Personal data

Surname/Name	Olimpia Musumeci	ID code	MSMLMP71A59C351B
Address	Via Panoramica 480 pal. B 98168 Messina		
Phone	090 2217178	Cell:--	
E-mail	omusumeci@unime.it		
Nationality	Italian		
DOB	19/01/1971		

Working experience

Date	- January 2022 to present
Type of work and position Role	Professor of Neurology, MED 12A, Department of Clinical and experimental Medicine, university of Messina, Messina Italy **Academic** - 2009-2021 University Researcher, MED/26 , Department of Clinical and experimental Medicine, University of Messina, Messina Italy -2009 to present Neurologist at University Hospital “AOU G. Martino”, Messina Italy
Main activities and responsabilities	- Medical doctor, specialist in Neurology. Expert in myopathology, clinical and diagnostic procedures in neuromuscular disorders. - Responsible of the in- and out-patients and DH activity for adult patients affected by rare neuromuscular disorders. - Responsible of the Regional Reference Center for Rare Neurological and Neuromuscular disorders **Academic activities as teacher in the following Programs** Medicine and Surgery Physical Sciences Psycological Sciences Residency in Neurology **Istitutional Academic responsabilities** Component of the phD commitee for Bioengineer Component of the Paritetic Commision of Neurophysiopatology Course Referent for Quality, Research and education for the DIMED at University of Messina
Name and Address of employer	Department of Clinical and Experimental Medicine, University of Messina via C. Valeria 1, Messina Italy
Type of business or Sector	Academy/HealthCare

Education	July 1995: **Degree in Medicine** from the University of Messina. Experimental thesis entitled: " Thyroid myopathies: biochemical and immunoistochemical studies of cytoskeletal proteins". Supervisor: Prof. C. Messina 1995-1999 **Residency in Neurology. Specialist in Neurology** (November 1999). Experimental thesis entitled "Mitochondrial encephalomyopathy due to Coenzyme Q10 deficiency: a clinical, morphological and biochemical study" 1998-1999 **Research fellowship,** Neuromuscular Group, Department of Neurology, "Houston Merritt Center of Muscular Dystrophy College of Physicians and Surgeons Department of Neurology" Prof. Salvatore Di Mauro, Columbia University di New York, USA 2000-2001 **Telethon fellowship** on Mitochondrial disorders (Columbia University, New York, USA) 2001-2003 **PhD** in "Neuromuscular disorders and genetic encephalo-neuro-myopathies", coordinator Prof. Giuseppe Vita, University of Messina
Personal skills and competences	Since 1994, she has been committed to providing specialized care to patients with neurological problems, particularly those suffering from rare neuromuscular diseases, attending the Neuropathology Unit of the University of Messina. She currently works in the Neurology and Neuromuscular Diseases Unit, providing inpatient care and performing on-call shifts, emergency services, and specialized outpatient services within the rare neuromuscular diseases clinic.
Language	English
Self evaluation	

Language	Comprehension		Oral		writing
Language	Listening	Reading	Oral interaction	Oral production	
	excellent	excellent	excellent	excellent	excellent

Social Competences	- Member of the Italian Society of Neurology (SIN) - Member of the Italian Association of Myology (AIM). - Member of the AIM Board of Directors as Secretary for the 2022-2024 three-year term and reappointed for the 2025-2027 three-year term. - Member of the World Muscle Society (WMS) - Member of the European Academy of Neurology (EAN). - Member of the EAN Management Panel for Rare Neurological Diseases - Member of the European Society for Mitochondrial Research and Medicine (E-MIT) and participated as a member of the organizing committee of the EUROMIT 2023 Congress held in Bologna in June 2023. - Member of the Medical-Scientific Commission of the Italian Ataxic Syndromes Association (AISA) - Member of the Medical-Scientific Commission of the Association for Mitochondrial Diseases (MITOCON) - Member of the Medical-Scientific Commission of the ARSACS Association - Orphanet contact for neuromuscular diseases, particularly mitochondrial diseases
Computer abilities	She can use Office, Adobe professional, Endnote and Canva

Other Skills

About research activity she is author and coauthorr of about 300 papers

Papers in extenso su riviste recensite da Current Contents/Life Sciences-Clinical Medicine 187

Scopus profile ID: 5,988 Citations 179 Documents H index 44
ORCID number: 0000-0002-9208-1527

The topics covered primarily concern the study of neuromuscular diseases, especially those of the muscle, such as metabolic myopathies and mitochondrial diseases. He studied these topics in depth, particularly with regard to the genetic-molecular definition of these diseases, at the Neuromuscular Group, Houston Merritt Center of Muscular Dystrophy, College of Physicians and Surgeons, Department of Neurology, directed by Professor Salvatore Di Mauro, Columbia University, New York, USA, where he attended for approximately two years. During this period, he conducted biochemical and genetic studies in the fields of mitochondrial encephalomyopathies, muscle glycogen storage diseases, and certain neurodegenerative diseases. In particular, he conducted a study on the pathogenesis of several families with autosomal recessive ataxic syndrome and cerebellar atrophy of unknown etiology, in which a marked deficiency of Coenzyme Q10 in the muscle was found. Over the years, she has focused on neuromuscular diseases, particularly metabolic myopathies such as Pompe disease and other muscle glycogen storage diseases, and hereditary neurodegenerative diseases such as familial spastic paraparesis and hereditary ataxias, both in research and clinical practice. She actively collaborates with several Italian and international centers on molecular characterization studies and genotype-phenotype correlations of these diseases.

Other Skills

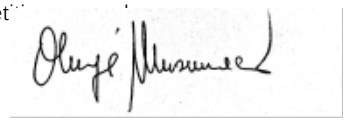
She was involved in the following Observational Studies:
2009-2014. **Principal investigator** for the project **GR-2009-1606283** " Recessive Spinocerebellar Ataxias: An integrated clinical, neuroradiological, biochemical and genetic approach for the identification of new phenotypes and possible biomarkers" supported by the **Italian Ministry of Health**.
2014-2016: **collaborator** in European project "European registry of patients with McArdle disease and very rare muscle glycogenolytic disorders (MGD) with exercise intolerance as the major symptom **(PR-MDMGD) (EUROMAC)" supported by European Commission**
2012-2015 **(Telethon): Collaborator** in the " Italian Register of Mitochondrial Diseases"
2014- 2016: **Collaborator in the Telethon** project " Building a Nation-wide Italian collaborative network for muscle glycogenoses: registry and natural history".
2024 to present **Principal investigator** - Centro Coordinatore Potential Prognostic Parameters in a Low/Paucisymptomatic Population with Late Onset Pompe Disease (LOPD)" –;
2023 to present **Collaborator** (PI Prof. J Diaz Manera NEWCASTLE) Transcribing Pompe: a full transcriptomics analysis of Pompe disease muscle biopsies.
2023 to present **Principal Investigator** "STOP-HSP.net: Storing a collection Of Patients with HSP – network" supported by **Telethon (GSP23001)**
2023 to present **Principal Investigator** Mitochondrial diseases: unraveling the clinical maze by omics and artificial intelligence, supported by MIUR and EU. **PRIN2022_ Cod 2022B9WY4A**
2024 to present **Principal Investigator THERAPY4ALL** - Towards therapies for mitochondrial diseases- **PNRR 2023** (Centro Coordinatore Università di Bologna)

She was involved in the followiong Clinical Trials
2013-2015 Local Study Coordinator for the clinical trial, Sanofy N": TDR12857, An open-label, multicenter, multinational, ascending dose study of the safety, pharmacokinetics, pharmacodynamics, Ind exploratory efficacy of repeated biweekly infusions of neoGAA in naïve and alglucosidase alfa treated late-onset Pompe disease patients"
2013 -2014 Local Study Coordinator for the clinical trial "A Phase 3 Switchover Study of the Efficacy and Safety of BMN 701 (GILT-tagged Recombinant Human GAA) in rhGAA Exposed Subjects with Late-onset Pompe Disease", Biomarin Pharmaceutical
2015-2017 : collaborator in Study "Historical Case Record Survey of Visual Acuity Data from Patients with Leber's Hereditary Optic Neuropathy (LHON)" Santhera Pharmaceuticals SNT-CRS-002

2019 -2020 Local Study Coordinator for the clinical trial . A Phase 3 Randomized, Double-Blind, Parallel-Group, Placebo-Controlled Trial to Evaluate the Efficacy and Safety of Daily Subcutaneous Injections of Elamipretide in Subjects with Primary Mitochondrial Myopathy Followed by an Open-Label Treatment Extension Prot. 107/18
2016-2023 Local Study Coordinator for the clinical trial "A Phase 3 Randomized, Multicenter, Multinational, Double-blinded Study Comparing the Efficacy and Safety of Repeated Biweekly Infusions of NeoGAA (GZ402666) and Alglucosidase Alfa in Treatment-naïve Patients With Late-onset Pompe Disease. Study EFC14028 COMET
2019-2020 A phase 3 double-blind randomized study to assess the efficacy and safety of intravenous ATB200 co-administered with oral AT2221 in adult subjects with late-onset pompe disease compared with alglucosidase alfa/placebo Protocol Number: ATB200-03 Prot. 01/19
2019-2020 A Multi-Center, Low Interventional Study with a Retrospective Component in Participants with Late Onset Pompe Disease (SPK-GAA-100) Prot. 30/19 (approvato 17/06/2019)
2019 to present Dieta chetogenica nella malattia di McArdle: Trial multicentrico randomizzato Prot. 66/19
2020 to present Subinvestigator in A phase 3 Open-Label-Extension Study to Assess the Long-term Safety and Efficacy of intravenous ATB200 Co-administrated with Oral AT2221 in Adult Subjects with Late-onset Pompe Disease (ATB200-07- OLE Study)
2021-2022 Principal Investigator in A Prospective, Multicenter, Non-Interventional Study To Investigate The Disease Characteristics Of Adult Patients With Long Chain Fatty Acid Oxidation Disorders (FAOD)" (REN001-903)
2021-2022 Principal Investigator in Double-Blind, Placebo-Controlled, Study To Evaluate The Efficacy and Safety of 24 weeks treatment with REN001 in patients with primary mitochondrial myopathy (PMM) (REN001-201)
2021-2022 A Retrospective Study of Subjects With Thymidine Kinase 2 Deficiency Treated With the Combination of Pyrimidine Nucleos(t)ides as well as Untreated Subjects to Collect Vital Status Data and Supporting Information . (MT-1621-107)
2022-2023 Principal Investigator An Open-Label, Multi-Centre Study To Evaluate The Long-Term Safety And Tolerability Of REN001 In Subjects With Primary Mitochondrial Myopathy (PMM)", Prot. V1.0 dated 04 Aug 2021 REN001-202 EudraCT Code 2021-003471-34
2022 to present Principal Investigator Phase III clinical trial "A Phase 3 Randomized, Double-Blind, Parallel-Group, Placebo-Controlled Trial to Evaluate the Efficacy and Safety of Daily Subcutaneous Injections of Elamipretide in Subjects with Primary Mitochondrial Disease Resulting from Pathogenic Nuclear DNA Mutations (nPMD)", Prot. No. SPIMD-301, EudraCT code 2021-003907-16
2023 to present Principal Investigator A Phase 2a Safety, Tolerability, and Pharmacodynamic Study of OMT-28 in PMD patients with myopathy and/or cardiomyopathy and inflammation (PMD-OPTION) Study Code: OMT28-C0203
2023-2024 Principal Investigator TK0110 A non-interventional study to collect supplemental information from participants with thymidine kinase 2 deficiency in the doxecitine and doxribtimine clinical studies
2024 to present Principal Investigator KL1333 2020-104. An adaptive, interventional, randomized, double-blind, parallel-group, placebo-controlled, flexible-dose study of the efficacy of KL1333 in adult patients with primary mitochondrial disease

The personal data collected will be treated in accordance with the principles and provisions of Legislative Decree no. 196/2003 regarding the protection of confidentiality and then only for the purpose of managing the compet

Date 15/07/2025



Signature Olimpia Musumeci _____

Publications

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- Mancuso, M., Lamperti, C., & **Musumeci, O.** (2025). National diagnostic gaps for TK2 Deficiency in Italy: insights from the AIM Multicenter Survey. Acta myologica : myopathies and cardiomyopathies : official journal of the Mediterranean Society of Myology, 44(3), 93–95. https://doi.org/10.36185/2532-1900-1424
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Servidei S, Bris C, Cano A, Furlan F, Gasperini S, Laborde N, Lamperti C, Lenz D, Mancuso M, Montano V, Menni F, **Musumeci O,** Nesbitt V, Procopio E, Rouzier C, Staufner C, Taanman JW, Tal G, Ticci C, Cordelli DM, Carelli V, Procaccio V, Prokisch H, Garone C. Deoxyguanosine kinase deficiency: natural history and liver transplant outcome. Brain Commun. 2024 May 6;6(3):fcae160. doi: 10.1093/braincomms/fcae160.

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