

Prof. Gabriele Siciliano

Curriculum vitae et studiorum

Date and place of birth:

ACADEMICS AND PROFESSIONALS

Degree in Medicine:

University of Pisa, 27/6/1980, cum laude

Thesis: *"Modifications of kidney function in patients affected by valvular cardiopathy after surgical defect correction"*.

Registration/Medical License Number:

Physicians register of Pisa, Inscr. N. 2167 – 20 November 1981

Specialization in Neurology:

University of Pisa, 15/11/1984, cum laude

Thesis: *"Methods of clinical evaluation of motor function in Duchenne Muscular Dystrophy"*

Specialization in Physical and Rehabilitation Medicine:

University of Pisa, 21/7/1992

Thesis: *"Muscle metabolic aspects of fatigue in normals as evaluated by means of Phosphorus31 Spectroscopic Nuclear Magnetic Resonance during submaximal contraction"*.

PhD in Neurological and Neurosensorial Sciences (University of Ancona, 2/10/1990)

Thesis: *"Central and peripheral mechanisms of muscle fatigue in normals and in neuromuscular diseases"*.

Post-Doctoral Research in Neurosciences:

University of Ancona, from 16/9/1991 to 15/9/1993

Neurologist Assistant Register:

at Clinica Neurologica, University of Pisa, from 1993 to 1997.

University Researcher

Department of Neuroscience, University of Pisa from 1997 to 2001

University Associated Professor in Neurology (MED/26)

Department of Clinical and Experimental Medicine, University of Pisa from 2001 to 2015

University Full Professor in Neurology (MED/26)

Department of Clinical and Experimental Medicine, University of Pisa from 2016

Present position:

Full Professor in Neurology and Head of Clinical Centre for Neuromuscular Diseases and Head of the Laboratory for Molecular Diagnosis, Neurological Clinic, Department of Neuroscience, Azienda Ospedaliero Universitaria Pisana.

FORMATIVE STAGES IN ITALY AND OVERSEAS

- 1983 Fellowship of Unione Italiana Lotta alla Distrofia Muscolare, 9 months, Laboratory of Neuropathology, University of Padova, argument: "Muscle histopathology in Neuromuscular Diseases"
- 1988 PhD Fellow, 6 months, Laboratory of Neuropathology, University of Padova, argument: "Biochemistry investigations in the diagnosis of Neuromuscular Diseases"
- 1989 PhD Fellow and "Guest Researcher", 8 months, Muscle Centre, Department of Medicine, Royal University Hospital, Liverpool (GB), argument: "Neurophysiopathologic techniques in the assessment of muscle fatigue in Neuromuscular Diseases"
- 1992 Post-doc Fellow, 1 month, Department of Biology, University of Padova, argument: "Methods of genetic and molecular analyses in the diagnosis of Muscular Dystrophies"
- 1992 Post-doc Fellow and "Guest Researcher", 2 months, Nuclear Magnetic Resonance Centre, University of Liverpool (GB), argument: "Magnetic Resonance Spectroscopic methods in the study of muscle function"
- 1999 Visiting researcher, Department of Neurology, Columbia University, New York (USA)

MEMBERSHIP IN SCIENTIFIC SOCIETIES

- Italian Neurology Society, since 1984;
- Italian Neuropathology Association, since 1985 (Advisory Board since 2001);
- Italian Clinical Neurophysiology Society, since 1989;
- International Society of Neuropathology, since 1990;
- World Society of Neurology, since 1990;
- Royal Society of Medicine, Section of Neurology, since 1991;
- International Society of Electromyography and Kinesiology, since 1992;
- World Society of Myology, since 1996;
- American Association for the Advancement of Science, since 1998;
- Italian Myology Association, since 2000.
- European Amyotrophic Lateral Sclerosis Consortium, since 2001
- European Federation of Neurological Societies, since 2012
- President of Italian Association of Myology (AIM), since 2015 to date

EDITORIAL BOARD OF INTERNATIONAL JOURNALS IN NEUROLOGIC AREA

- Bioscience Reports, Kluwer Academic/Plenum Publishers, ISSN 0144-8463
- Basic and Applied Myology, Unipress, ISSN: 1120-9992
- Journal of Alzheimers Disease, IOS Press, ISSN:1387-2877 (Print) ; 1875-8908 (Electronic)
- Nutrition, Tarrytown, NY : Elsevier Science, 0899-9007 (Print) (dal novembre 2010)

B) Editorial board di numeri speciale/raccolte su riviste internazionali in ambito neurologico

- Neurological Sciences Supplement- Perspectives In Molecular Therapy On Muscle Diseases, Springer-Verlag Italia, Issn:1590-1874 (Print)
- Bioscience Reports SUPPLEMENT- Mitochondrial Diseases: Advances In Understanding and

- CNS Neurol Disord Drug Targets SUPPLEMENT- Neuroprotection In ALS: From Pathology To Treatment, Bentham Science Publishers, 1871-5273 (Print)
- Neuromuscular Disorders SUPPLEMENT- Muscle Fatigue in Neuromuscular Disorders: Pathogenic Mechanisms and Treatment, Pergamon Press, ISSN:0960-8966 (Print)

REFeree OF INTERNATIONAL JOURNALS IN NEUROLOGIC AREA

- Acta Neurologica Scandinavica, Wiley-Blackwell, ISSN: 0001-6314
- Amyotrophic Lateral Sclerosis and other Motor Neuron Disorders, Martin Dunitz, ISSN: 1466-
- Annals of Neurology, Wiley-Liss, ISSN: 0364-5134 (Print)
- Antioxidant and Redox Signaling, Mary Ann Liebert, Inc., ISSN: 1523-0864 (Print)
- Archives of Neurology, American Medical Assn, ISSN: 0003-9942
- Behavioural Brain Research, Biomedical Press, ISSN: 0166-4328 (Print)
- Biomarkers in Medicine, Future Medicine, ISSN: 1752-0363 (Print)
- Brain , Oxford University Press, ISSN: 0006-8950
- Brain Research Bulletin, Elsevier, ISSN 0361-9230
- Clinical and Experimental Rheumatology, S.A.S., ISSN 0392-856X
- Clinical Investigation, Future Science, ISSN: 2041-6792 (Print)
- Drug Design, Development and Therapy, Dove Press Limited, ISSN:1177-8881 (Electronic)
- European Journal of Neurology, Blackwell Science, ISSN: 1351-5101 (Print)
- European Journal of Neurology, Blackwell, ISSN 1351-5101
- Expert Opinion on Orphan Drugs
- Expert Opinion on Medical Diagnostics, Informa Healthcare, ISSN: 1753-0059 (Print)
- Expert Opinion on Therapeutic Targets, Informa Healthcare, ISSN: 1472-8222 (Print)
- Functional Neurology, Cic Edizioni Internazionali S.R.L, ISSN: 0393-5264 (Print)
- Gene, Elsevier, ISSN: 0378-1119
- Journal of Medical Genetics, British Medical Association, SSN:0022-2593 (Print)
- Journal of Neural Transmission, Springer, ISSN: 0300-9564 (print version)
- Journal of Neurology, Neurosurgery and Psychiatry, BMJ Publishing Group, ISSN: 0022-3050
- Journal of Neurology, Springer, ISSN 0340-5354
- Journal of Neuroscience Research, Wiley Interscience, ISSN: 0360-4012
- Journal of Neurosciences Methods, Elsevier, ISSN: 0165-0270
- Journal of Pharmacy and Pharmacology, Wiley, ISSN:0022-3573 (Print)
- Life Science, Pergamon-Elsevier, ISSN 0024-3205
- Medicinal chemistry, Omics Pub. Group, ISSN: 2161-0444 (Electronic)
- *Mini Reviews in Medicinal Chemistry, Bentham Science, c2001-ISSN: 1389-5575 (Print)*
- Mitochondrion, Elsevier Science, ISSN: 1567-7249
- Molecular neurodegeneration, BioMed Central, ISSN: 1750-1326 (Electronic)
- Muscle and Nerve, John Wiley & Sons, ISSN: 0148-639X (Print)
- Neurochemistry, Pergamon Press, ISSN:0197-0186 (Print)
- Neurological research, Maney Pub, ISSN:0161-6412 (Print)
- Neurological Sciences, Springer-Verlag Italia, ISSN: 1590-1874
- Neurological Sciences, Springer-Verlag Italia, ISSN:1590-1874 (Print)
- Neurology, Lippincott Williams & Wilkins, ISSN: 0028-3878
- Neuromuscular Disorders, Pergamon, Pergamon Press, ISSN:0960-8966 (Print)
- Neuropathology and Applied Neurobiology, Blackwell Scientific, ISSN: 0305-1846 (Print)
- Neurotoxicology, Elsevier Science, ISSN: 0161-813X (Print)
- Nutrition journal, BioMed Central, ISSN:1475-2891 (Electronic)
- Orphanet Journal of Rare Diseases, [London] : BioMed Central, 2006-, ISSN: 1750-1172 (Electronic)
- Redox Report: Churchill Livingstone, ISSN: 1351-0002 (Print)

RESEARCH ACTIVITIES AND INTERESTS

Since 1993 he is responsible for the Unit of Neuromuscular Diseases and for the Laboratory of Molecular Diagnosis in the former Department of Neuroscience, now Department of Clinical and Experimental Medicine - Neurological Clinics - University of Pisa, with main interests in clinical, laboratory and molecular aspects of neurodegenerative and neurogenetic disorders, acquired and genetic diseases of peripheral nervous system and skeletal muscle. These activities are conducted in collaboration with some overseas centres and laboratories. Since 1986 he has been engaged in studies on muscular dystrophies, metabolic diseases of muscle and motorneuron diseases. In particular, he has studied several issues on muscle fatigue, in collaboration with the Muscle Research Centre, University of Liverpool (Prof. RHT Edwards), exercise in normal and myopathic subjects, motorneuron output evaluation in amyotrophic lateral sclerosis, subsarcolemmal excitability in myotonic syndromes and muscular dystrophies, analysis of metabolic parameters in McArdle's disease and mitochondrial myopathies. In the same period he has been engaged in epidemiological and therapeutical studies on neuromuscular disorders, in particular the use of selenium in Steinert disease, idebenone in mitochondrial myopathies, mexiletine in congenital myotonia, TRH and riluzole in amyotrophic lateral sclerosis. As far as mitochondrial myopathies are concerned, he is currently studying the effects of aerobic training programs, the relationship between metabolic and catecholaminergic systems during exercise, the pathogenic role of mitochondrial transcription factors, the diagnostic usefulness of magnetic resonance spectroscopic techniques. Furthermore, he is working on the possible role of mitochondrial involvement and oxidative stress in the pathogenesis of neurodegenerative disorders. All these activities have led to a scientific production volume represented by near 300 full papers, in specialized cited international journals, dealing with the above indicated arguments, funded projects from Institutional and Charity bodies, responsibility in research evaluation committees for public and private funding research bodies, participation to national and international clinical trials performed according to GCP (last GCP training certificate obtained in 19 February 2016). More recent ongoing GCP studies as PI: CFTY720I2201, IGD002, 161403, SNT-CRS-002

PARTICIPATION IN CLINICAL TRIALS (LAST 5 YEARS) as PI

- 1) Phase III Efficacy, Safety, and Tolerability Study of HYQVIA/HyQvia and GAMMAGARD LIQUID/KIOVIG in CIDP -161403, ongoing
- 2) Long-Term Tolerability and Safety of HYQVIA/HyQvia in CIDP-161505, as PI , ongoing
- 3) A Non-interventional Study of Clinical Experience in Patients Prescribed Raxone® for the Treatment of Leber's Hereditary Optic Neuropathy (LHON), SNT-IV-003
- 4) Effects of oral levosimendan (ODM-109) on respiratory function in patients with ALS-3119002, center activation
- 5) A Double-blind, Randomized, Multicenter, Placebo-controlled, Parallel-group Study to Evaluate the Efficacy and Safety of Fingolimod 0.5 mg Administered Orally Once Daily Versus Placebo in Patients With Chronic Inflammatory Demyelinating Polyradiculoneuropathy (CIDP)-FTY720
- 6) Study to assess the effectiveness of high doses of immunoglobulin in the treatment of polyneuropathy painful diabetic resistant to conventional therapies. –IGD002

PUBLICATIONS

1: Daniele S, Pietrobono D, Fusi J, Lo Gerfo A, Cerri E, Chico L, Iofrida C, Petrozzi L, Baldacci F, Giacomelli C, Galetta F, Siciliano G, Bonuccelli U, Trincavelli ML, Franzoni F, Martini C. α -Synuclein Aggregated with Tau and β -Amyloid in Human Platelets from Healthy Subjects: Correlation with Physical Exercise. *Front Aging Neurosci.* 2018 Jan 30;10:17. doi: 10.3389/fnagi.2018.00017. eCollection 2018. PubMed PMID: 29441013; PubMed Central PMCID: PMC5797553.

2: Chico L, Modena M, Lo Gerfo A, Ricci G, Caldarazzo Ienco E, Ryskalin L, Fornai F, Siciliano G. Cross-talk between pathogenic mechanisms in neurodegeneration: the role of oxidative stress in Amyotrophic Lateral Sclerosis. *Arch Ital Biol.* 2017 Dec 1;155(4):131-141. doi: 10.12871/00039829201744. PubMed PMID: 29405030.

3: Cassandrini D, Trovato R, Rubegni A, Lenzi S, Fiorillo C, Baldacci J, Minetti C, Astrea G, Bruno C, Santorelli FM; Italian Network on Congenital Myopathies. Congenital myopathies: clinical phenotypes and new diagnostic tools. *Ital J Pediatr.* 2017 Nov 15;43(1):101. doi: 10.1186/s13052-017-0419-z. Review. PubMed PMID: 29141652; PubMed Central PMCID: PMC5688763.

4: Donatelli G, Retico A, Caldarazzo Ienco E, Cecchi P, Costagli M, Frosini D, Biagi L, Tosetti M, Siciliano G, Cosottini M. Semiautomated Evaluation of the Primary Motor Cortex in Patients with Amyotrophic Lateral Sclerosis at 3T. *AJNR Am J Neuroradiol.* 2018 Jan;39(1):63-69. doi: 10.3174/ajnr.A5423. Epub 2017 Nov 9. PubMed PMID: 29122765.

5: Mancuso M, McFarland R, Klopstock T, Hirano M; consortium on Trial Readiness in Mitochondrial Myopathies. International Workshop:: Outcome measures and clinical trial readiness in primary mitochondrial myopathies in children and adults. Consensus recommendations. 16-18 November 2016, Rome, Italy. *Neuromuscul Disord.* 2017 Dec;27(12):1126-1137. doi: 10.1016/j.nmd.2017.08.006. Epub 2017 Sep 8. PubMed PMID: 29074296.

6: Vergallo A, Giampietri L, Baldacci F, Volpi L, Chico L, Pagni C, Giorgi FS, Ceravolo R, Tognoni G, Siciliano G, Bonuccelli U. Oxidative Stress Assessment in Alzheimer's Disease: A Clinic Setting Study. *Am J Alzheimers Dis Other Dement.* 2018 Feb;33(1):35-41. doi: 10.1177/1533317517728352. Epub 2017 Sep 21. PubMed PMID: 28931301.

7: Barone V, Del Re V, Gamberucci A, Polverino V, Galli L, Rossi D, Costanzi E, Toniolo L, Berti G, Malandrini A, Ricci G, Siciliano G, Vattemi G, Tomelleri G, Pierantozzi E, Spinozzi S, Volpi N, Fulceri R, Battistutta R, Reggiani C, Sorrentino V. Identification and characterization of three novel mutations in the

CASQ1 gene in four patients with tubular aggregate myopathy. *Hum Mutat.* 2017 Dec;38(12):1761-1773. doi: 10.1002/humu.23338. Epub 2017 Sep 26. PubMed PMID: 28895244.

8: Aimo A, Giannoni A, Castiglione V, Mancuso M, Siciliano G, Piepoli MF, Passino C, Emdin M. Neurohormonal modulation for treatment of cardiac involvement in dystrophinopathies and mitochondrial disease. *Eur J Prev Cardiol.* 2017 Nov;24(16):1718-1724. doi: 10.1177/2047487317725018. Epub 2017 Aug 14. PubMed PMID: 28805065.

9: Bella ED, Tramacere I, Antonini G, Borghero G, Capasso M, Caponnetto C, Chiò A, Corbo M, Eleopra R, Filosto M, Giannini F, Granieri E, Bella V, Lunetta C, Mandrioli J, Mazzini L, Messina S, Monsurrò MR, Mora G, Riva N, Rizzi R, Siciliano G, Silani V, Simone I, Sorarù G, Volanti P, Lauria G. Protein misfolding, amyotrophic lateral sclerosis and guanabenz: protocol for a phase II RCT with futility design (PromISE trial). *BMJ Open.* 2017 Aug 11;7(8):e015434. doi: 10.1136/bmjopen-2016-015434. PubMed PMID: 28801400; PubMed Central PMCID: PMC5724081.

10: Vergallo A, Carlesi C, Pagni C, Giorgi FS, Baldacci F, Petrozzi L, Ceravolo R, Tognoni G, Siciliano G, Bonuccelli U. A single center study: A β 42/p-Tau(181) CSF ratio to discriminate AD from FTD in clinical setting. *Neurol Sci.* 2017 Oct;38(10):1791-1797. doi: 10.1007/s10072-017-3053-z. Epub 2017 Jul 19. PubMed PMID: 28726050.

11: Baldanzi S, Ricci G, Bottari M, Chico L, Simoncini C, Siciliano G. The proposal of a clinical protocol to assess central and peripheral fatigue in myotonic dystrophy type 1. *Arch Ital Biol.* 2017 Jul 1;155(1-2):43-53. doi: 10.12871/000398292017125. PubMed PMID: 28715597.

12: Chico L, Ricci G, Cosci O Di Coscio M, Simoncini C, Siciliano G. Physical exercise and oxidative stress in muscular dystrophies: is there a good balance? *Arch Ital Biol.* 2017 Jul 1;155(1-2):11-24. doi: 10.12871/000398292017122. Review. PubMed PMID: 28715594.

13: Orsucci D, Angelini C, Bertini E, Carelli V, Comi GP, Federico A, Minetti C, Moggio M, Mongini T, Santorelli FM, Servidei S, Tonin P, Ardisson A, Bello L, Bruno C, Ienco EC, Diodato D, Filosto M, Lamperti C, Moroni I, Musumeci O, Pegoraro E, Primiano G, Ronchi D, Rubegni A, Salvatore S, Sciacco M, Valentino ML, Vercelli L, Toscano A, Zeviani M, Siciliano G, Mancuso M. Revisiting mitochondrial ocular myopathies: a study from the Italian Network. *J Neurol.* 2017 Aug;264(8):1777-1784. doi: 10.1007/s00415-017-8567-z. Epub 2017 Jul 10. PubMed PMID: 28695364.

- 14: Simoncini C, Siciliano G, Tognoni G, Mancuso M. Mitochondrial ANT-1 related adPEO leading to cognitive impairment: is there a link? *Acta Myol.* 2017 Mar;36(1):25-27. PubMed PMID: 28690391; PubMed Central PMCID: PMC5479106.
- 15: Stocco A, Siciliano G, Migliore L, Coppedè F. Decreased Methylation of the Mitochondrial D-Loop Region in Late-Onset Alzheimer's Disease. *J Alzheimers Dis.* 2017;59(2):559-564. doi: 10.3233/JAD-170139. PubMed PMID: 28655136.
- 16: Baldanzi S, Ricci G, Simoncini C, Cosci O Di Coscio M, Siciliano G. Hard ways towards adulthood: the transition phase in young people with myotonic dystrophy. *Acta Myol.* 2016 Dec;35(3):145-149. Review. PubMed PMID: 28484315; PubMed Central PMCID: PMC5416743.
- 17: Siciliano G, Santorelli FM, Battini R. EDITORIAL. *Acta Myol.* 2016 Dec;35(3):121. PubMed PMID: 28484311; PubMed Central PMCID: PMC5416738.
- 18: Daniele S, Pietrobono D, Fusi J, Iofrida C, Chico L, Petrozzi L, Gerfo AL, Baldacci F, Galetta F, Siciliano G, Bonuccelli U, Santoro G, Trincavelli ML, Franzoni F, Martini C. α -Synuclein Aggregates with β -Amyloid or Tau in Human Red Blood Cells: Correlation with Antioxidant Capability and Physical Exercise in Human Healthy Subjects. *Mol Neurobiol.* 2017 Apr 18. doi: 10.1007/s12035-017-0523-5. [Epub ahead of print] PubMed PMID: 28421539.
- 19: Giannoni A, Aimo A, Mancuso M, Piepoli MF, Orsucci D, Aquaro GD, Barison A, De Marchi D, Taddei C, Cameli M, Raglianti V, Siciliano G, Passino C, Emdin M. Autonomic, functional, skeletal muscle, and cardiac abnormalities are associated with increased ergoreflex sensitivity in mitochondrial disease. *Eur J Heart Fail.* 2017 Dec;19(12):1701-1709. doi: 10.1002/ejhf.782. Epub 2017 Feb 24. PubMed PMID: 28233467.
- 20: Stocco A, Tannorella P, Salluzzo MG, Ferri R, Romano C, Nacmias B, Siciliano G, Migliore L, Coppedè F. The Methylenetetrahydrofolate Reductase C677T Polymorphism and Risk for Late-Onset Alzheimer's disease: Further Evidence in an Italian Multicenter Study. *J Alzheimers Dis.* 2017;56(4):1451-1457. doi: 10.3233/JAD-161081. PubMed PMID: 28211809.
- 21: Mancuso M, Orsucci D, Angelini C, Bertini E, Bruno C, Carelli V, Comi GP, Filosto M, Lamperti C, Moggio M, Mongini T, Moroni I, Tonin P, Toscano A, Siciliano G; Nation-wide Italian Collaborative Network of Mitochondrial Diseases. Corrigendum to "Response to: Mitochondrial neuropathy affects peripheral and cranial nerves and is primary or secondary or both" [*Neuromuscular Disorders* 26/8 (2016) 549]. *Neuromuscul Disord.* 2017 Apr;27(4):e1. doi:

10.1016/j.nmd.2017.01.003. Epub 2017 Feb 8. PubMed PMID: 28189480.

22: Lucchesi C, Baldacci F, Cafalli M, Dini E, Siciliano G, Bonuccelli U, Gori S. P041. Analysis of body mass index, psychiatric comorbidity, sleep-wake pattern and occurrence of fatigue in episodic and chronic migraine patients. *J Headache Pain*. 2015 Dec;16(Suppl 1):A188. doi: 10.1186/1129-2377-16-S1-A188. PubMed PMID: 28132200; PubMed Central PMCID: PMC4715196.

23: Da Pozzo P, Cardaioli E, Rubegni A, Gallus GN, Malandrini A, Rufa A, Battisti C, Carluccio MA, Rocchi R, Giannini F, Bianchi A, Mancuso M, Siciliano G, Dotti MT, Federico A. Novel POLG mutations and variable clinical phenotypes in 13 Italian patients. *Neurol Sci*. 2017 Apr;38(4):563-570. doi: 10.1007/s10072-016-2734-3. Epub 2017 Jan 27. PubMed PMID: 28130605.

24: Train the Brain Consortium. Randomized trial on the effects of a combined physical/cognitive training in aged MCI subjects: the Train the Brain study. *Sci Rep*. 2017 Jan 3;7:39471. doi: 10.1038/srep39471. PubMed PMID: 28045051; PubMed Central PMCID: PMC5206718.

25: Costagli M, Donatelli G, Biagi L, Caldarazzo Ienco E, Siciliano G, Tosetti M, Cosottini M. Magnetic susceptibility in the deep layers of the primary motor cortex in Amyotrophic Lateral Sclerosis. *Neuroimage Clin*. 2016 May 2;12:965-969. eCollection 2016. PubMed PMID: 27995062; PubMed Central PMCID: PMC5153607.

26: Magri F, Nigro V, Angelini C, Mongini T, Mora M, Moroni I, Toscano A, D'angelo MG, Tomelleri G, Siciliano G, Ricci G, Bruno C, Corti S, Musumeci O, Tasca G, Ricci E, Monforte M, Sciacco M, Fiorillo C, Gandossini S, Minetti C, Morandi L, Savarese M, Fruscio GD, Semplicini C, Pegoraro E, Govoni A, Brusa R, Del Bo R, Ronchi D, Moggio M, Bresolin N, Comi GP. The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. *Muscle Nerve*. 2017 Jan;55(1):55-68. doi: 10.1002/mus.25192. Epub 2016 Oct 28. PubMed PMID: 27184587.

27: Mancuso M, Montano V, Orsucci D, Peverelli L, Caputi L, Gambaro P, Siciliano G, Lamperti C. Mitochondrial m.3243A > G mutation and carotid artery dissection. *Mol Genet Metab Rep*. 2016 Sep 1;9:12-4. doi: 10.1016/j.ymgmr.2016.08.010. eCollection 2016 Dec. PubMed PMID: 27656415; PubMed Central PMCID: PMC5021764.

28: Fattori B, Siciliano G, Mancini V, Bastiani L, Bongioanni P, Caldarazzo Ienco E, Barillari MR, Romeo SO, Nacci A. Dysphagia in Amyotrophic Lateral Sclerosis: Relationships between disease progression and Fiberoptic Endoscopic Evaluation of Swallowing. *Auris Nasus Larynx*. 2017 Jun;44(3):306-312. doi: 10.1016/j.anl.2016.07.002. Epub 2016 Aug 25. PubMed PMID: 27569290.

- 29: Lucchesi C, Caldarazzo Ienco E, Fabbrini M, Pasquali L, Lo Gerfo A, Fogli A, Siciliano G. Amyotrophic lateral sclerosis with long lasting disease course and SOD1 and TARDBP mutations: Report of two cases and overview of the literature. *Amyotroph Lateral Scler Frontotemporal Degener.* 2017 Feb;18(1-2):137-139. doi: 10.1080/21678421.2016.1212896. Epub 2016 Aug 5. PubMed PMID: 27494151.
- 30: Caldarazzo Ienco E, Orsucci D, Simoncini C, Montano V, LoGerfo A, Siciliano G, Bonuccelli U, Mancuso M. Acute encephalopathy of the temporal lobes leading to m.3243A>G. When MELAS is not always MELAS. *Mitochondrion.* 2016 Sep;30:148-50. doi: 10.1016/j.mito.2016.07.008. Epub 2016 Jul 21. PubMed PMID: 27453332.
- 31: Baldanzi S, Cecchi P, Fabbri S, Pesaresi I, Simoncini C, Angelini C, Bonuccelli U, Cosottini M, Siciliano G. Relationship between neuropsychological impairment and grey and white matter changes in adult-onset myotonic dystrophy type 1. *Neuroimage Clin.* 2016 Jun 15;12:190-7. doi: 10.1016/j.nicl.2016.06.011. eCollection 2016. PubMed PMID: 27437180; PubMed Central PMCID: PMC4939389.
- 32: di Russo P, Perrini P, Benedetto N, Siciliano G. Phenytoin-induced rhabdomyolysis: Timely recognition for safely remission. *Neurol India.* 2016 Jul-Aug;64(4):793-4. doi: 10.4103/0028-3886.185415. PubMed PMID: 27381132.
- 33: Simoncini C, Chico L, Concolino D, Sestito S, Fancellu L, Boadu W, Sechi GP, Feliciani C, Gnarra M, Zampetti A, Salviati A, Scarpelli M, Orsucci D, Bonuccelli U, Siciliano G, Mancuso M. Mitochondrial DNA haplogroups may influence Fabry disease phenotype. *Neurosci Lett.* 2016 Aug 26;629:58-61. doi: 10.1016/j.neulet.2016.06.051. Epub 2016 Jun 27. PubMed PMID: 27365132.
- 34: Imbrici P, Altamura C, Camerino GM, Mangiatordi GF, Conte E, Maggi L, Brugnoli R, Musaraj K, Caloiero R, Alberga D, Marsano RM, Ricci G, Siciliano G, Nicolotti O, Mora M, Bernasconi P, Desaphy JF, Mantegazza R, Camerino DC. Multidisciplinary study of a new ClC-1 mutation causing myotonia congenita: a paradigm to understand and treat ion channelopathies. *FASEB J.* 2016 Oct;30(10):3285-3295. Epub 2016 Jun 20. PubMed PMID: 27324117; PubMed Central PMCID: PMC5024700.
- 35: Savarese M, Di Fruscio G, Torella A, Fiorillo C, Magri F, Fanin M, Ruggiero L, Ricci G, Astrea G, Passamano L, Ruggieri A, Ronchi D, Tasca G, D'Amico A, Janssens S, Farina O, Mutarelli M, Marwah VS, Garofalo A, Giugliano T, Sampaolo S, Del Vecchio Blanco F, Esposito G, Piluso G, D'Ambrosio P, Petillo R, Musumeci O, Rodolico C, Messina S, Evilä A, Hackman P, Filosto M, Di Iorio G, Siciliano G, Mora M, Maggi L, Minetti C, Sacconi S, Santoro L, Claes K, Vercelli L, Mongini T, Ricci E, Gualandi F, Tupler R, De Bleecker J, Udd B, Toscano A, Moggio M,

Pegoraro E, Bertini E, Mercuri E, Angelini C, Santorelli FM, Politano L, Bruno C, Comi GP, Nigro V. The genetic basis of undiagnosed muscular dystrophies and myopathies: Results from 504 patients. *Neurology*. 2016 Jul 5;87(1):71-6. doi: 10.1212/WNL.0000000000002800. Epub 2016 Jun 8. PubMed PMID: 27281536; PubMed Central PMCID: PMC4932234.

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Invited speaker, seminars, lectures and/or chairmanships in International and National Congresses: more than 300

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Pisa, 02 March 2018

Prof. Gabriele Siciliano