

Pennuto, Maria, PhD

Maria Pennuto, PhD

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Work Experience

Associate Professor 2017-present
Pathogenesis of Neurological and Neurodegenerative Diseases Unit
Department of Biomedical Sciences (DBS), University of Padova, Padova, Italy

Assistant and then Associate Professor 2013-2017
Head of the Dulbecco Telethon Institute Lab of Neurodegenerative Diseases
Centre for Integrative Biology (CIBIO), University of Trento, Trento, Italy

Group Leader 2009-2013
Head of Motor Neuron Disease Unit
Department of Neuroscience and Brain Technologies, Italian Institute of Technology, Genoa, Italy

Staff Scientist 2008- 2009
Department of Neurology, University of Pennsylvania, Philadelphia, PA, USA
Experimental work on "Molecular mechanisms of neurodegeneration in polyglutamine and neuromuscular diseases", Mentor: Dr J. Paul Taylor, MD, PhD

Senior Post-Doctoral Visiting Fellow (Genetics) 2005-2007
National Institute for Neurological Disorders and Stroke (NINDS), National Institutes of Health, Bethesda, MD, USA
Experimental work on "Molecular mechanisms of spinal and bulbar muscular atrophy", Mentor: Dr Kenneth Fischbeck, MD

Post-Doctoral Fellow (Mouse Genetics) 2001-2004
S. Raffaele Scientific Institute, Milan, Italy
Experimental work on "Protein quality control in the pathogenesis of Charcot-Marie-Tooth 1B", Mentor: Dr Lawrence Wrabetz, MD

Education

Full Professor Habilitation (*venia legendi*) 2017
General Pathology & Clinical Pathology (SSD: MED/04/05/46/02), Ministry of Education, Universities and Research (MIUR), Italy

Associate Professor Habilitation (*venia legendi*) 2014
Molecular Biology (SSD: BIO/11), Ministry of Education, Universities and Research (MIUR), Italy

Doctor of Philosophy (Molecular and Cellular Biology) 1997-2000

Pennuto, Maria, PhD

University of Milan, Milan, Italy

Experimental thesis work: "Single-synapse analysis of protein-protein interactions in live neurons", Mentor: Prof Flavia Valtorta, MD

Master in Science (Diploma, Biological Sciences)

1990-1996

University La Sapienza, Rome, Italy

Experimental thesis work: "NGF-dependent and tissue-specific transcription of vgf", Mentor: Dr Sergio Nasi, PhD

Honors and Awards

- 2017 Arimura Foundation State-of-Art Lecture Award, 13th International PACAP meeting, Hong Kong.
- 2013 Dulbecco Telethon Institute Career Award
- 2011 IBRO selected Lecture "Woman in Neuroscience", 10th International PACAP meeting, Israel.
- 2010 Cover, Neuron, Vol 67
- 2009 Cover, Neuron, Vol 63
- 2005 Telethon Post-Doctoral Competitive Fellowship Award (GFP04005)
- 2003 Euresco Travel Award
- 1998 European Science Foundation Travel Award
- 1997 A. Marzullo National Award for the Undergraduate Thesis, University of Trieste (Italy)

Teaching and mentoring experience

- 2018- Lecturer, Introduction to Molecular Biology (48h/year), Data Science Master in Science program, Department of Matematics, University of Padova, Italy
- 2018- Lecturer, Neurodegenerative diseases (10h/year), PhD school in Biomedical Sciences, University of Padova, Italy
- 2017- Lecturer, Molecular Biology (48h/year), Medical School, University of Padova, Italy
- 2017- Lecturer, Molecular Biology (24h/year), Medical School, University of Padova, Italy
- 2014-2017 Lecturer, Neurodegenerative diseases (48h/year), Master in Science program, University of Trento, Italy
- 2015-2017 Lecturer, Neurodegenerative diseases (12h/year), PhD school in Biotechnology, University of Trento, Italy
- 2012-2013 Instructor, General Pharmacology (20h/year), Medical School and the International MD Program, Università Vita e Salute San Raffaele Medical School, Milan, Italy
- 2009-2013 Instructor, "Trinucleotide repeat disorders, Life and Humanoid Technologies PhD program, Istituto Italiano di Tecnologia and University of Genoa
- 2000- Tutoring of the following PhD students:
Dario Bonanomi (ERC awardee, S. Raffaele, Milan, Italy); Elisa Tinelli (post-doc at ETH, Zurich, Switzerland); Isabella Palazzolo (post-doc Harvard, Boston, USA); Chiara Scaramuzzino (post-doc at the University of Grenoble, France); Tanya Aggarwal (post-doc at Karolinska Institute, Sweden); Carmelo Milioto (post-doc at the UCL, UK), Diana Piol, Alice Migazzi, Caterina Marchioretti.

Professional experience and membership

- 2019 Conference committee and organizer, II ENMC-sponsored International meeting on SBMA, Naarden, Netherlands.

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- 2018 Conference committee and organizer: IV SBMA Italian meeting, University of Padova, Italy, 5-6 October
- 2016 Conference committee and organizer: III SBMA Italian meeting, University of Milan, Italy, 11-12 November
- 2016 Conference committee and organizer: Fondazione Telethon Tri-Retreat, Rome, Italy, 26-28 May
- 2015 Conference committee and organizer: II SBMA Italian meeting, University of Trento, Italy, 18 April
- 2015 Conference committee and organizer: I ENMC-sponsored International meeting on SBMA, March 27-29, Naarden, Netherlands.
- 2015 Chair of section: "Motor neuron diseases: from molecular targets to trial design", XVII Telethon Scientific Convention, Riva del Garda (Trento), 9-11 March
- 2014 Chair, Motor Neuron Disease Satellite, FENS, Milan, 3-4 July
- 2013 Conference committee and organizer: I SBMA Italian meeting, University of Padova, 29-30 October.
- 2013 Scientific Committee and Organizer, Summer school: "The core of neuronal communication: axonal biology, degeneration and regeneration", Cavalese, Trento, Italy, September 30-October 4
- 2011 Chair, Meeting: Molecular Mechanisms of Neurodegeneration, Milan (Italy)
- 2007 Federation of European Neuroscience Societies (FENS) member

Editor

- 2018- Editorial board member of "Journal of Clinical and Molecular Pathology"
- 2016- Editorial board member of "Scientific Reports"
- 2013-2017 Editorial board member of "Advances in Neuroscience"

Ad hoc reviewer

Research articles:

PlosOne, Neurobiology of Disease, J. of Molecular Neuroscience, Neuron, Progress in Neurobiology, Human Molecular Genetics, Developmental Cell, European J. Neuroscience, J. Neuroscience Research, Cell Report, Experimental Neurology, Brain Research, Scientific Reports, Acta Neuropathologica, J. Neurological Sciences, JCI

Grant applications:

Reviewer for grant agencies: Association Francaise for Myopathies (AFM), Neuromuscular Disease Association UK, MRC UK.

Publications (* indicates corresponding authorship)

- 2017 Mutations in TGM6 induce the unfolded protein response in SCA35. Tripathy D, Vignoli B, Ramesh N, Polanco MJ, Coutelier M, Stephen CD, Canossa M, Monin ML, Aeschlimann P, Turberville S, Aeschlimann D, Schmahmann JD, Hadjivassiliou M, Durr A, Pandey UB, **Pennuto M**, Basso M. Hum Mol Genet. 26:3749-3762.
- 2017 Jiménez Garduño AM, Juárez-Hernández LJ, Polanco MJ, Tosatto L, Michelatti D, Arosio D, Basso M, **Pennuto M**, Musio C. *Altered ionic currents and amelioration by IGF-1 and PACAP in motoneuron-derived cells modelling SBMA.* Biophys Chem 229:68-76.
- 2017 Querin G, Martinelli I, Bertolin C, Pegoraro E, **Pennuto M**, Sorarù G. *The role of AR polyQ tract in male breast carcinoma: lesson from a SBMA case.* Ann Oncol 28:1160-1161.

- 2017 Borgia D, Malena A, Spinazzi M, Andrea Desbats M, Salviati L, Russell AP, Miotto G, Tosatto L, Pegoraro E, Sorarù G, **Pennuto M***, Vergani L*. *Increased mitophagy in the skeletal muscle of spinal and bulbar muscular atrophy patients.* Hum Mol Genet 26:1087-1103.
- 2017 Milioto C, Malena A, Maino E, Polanco MJ, Marchioretto C, Borgia D, Pereira MG, Blaauw B, Lieberman AP, Venturini R, Plebani M, Sambataro F, Vergani L, Pegoraro E, Sorarù G*, **Pennuto M***. *Beta-agonist stimulation ameliorates the phenotype of spinal and bulbar muscular atrophy mice and patient-derived myotubes.* Sci Rep 7:41046.
- 2016 Polanco MJ, Parodi S, Piol D, Stack C, Chivet M, Contestabile A, Miranda HC, Lievens PMJ, Espinoza S, Jochum T, Rocchi A, Grunseich C, Gainetdinov RR, Cato ACB, Lieberman A, La Spada AR, Sambataro F, Fischbeck KH, Gozes I, **Pennuto M***. *CDK2 inhibition by PACAP/AC/PKA signaling reduces polyglutamine-expanded androgen receptor phosphorylation and toxicity in SBMA.* Sci Transl Med 8:370ra181.
- 2016 Giorgetti E, Yu Z, Chua JP, Shimamura R, Zhao L, Zhu F, Venneti S, **Pennuto M**, Guan Y, Hung G, Lieberman AP. *Rescue of metabolic alterations in AR113Q skeletal muscle by peripheral androgen receptor gene silencing.* Cell Rep 17: 125–136.
- 2016 Bertolin C, Querin G, Da Re E, Sagnelli A, Bello L, Cao M, Muscas M, **Pennuto M**, Ermani M, Pegoraro E, Mariotti C, Gellera C, Hanna MG, Pareyson D, Fratta P, Sorarù G. *No effect of AR polyG polymorphism on spinal and bulbar muscular atrophy phenotype.* Eur J Neurol. 23:1134-6.
- 2016 Rocchi A, Milioto C, Parodi S, Armirotti A, Borgia D, Pellegrini M, Urciuolo A, Molon S, Morbidoni V, Marabita M, Romanello V, Gatto P, Blaauw B, Bonaldo P, Sambataro F, Robins DM, Lieberman AP, Sorarù G, Vergani L, Sandri M, **Pennuto M***. *Glycolytic-to-oxidative fiber-type switch and mTOR signaling activation are early-onset features of SBMA muscle modified by high-fat diet.* Acta Neuropathol 132:127-44.
- 2016 Querin G, Bertolin C, Da Re E, Volpe M, Zara G, Pegoraro E, Caretta N, Foresta C, Silvano M, Corrado D, Iafrate M, Angelini L, Sartori L, **Pennuto M**, Gaiani A, Bello L, Semplicini C, Pareyson D, Silani V, Ermani M, Ferlin A, Sorarù G; Italian Study Group on Kennedy's disease. *Non-neural phenotype of spinal and bulbar muscular atrophy: results from a large cohort of Italian patients.* J Neurol Neurosurg Psychiatry 87: 810--816.
- 2015 Rusmini P, Polanco MJ, Cristofani R, Cicardi ME, Meroni M, Galbiati M, Piccolella M, Messi E, Giorgetti E, Lieberman AP, Milioto C, Rocchi A, Aggarwal T, **Pennuto M**, Crippa V, Poletti A. *Aberrant Autophagic Response in The Muscle of A Knock-in Mouse Model of Spinal and Bulbar Muscular Atrophy.* Sci Rep 5: 15174.
- 2015 Scaramuzzino C, Casci I, Parodi S, Lievens PMJ, Polanco MJ, Milioto C, Chivet M, Monaghan J, Mishra A, Badders N, Aggarwal T, Grunseich C, Sambataro F, Basso M, Fackelmayer FO, Taylor JP, Pandey UB, **Pennuto M***. *Protein arginine methyltransferase 6 enhances polyglutamine-expanded androgen receptor function and toxicity in spinal and bulbar muscular atrophy.* Neuron 85: 88-100.
- 2015 Fossati SM, Candiani S, Nödl MT, Maragliano L, **Pennuto M**, Domingues P, Benfenati F, Pestarino M, Zullo L. *Identification and Expression of Acetylcholinesterase in Octopus vulgaris Arm Development and Regeneration: a Conserved Role for ACHE?* Mol Neurobiol 52: 45-56.

- 2014 Aggarwal T, Polanco JM, Scaramuzzino C, Rocchi A, Milioto C, Emionite L, Ognio E, Sambataro F, Galbiati M, Poletti A, **Pennuto M***. *Androgens affect muscle, motor neuron and survival of a mouse model of SOD1-related ALS*. Neurobiol Aging 35:1929-38.
- 2014 Sambataro F, Wolf ND, **Pennuto M**, Vasic N, Wolf RC. *Revisiting default mode network function in major depression: evidence for disrupted subsystem connectivity*. Psychol Med 1-11.
- 2013 Crippa V, Boncoraglio A, Galbiati M, Aggarwal T, Rusmini P, Giorgetti E, Cristofani R, Carra S, **Pennuto M**, Poletti A. *Differential autophagy power in the spinal cord and muscle of transgenic ALS mice*. Front Cell Neurosci 7:234
- 2013 Malena A, **Pennuto M**, Tezze C, Querin G, D'Ascenzo C, Silani V, Cenacchi G, Scaramozza A, Romito S, Morandi L, Pegoraro E, Russell AP, Soraru` G, Vergani L. *Androgen-dependent impairment of myogenesis in spinal and bulbar muscular atrophy*. Acta Neuropathol 126:109-21.
- 2013 Scaramuzzino C, Monaghan J, Milioto C, Lanson NA, Maltare A, Aggarwal T, Casci I, Fackelmeyer FO, **Pennuto M***, Pandey UB. *Protein arginine methyltransferase 1 and 8 interact with FUS to modify its sub-cellular distribution and toxicity in vitro and in vivo*. PLoS One 8:e61576.
- 2012 Rinaldi C, Bott LC, Chen KL, Harmison GG, Katsuno M, Sobue G, **Pennuto M**, Fischbeck KH. *IGF-1 administration ameliorates disease manifestations in a mouse model of spinal and bulbar muscular atrophy*. Mol Med 18:1261-8.
- 2012 Saporta MA, Shy BR, Patzko A, Bai Y, **Pennuto M**, Ferri C, Tinelli E, Saveri P, Kirschner D, Crowther M, Southwood C, Wu X, Gow A, Feltri ML, Wrabetz L, Shy ME. *MpzR98C arrests Schwann cell development in a mouse model of early-onset Charcot-Marie-Tooth disease type 1B*. Brain 135:2032-47.
- 2012 Parodi S, Di Zanni E, Di Lascio S, Bocca P, Prigione I, Fornasari D, **Pennuto M**, Bachetti T, Ceccherini I. *The E3 ubiquitin ligase TRIM11 mediates the degradation of congenital central hypoventilation syndrome associated with polyalanine expanded PHOX2B proteins*. J Mol Med 90:1025-35.
- 2012 Sambataro F, **Pennuto M**, Wolf C. *Catechol-O-methyl transferase modulates cognition in late life: evidence and implications for cognitive enhancement. Invited review*. CNS & Neurological Disorder-Drug Targets 11:195-208.
- 2012 Bruson A, Sambataro F, Querin G, D'Ascenzo C, Palmieri A, Agostini J, Gaiani A, Angelini C, Galbiati M, Poletti A, **Pennuto M**, Pegoraro E, Clementi M, Soraru` G. *CAG repeat length in androgen receptor gene is not associated with amyotrophic lateral sclerosis*. Eur J Neurol 19:1373-5.
- 2010 Nedelsky NB, **Pennuto M**, Smith RB, Palazzolo I, Moore J, Nie Z, Neale G, Taylor JP. *Native functions of the androgen receptor are essential to pathogenesis in a Drosophila model of spinobulbar muscular atrophy*. Neuron 67: 936-52.
-Featured by Kratter and Finkbeiner, Neuron 67: 897-99.
- 2010 Mbaya E, Oulès B, Caspersen C, Tacine R, Massinet H, **Pennuto M**, Chrétien D, Munnich A, Rötig A, Rizzuto R, Rutter GA, Paterlini-Bréchet P, Chami M. *Calcium signalling-dependent mitochondrial dysfunction and bioenergetics regulation in respiratory chain Complex II deficiency*. Cell Death Differ 17:1855-66.
- 2010 Palazzolo I, Nedelsky NB, Askew CE, Harmison GG, Kazantsev AG, Taylor JP, Fischbeck KH, **Pennuto M***. *B2 attenuates polyglutamine-expanded androgen receptor toxicity in cell and fly models of spinal and bulbar muscular atrophy*. J Neurosci Res 88: 2207-16.

- 2009 Palazzolo I, Stack C, Kong L, Musaro M, Adachi H, Katsuno M, Sobue G, Taylor JP, Sumner JC, Fischbeck HK, **Pennuto M*** *Overexpression of muscle-specific isoform of IGF-1 in the skeletal muscle of SBMA mice extends life and attenuates disease manifestations.* Neuron 63: 316-28.
-Featured by Papanikolaou and Ellerby, *Neuron* 63: 277-78.
-Press coverage: *Neurology Today*, *ScienceBX*, MDA press release, KDA press release
- 2009 Ranganathan S, Harmison G, Meyert K, **Pennuto M**, Burnett B, Fischbeck K *Mitochondrial abnormalities in spinobulbar muscular atrophy.* Hum Mol Genet 18: 27-42.
- 2008 **Pennuto M**, Tinelli E, Malaguti M, Del Carro U, D'Antonio M, Ron D, Quattrini A, Feltri ML, Wrabetz L. *Ablation of the UPR-mediator CHOP restores motor function and reduces demyelination in Charcot-Marie-Tooth 1B mice.* Neuron 57:393-405.
-Featured by Khajavi and Lupski, *Neuron* 57: 329-30.
- 2007 Palazzolo I, Burnett BG, Young JE, Brenne PL, La Spada AR, Fischbeck KH, Howell BW, **Pennuto M***. *Akt blocks ligand binding and protects against expanded polyglutamine androgen receptor toxicity.* Hum Mol Genet 16:1593-603.
- 2006 Wrabetz L, D'Antonio M, **Pennuto M**, Dati G, Tinelli E, Fratta P, Previtali S, Imperiale D, Zielasek J, Toyka K, Avila RL, Kirschner DA, Messing A, Feltri ML, Quattrini A. *Different intracellular pathomechanisms produce diverse Myelin Protein Zero neuropathies in transgenic mice.* J Neurosci 26:2358-68.
- 2005 Bonanomi D, **Pennuto M**, Rigoni M, Rossetto O, Montecucco C, Valtorta F. *Taipoxin induces synaptic vesicle exocytosis and disrupts the interaction of synaptophysin I with VAMP2.* Mol Pharmacol 67:1901-8.
- 2004 Rigoni M, Schiavo G, Weston AE, Caccin P, Allegrini F, **Pennuto M**, Valtorta F, Montecucco C, Rossetto O. *Snake presynaptic neurotoxins with phospholipase A2 activity induce punctate swellings of neurites and exocytosis of synaptic vesicles.* J Cell Sci 117:3561-70.
- 2003 **Pennuto M**, Bonanomi D, Benfenati F, Valtorta F. *Synaptophysin I controls the targeting of VAMP2/synaptobrevin II to synaptic vesicles.* Mol Biol Cell 14:4909-19.
- 2002 **Pennuto M**, Dunlap D, Contestabile A, Benfenati F, Valtorta F. *Fluorescence resonance energy transfer detection of synaptophysin I and vesicle-associated membrane protein 2 interactions during exocytosis from single live synapses.* Mol Biol Cell 13:2706-17.
- 2002 Mandolesi G, Gargano S, **Pennuto M**, Illi B, Molfetta R, Soucek L, Mosca L, Levi A, Jucker R, Nasi S. *NGF-dependent and tissue-specific transcription of vgf is regulated by a CREB-p300 and bHLH factor interaction.* FEBS Lett 510:50-6.
- 1999 Leoni C, Menegon A, Benfenati F, Toniolo D, **Pennuto M**, Valtorta F. *Neurite extension occurs in the absence of regulated exocytosis in PC12 subclones.* Mol Biol Cell 10:2919-31.
- 1997 Di Rocco G[§], **Pennuto M**[§], Illi B, Canu N, Filocamo G, Trani E, Rinaldi AM, Possenti R, Mandolesi G, Sirinian MI, Jucker R, Levi A, Nasi S. *Interplay of the E box, the cyclic AMP response element, and HTF4/HEB in transcriptional regulation of the neurospecific, neurotrophin-inducible vgf gene.* Mol Cell Biol 17:1244-53.
([§]Equal Contribution).

Reviews and book chapters

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- 2018 Manzano R, Sorarú G, Grunseich C, Fratta P, Zuccaro E, **Pennuto M**, Rinaldi C. *Beyond motor neurons: expanding the clinical spectrum in Kennedy's disease.* J Neurol Neurosurg Psychiatry (Epub).
- 2017 **Pennuto M*** & Rinaldi C*. *From gene to therapy in spinal and bulbar muscular atrophy: Are we there yet?* Mol Cell Endocrinol (Epub).
- 2017 Sambataro F & **Pennuto M***. *Post-translational modifications and protein quality control in motor neuron and polyglutamine diseases.* Frontiers Mol Neurosci 10:82.
- 2016 **Pennuto M*** & Gozes I*. *Introduction to the Special Issue on Spinal and Bulbar Muscular Atrophy.* J Mol Neurosci 58: 313-6.
- 2016 **Pennuto M*** & Basso M*. *In vitro and in vivo modeling of spinal and bulbar muscular atrophy.* J. Mol Neurosci 58: 365-73.
- 2015 Finsterer J, Mishra A, Wakil S, **Pennuto M**, Soraru G. *Mitochondrial implications in bulbospinal muscular atrophy (Kennedy disease).* Amyotroph Lateral Scler Frontotemporal Degener 17:112-118.
- 2015 **Pennuto M***, Greensmith L, Pradat PF, Sorarù G*. *210th ENMC international workshop: Research and clinical management of patients with spinal and bulbar muscular atrophy. 27-29 March, 2015, Naarden, The Netherlands.* Neuromusc Dis 25: 802-12.
- 2015 Basso M* & **Pennuto M***. *Serine phosphorylation and arginine methylation at the crossroads to neurodegeneration.* Experimental Neurology 271: 77-83.
- 2014 Sorarù G, Querin G, **Pennuto M**. *Skeletal muscle as an emerging therapeutic target in spinal and bulbar muscular atrophy.* Clinic Invest 4: 293-95.
- 2013 Rocchi A & **Pennuto M***. *New routes to therapy for spinal and bulbar muscular atrophy.* J. Mol Neurosci 50:514-23.
- 2013 Parodi S & **Pennuto M***. *Huntington's Disease: From Disease Pathogenesis to Clinical Perspectives.* In K. Guillory & A.M. Carrasco (Eds): *Huntington's disease: Symptoms, risk factors and prognosis*, NOVA Publishers.
- 2012 Sambataro F & **Pennuto M***. *Cell-autonomous and non-cell-autonomous toxicity in polyglutamine diseases.* Prog Neurobiol 97: 152-72.
- 2011 Parodi S & **Pennuto M***. *Neurotoxic effects of androgens in spinal and bulbar muscular atrophy.* Front Neuroendoc 32: 416-25.
- 2010 **Pennuto M*** and Sambataro F. *Pathogenesis of Polyglutamine Diseases.* In: ENCYCLOPEDIA OF LIFE SCIENCES 2010, John Wiley & Sons, Ltd: Chichester [DOI: 10.1002/9780470015902.a0021486]
- 2010 **Pennuto M**, Fischbeck KH: *Therapeutic prospects for polyglutamine disease.* In Dobson CM, Kelly JW, Ramirez-Alvarado M (eds.): *Protein Misfolding Diseases: Current and Emerging Principles.* Hoboken, John Wiley & Sons.
- 2009 **Pennuto M***, Palazzolo I, Poletti A. *Post-translational modifications and polyglutamine toxicity in neurons.* Hum Mol Genet 18:R40-7.
- 2004 Valtorta F, **Pennuto M**, Bonanomi D, Benfenati F. *Synaptophysin: leading actor or walk-on role in synaptic vesicle exocytosis?* Bioessays 26:445-53.

Invited Talks

- 4/12/2017 13th International symposium on VIP, PACAP & related peptides, University of Hong Kong, Hong Kong SAR, *PACAP/AC/PKA signaling attenuates SBMA by targeting androgen receptor phosphorylation.*
- 12/09/2017 Padua-Mit-Innsbruck: Mitochondrial Conference, University of Padova, Italy, *Mitochondrial alterations and mitophagy in the muscle of SBMA patients and mice.*
- 21/07/2017 EBSA2017 Satellite meeting "Biophysical Approaches to Protein Folding and Disease", Edinburg, UK, Keynote invited speaker, *Brain folding diseases: From disease pathogenesis to therapy development.*
- 18/05/2017 53° CONGRESSO AINeNC – 43° CONGRESSO AIRIC, PALAZZO DELLA SALUTE – PADOVA, *Autofagia nella malattia di Kennedy e nelle malattie del motoneurone.*
- 2016 Declined (maternity)
- 20/10/2015 University of Trieste (Italy), hosted by Prof G. Manfioletti, *Protein arginine methylation in motor neuron diseases: ALS and SBMA.*
- 10/11/2015 German Centre for Neurodegenerative Diseases (DZNE), Tübingen (Germany), hosted by Dr M. Neumann, *Protein arginine methylation in ALS and SBMA.*
- 09/10/2015 SINS, Cagliari (Italy), *The involvement of muscle in SBMA and ALS.*
- 22/09/2015 12th International Symposium on VIP, PACAP and Related Peptides, *PACAP is a novel modifier of spinal and bulbar muscular atrophy.*
- 29/05/2015 Molecular Mechanisms of Neurodegeneration, Milan (Italy), *Protein arginine methylation as a major modifier of motor neuron diseases.*
- 04/05/2015 Mario Negri Institute, Milan (Italy), hosted by Dr E. Zito, *Muscle dysfunction in spinal and bulbar muscular atrophy.*
- 09/04/2015 Department of Biomedical Sciences, University of Padova (Italy), hosted by Prof P. Bernardi, *Skeletal muscle abnormalities and metabolic alterations in spinal and bulbar muscular atrophy.*
- 27/03/2015 ENMC-sponsored International meeting on spinal and bulbar muscular atrophy, March 27-29, Naarden, Netherlands, *Cell and mouse models of SBMA.*
- 09/03/2015 XVIII Telethon Scientific Convention, Riva del Garda, Trento (Italy), *Skeletal muscle degeneration in spinal and bulbar muscular atrophy.*
- 03/03/2015 TIGEM, Naples (Italy), hosted by Dr A. Ballabio, *Spinal and bulbar muscular atrophy: a pure motor neuron or a metabolic disease?*
- 13/10/2014 Dulbecco Telethon Institute Retreat, TIGEM, Naples (Italy), *Targeting Akt signaling in muscle to identify new therapeutic strategies for SBMA.*
- 30/10/2013 First SBMA Italian meeting, University of Padua (Italy), *Role of muscle and IGF-1 in the pathogenesis of SBMA.*
- 28/08/2013 11th International PACAP meeting, Pecs (Hungary), *Pacap/PKA reduces polyglutamine androgen receptor toxicity in cell models of spinal and bulbar muscular atrophy.*
- 31/05/2013 Thierry Latran Foundation meeting Sheffield (UK), *The interplay between androgenic/anabolic steroid and IGF-1 signaling in ALS.*
- 19/10/2012 Hayward Genetics Center, Tulane University, New Orleans (LA, USA), hosted by Dr K. Weissbecker, *Androgen receptor and IGF-1 signaling in spinobulbar muscular atrophy.*
- 24/05/2012 Thierry Latran Foundation meeting Dublin (Ireland), *Interplay between androgenic/anabolic steroid and IGF-1 signaling in ALS.*
- 19/04/2012 1st Joint Israeli/SINS Meeting, Catania (Italy), *Phosphorylation of polyQ-AR by PKA in SBMA.*
- 03/02/2012 CIBIO, University of Trento (Italy), hosted by Prof A. Quattrone, *PolyQ-AR: from post-translational modifications to therapeutic development for SBMA.*

- 14/12/2011 IBRO sponsored lecture, 10th International Symposium on VIP-PACAP and related peptides, Eilat (Israel), *PACAP/PKA reduces polyglutamine androgen receptor toxicity in cell models of spinal and bulbar muscular atrophy.*
- 29/09/2011 Keynote Lecture, Istituto Interuniversitario di Miologia Annual Meeting, Sestri Levante (Italy), *Phosphorylation of polyglutamine androgen receptor in skeletal muscle attenuates disease manifestations in SBMA muscle.*
- 08/06/2011 Department of Neurology, Aachen University (Germany), hosted by Prof J.B. Schulz, *Phosphorylation of polyglutamine androgen receptor in skeletal muscle attenuates disease manifestations in SBMA mice.*
- 14/05/2011 Meeting: Molecular Mechanisms of Neurodegeneration, Milan (Italy), *Phosphorylation of androgen receptor in muscle attenuates disease in SBMA mice.*
- 13/05/2011 Meeting: Thierry Latran Foundation, Hanover (Germany), *The interplay between androgenic/anabolic steroid and IGF-1 signaling in amyotrophic lateral sclerosis.*
- 05/04/2011 University of Padova (Italy), hosted by Prof C. Angelini and Dr G. Sorarú. *CAG trinucleotide repeat disorders: from disease pathogenesis to development of therapy.*
- 19/11/2010 CNR Institute for Neuroscience, Milan (Italy), hosted by Dr N. Borgese, *Native protein function in the pathogenesis of polyglutamine disease.*
- 10/11/2010 Kennedy's Disease Association Annual Meeting, S. Diego (USA), *Phosphorylation of androgen receptor in SBMA pathogenesis.*
- 08/09/2010 First Italian SBMA meeting, Milan (Italy), *IGF-1 and post-translational modifications in SBMA.*
- 12/04/2010 S. Raffaele Scientific Institute, Milan (Italy), hosted by Dr L. Wrabetz, *Polyglutamine disease: from post-translational modification of the disease protein to development of pharmacologic intervention for therapy.*
- 29/01/2009 Institute of Functional Genomics, Montpellier (France), hosted by Prof J. Bockaert, *Polyglutamine androgen receptor and insulin-like growth factor 1.*
- 23/07/2008 Department of Endocrinology, University of Milan (Italy), hosted by Prof A. Poletti, *Polyglutamine diseases: Therapeutic perspectives.*
- 11/07/2008 Institute of Healthy Aging, University College of London (UK), hosted by Prof L. Partridge, *Polyglutamine diseases and aging.*
- 11/06/2008 NeuroCure Symposium, Charité, Berlin (Germany), *Molecular mechanisms of neurodegeneration in polyglutamine diseases.*

Research Support

ACTIVE

MUSCULAR DYSTROPHY ASSOCIATION USA (479363) 2017-2020 \$300,000

Pennuto (PI)

Targeting activity-regulated (dys)function of androgen receptor in SBMA neurons

The goal is to identify androgen receptor as a transcription factor whose activity is subjected to regulation by neuronal excitability in SBMA neurons.

UNIVERSITY OF PADOVA (BIRD PROJECT) 2018-2019 €45,000

Pennuto (PI)

Native androgen receptor functions in normal and spinobulbar muscular atrophy neurons

The goal of this project is to elucidate the basic functions of androgen receptors in neurons in physiological and pathological conditions.

AKIRA ARIMURA RESEARCH GRANT 2018-2019 €50,000

Pennuto (Mentor)

Pennuto, Maria, PhD

Dissecting the molecular logics of neurodegeneration in SBMA and the molecular mechanisms of PACAP

The goal of this project is to elucidate the effect of activation of PACAP signaling in neurodegenerative disease

ASSOCIATION FRANCAISE MYOPATHIES (AFM-18722) 2014-2018 €32,000
Pennuto (Co-PI)

Skeletal muscle: target tissue to cure Spinal and Bulbar Muscular Atrophy (SBMA)

This project supports the pre-clinical evaluation of beta-agonist stimulation in SBMA.

ITALIAN MINISTRY OF HEALTH (RF-2011-02350097) 2014-2018 €165,000
Pennuto (Co-PI)

Skeletal muscle in spinal and bulbar muscular atrophy (SBMA): Therapeutic target and delivery route for potential therapy

This is a pre-clinical study to test the effect of beta-agonists in SBMA mice.

COMPLETED

DULBECCO TELETHON INSTITUTE CAREER AWARD (TCP12013) 2013-2018 €510,000
Pennuto (PI)

Targeting Akt signaling in muscle to identify new therapeutic strategies for spinal and bulbar muscular atrophy.

The goal of this project is to characterize the impact of the IGF-1/Akt signaling in SBMA muscle atrophy.

AKIRA ARIMURA RESEARCH GRANT 2016-2017 €50,000
Pennuto (Mentor)

Targeting PACAP/AC/PKA/CDK2 axis to ameliorate spinal and bulbar muscular atrophy

The goal of this project is to elucidate the effect of activation of PACAP signaling in SBMA pathogenesis

BANDO DI ATENEIO UNIVERSITA' DI TRENTO 2015-2017 €104,000
Pennuto (Coordinator, 2 research groups)

Innovative multidisciplinary approach to neurodegeneration: the impact of chloride signalling dysregulation

This project goal is to characterize the impact of chloride ion dysregulation in neurodegenerative diseases.

MUSCULAR DYSTROPHY ASSOCIATION USA (196646) 2011-2014 \$330,000
Pennuto (PI)

PKA signaling in SBMA pathogenesis

The goals are to identify PKA as a novel modifier of mutant androgen receptor in SBMA.

TELETHON-Italy (GGP10037) 2011- 2013 €190,300
Pennuto (PI)

Insulin-like growth factor 1/Akt and androgen signaling crosstalk in the pathogenesis of spinal and bulbar muscular atrophy.

The goal of this project is to characterize the regulation of phosphorylation of polyglutamine androgen receptor by Akt in the context of SBMA pathogenesis.

THIERRY-LATRAN FOUNDATION (France) 2011–2012 €125,000
Pennuto (Coordinator, 4 research groups)

Pennuto, Maria, PhD

Interplay between Androgenic/Anabolic Steroid and IGF-1 Signaling in Amyotrophic Lateral Sclerosis

The aim of this project is to establish androgens and androgen receptor as risk factors for sporadic ALS.

**Marie-Curie FP7-PEOPLE-2009-RG (FP7-256448) 2009-2014 €100,000
Pennuto (PI)**

Polyglutamine diseases: impact of protein and cell context on neurotoxicity

The aims of this project are to determine the contribution of different factors to polyglutamine toxicity, including different domains and the pattern of expression of the disease protein.

**KENNEDY DISEASE ASSOCIATION USA 2009 \$20,000
Pennuto (PI)**

Role of phosphorylation of polyglutamine androgen receptor by PKA in SBMA pathogenesis

The goal of this project is to identify PKA as a novel modifier of polyglutamine androgen receptor toxicity.

**MUSCULAR DYSTROPHY ASSOCIATION USA (92333) 2008-2011 \$179,620
Pennuto (PI)**

Targeting androgen receptor for development of SBMA therapeutics

The goals are to identify post-translational modifications of androgen receptor as modifiers of polyglutamine toxicity in SBMA.

**KENNEDY DISEASE ASSOCIATION USA 2007 \$25,000
Pennuto (PI)**

Targeting androgen receptor for development of SBMA therapeutics

The goal of this project is to identify specific phosphorylation sites on mutant androgen receptor that modify the toxicity of mutant protein in SBMA.

References:

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