F, Pescatore A, Poeta L, Ursini MV, Lioi MB, Miano MG. (2011). NF-kappa-B-inhibitor alpha (NFKBIA) is a Developmental Marker of NF-kB/p65 Activation during Oocyte Maturation and Early Embryogenesis. *Hum Reprod.* 26:1191-1201.
16. Fusco F, Paciolla M, Chen E, Li X, Genesio R, Conti A, Jones J, Poeta L, Lioi MB, Ursini MV, Miano MG. (2011). Genetic and molecular analysis of a new unbalanced X;18 rearrangement: localization of the diminished ovarian reserve disease locus in the distal Xq POF1 region. *Hum Reprod.* 26:3186-3196.
17. Fusco F, Paciolla M, Napolitano F, Pescatore A, D'Addario I, Bal E, Lioi MB, Smahi A, Miano MG, Ursini MV. (2012). Genomic architecture at the Incontinentia Pigmenti locus favours de novo pathological alleles through different mechanisms. *Hum Mol Genet.* 21(6):1260-71.
18. Ursini MV, Conte M, Pescatore A, Miano MG, Fusco F. Molecular Genetics of Incontinentia Pigmenti. *eLS*. 15, 10, 2012. https://doi.org/10.1002/9780470015902.a0024332.
19. Poeta L, Fusco F, Drongitis D, Shoubridge C, Manganelli G, Filosa S, Paciolla M, Courtney M, Collombat P, Lioi MB, Gecz J, Ursini MV, Miano MG. (2013). A Regulatory Path Associated with X-Linked Intellectual Disability and Epilepsy Links KDM5C to the Polyalanine Expansions in ARX. *Am J Hum Genet.* 92:114-125.
20. Conte MI, Pescatore A, Paciolla M, Esposito E, Miano MG, Lioi MB, McAleer MA, Giardino G, Pignata C, Irvine AD, Scheuerle AE, Royer G, Hadj-Rabia S, Bodemer C, Bonnefont JP, Munnich A, Smahi A, Steffann J, Fusco F, Ursini MV. (2013). Insight into IKBKG/NEMO Locus: Report of New Mutations and Complex Genomic Rearrangements Leading to Incontinentia Pigmenti Disease. *Hum Mutat.* 35:165-177.
21. Poeta L, Padula A, Attianese B, Valentino M, Verrillo L, Filosa S, Shoubridge C, Barra A, Schwartz CE, Christensen J, van Bokhoven H, Helin K, Lioi MB, Collombat P, Gecz J, Altucci L, Di Schiavi E, Miano MG. (2019) Histone demethylase KDM5C is a SAHA-sensitive central hub at the crossroads of transcriptional axes involved in multiple neurodevelopmental disorders. *Hum Mol Genet.* Dec 15;28(24):4089-4102.
22. Poeta, L, Drongitis, D, Verrillo, L, Miano, M.G. (2020) DNA Hypermethylation and Unstable Repeat Diseases: A Paradigm of Transcriptional Silencing to Decipher the Basis of Pathogenic Mechanisms. *Genes*, 11, 684-696.
23. Verrillo L, Mangano E, Drongitis D, Merelli I, Pischedda F, Piccoli G, Consolandi C, Bordoni R, Miano MG. (2020) A reliable strategy for single-cell RNA sequencing analysis using cryoconserved primary cortical cells. *J Neurosci Methods*. Sep 25;347:108960.
24. Petrone P, Giordano G, Vezzoli E, Pensa A, Castaldo G, Graziano V, Sirano F, Capasso E, Quaremba G, Vona A, Miano MG, Savino S, Niola M. (2020) Preservation of neurons in an AD 79 vitrified human brain. *PLoS One*. Oct 6;15(10):e0240017.
25. Poeta L, Padula A, Lioi MB, van Bokhoven H, Miano MG. (2021) Analysis of a Set of KDM5C Regulatory Genes Mutated in Neurodevelopmental Disorders Identifies Temporal Coexpression Brain Signatures. *Genes.* Jul 18;12(7):1088. doi: 10.3390/genes12071088.
26. Drongitis D, Caterino M, Verrillo L, Santonicola P, Costanzo M, Poeta L, Attianese B, Barra A, Terrone G, Lioi MB, Paladino S, Di Schiavi E, Costa V, Ruoppolo M, Miano M. G. **(**2022) Deregulation of microtubule organization and RNA metabolism in *Arx* models for Lissencephaly and Developmental epileptic encephalopathy. *Hum Mol Genet.* 2022 Jun 4;31(11):1884-1908. COVER.
27. Poeta L, Malacarne M, Padula A, Drongitis D, Verrillo L, Chiariello AM, Nicodemi M, Lioi MB, Piccione M, Coviello D, Miano MG. (2022) Further delineation of duplications of *ARX* locus detected in male patients with varying degrees of intellectual disability". *Int J Mol Sci. 2022 Mar 13;23(6):3084.*
28. Drongitis D, Verrillo L, De Marinis P, Orabona P, Caiola A, Turitto G, Alfieri A, Bruscella S, Gentile M, Moriello V, Sannino E, Di Muccio I, Costa V, Miano MG, de Bellis A. The Chromatin-Oxygen Sensor Gene *KDM5C* Associates with Novel Hypoxia-Related Signatures in Glioblastoma Multiforme. Int J Mol Sci. 2022 Sep 6;23(18):10250. doi: 10.3390/ijms231810250. PMID: 36142158; PMCID: PMC9498997.
29. Leonardi E, Aspromonte MC, Drongitis D, Bettella E, Verrillo L, Polli R, McEntagart M, Licchetta L, Dilena R, D'Arrigo S, Ciaccio C, Esposito S, Leuzzi V, Torella A, Baldo D, Lonardo F, Bonato G, Pellegrin S, Stanzial F, Posmyk R, Kaczorowska E, Carecchio M, Gos M, Rzońca-Niewczas S, A Miano MG*§ and Murgia A*§. Expanding the genetics and phenotypic spectrum of Lysine-specific demethylase 5C (KDM5C): a report of 13 novel variants. Eur J Hum Genet. 2023 Feb;31(2):202-215. **§ co-last and co-corresponding authors.*
30. Alagia M, Fecarotta S, Romano A, Parrini E, Auricchio G, Miano MG, Terrone G. A Novel Splicing SCN2A

Mutation in an Adolescent With Low-Functioning Autism, Acute Dystonic Movement Disorder, and Late-Onset Generalized Epilepsy. Pediatr Neurol. 2023 Jan;138:58-61. doi: 10.1016/j.pediatrneurol.2022.10.011. Epub 2022 Oct 27. PMID: 36401981.