europass Curriculum vitae

PERSONAL INFORMATION

Alessandra Patitucci

(Italy)

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Sex Female | Nationality Italian

POSITION

Researcher

WORK EXPERIENCE

3 Nov 2017–Present

Researcher

C.N.R.-National Research Council - ISAFoM - Institute for agricultural and forest systems in the Mediterranean
2-4, Via Cavour, 87036 Rende (Cosenza) (Italy)
www.isafom.cnr.it/index.php/it/

Research activity: Studies in the History of Medicine and Ethnomedicine with particular attention to the remedies used by Italian folk medicine for the treatment of different diseases: evaluation of the antimicrobial, anti-inflammatory and modulating properties of hormonal cascades of active ingredients contained in plants in light of current physiological and pharmacological knowledge.

Business or sector Ecosystems, Agriculture, Forestry, Anthropology, Ethnomedicine

1 Feb 2010–2 Nov 2017

Researcher

C.N.R.-National Research Council-I.S.N. Institutes of Neurological Sciences
Contrada Burga, 87050 Mangone (Cosenza) (Italy)
www.isn.cnr.it

Research activity: the main fields of research are focused on the study of the hereditary diseases of the nervous system with particular attention to: Peripheral Neuropathies, Huntington Disease, Kennedy Disease, Neurofibromatosis, Amyotrophic Lateral Sclerosis, Rett Syndrome , Hereditary Spastic Paraparesis, CADASIL, Spinal Muscular Atrophy. The studies are based on mutational screening of candidate genes, on the genotype-phenotype correlation and on the linkage analysis.

Business or sector Molecular Biology and Human Genetic

11 Jan 2016–29 Apr 2016

Theacher

Fondazione Istituto Tecnico Superiore Tirreno, Fuscaldo (Cosenza) (Italy)

Theaching for Biomedical Equipment

14 Oct 2015–20 Dec 2015

University teaching assistant

Fondazione Istituto Tecnico Tirreno, Fuscaldo (Cosenza) (Italy)

Tutor for teaching Biomedical Equipment as part of the Course for the formation of Higher Technicians for the production, maintenance of equipment, diagnostic and biomedical devices.

1 Feb 2001–14 Dec 2010

Technical collaborator research institutes

C.N.R.-National Research Council-I.S.N.-Institutes of Neurological Sciences
Contrada Burga, 87050 Mangone (Cosenza) (Italy)
www.isn.cnr.it

Molecular study of the main diseases of the nervous system.

Business or sector Molecular Biology and Human Genetic

1 Mar 2000–31 Jan 2001

Researcher assistant (research grant)

C.N.R.-National Research Council-I.S.N.-Institutes of Neugrological Sciences
Contrada Burga, 87050 Mangone (Cosenza) (Italy)
www.isn.cnr.it

Genetical study in subject affected by hereditary neorupaties.

Business or sector Molecular Biology and Human Genetic

15 Jan 1998–28 Feb 2000

Researcher assistant (professional collaboration)

C.N.R.-National Research Council-I.S.N.-Institutes of Neugrological Sciences
Contrada Burga, 87050 Mangone (Cosenza) (Italy)
www.isn.cnr.it

Screening in the territory of subjects affected from hereditary neuropathies.

Business or sector Molecular biology and Human Genetic

EDUCATION AND TRAINING

1 Aug 1994–31 Jul 1996

Fellowship

C.N.R.-National Research Council-I.M.S.E.B.-Institute of Experimental Medicine and Biotechnology, Mangone (Cosenza) (Italy)

Identification of Hemoglobinopathies in its various forms, with particular attention to the Microcytemias examined also from the epidemiological point of view. The genetical study was carried aout in order to identify subjects carrying these anomalies.

Jul 1991

Biologist

University of Calabria, Arcavacata di Rende (Cosenza) (Italy)

Qualification to the profession of biologist

1990

Theacher in high school

Regione Calabria, Catanzaro (Italy)

Qualification for teaching in high schools for the competition class Natural Sciences, Chemistry and Geography, Microbiology, Agricultural Entomology and Phytopathology, BAND 1990.

7 Mar 1990

Graduation

Degree in Biological Sciences with laude

University of Calabria, Arcavacata di Rende (Cosenza) (Italy)

Experimental thesis entitled “Ultrastructural characteristics of the spermiogenesis of the Wren (Troglodytes troglodytes)"

PERSONAL SKILLS

Mother tongue(s): Italian

Foreign language(s)

	UNDERSTANDING		SPEAKING		WRITING
	Listening	Reading	Spoken interaction	Spoken production	
English	B1	B1	B1	B1	B1

Levels: A1 and A2: Basic user - B1 and B2: Independent user - C1 and C2: Proficient user
Common European Framework of Reference for Languages

Job-related skills: RESPONSIBILITY DUTIES:

• Head of the correct functionality and management of high performance liquid chromatography (DHPLC), protocol no. 66 of 18.12.2002, by the Director of the Institute of Neurological Sciences of Mangone (CS).

TUTORING:

• Responsible for training during the preparation of the experimental thesis on molecular biology of the student Annunziato Morabito with appointment of Tutor by the Director of the Institute of Neurological Sciences of Mangone (CS), prof. Aldo Quattrone pursuant to Legislative Decree n. 629/94.

• Responsible for training during the preparation of the experimental thesis on molecular biology of the student Rosellina Margherita Mancina with the appointment of Tutor by the Director of the Institute of Neurological Sciences of Mangone (CS), prof. Aldo Quattrone pursuant to Legislative Decree n. 629/94.

• Head of training during the preparation of the experimental thesis on molecular biology of the student Gabriella Livide with the appointment of Tutor by the Director of the Institute of Neurological Sciences of Mangone (CS), prof. Aldo Quattrone pursuant to Legislative Decree n. 629/94.

• Head of training during the preparation of the experimental thesis on molecular biology of the student Cesare Daniele Venneri with appointment of Tutor by the Director of the Institute of Neurological Sciences of Mangone (CS), prof. Aldo Quattrone pursuant to Legislative Decree n. 629/94.

TECHNICAL SKILLS:

• DNA manipulation techniques:

• Genomic DNA extraction, Genomic DNA digestion with restriction endonucleases, Agarose gel electrophoresis, Southern Blot, Radioactive probe DNA marking, Hybridization, Acrylamide gel electrophoresis, Silver nitrate staining

• DNA probes:Transformation of bacteria with plasmid DNA, Minipreparations of plasmid DNA, Preparation of plasmid DNA, Isolation of DNA probe

• RNA manipulation techniques:

• Extraction of RNA from blood and tissues, Northern blot

• PCR, Nested PCR, RT-PCR, VNTR, SSCP, RFLP, REAL TIME PCR

• Linkage analysis

• PCR of Microsatellites, Gene Scan, Analysis of Aplotypes, Calculation of Lod Score

• Automatic Sequencing: Sequence PCR, Sequences Analysis

• DHPLC analisys (Denaturing High Performance Liquid Chromatography)

Digital skills

SELF-ASSESSMENT				
Information processing	Communication	Content creation	Safety	Problem-solving
Proficient user	Proficient user	Proficient user	Proficient user	Proficient user

Digital skills - Self-assessment grid

Good knowledge of Database use:

http://www.ncbi.nih.gov (PubMed. Blast, OMIM)

http://www.gdb.org (Markers, Hugo)

http://www.ensembl.org (Markers, physical maps, proteins)

http://www.genome.wi.mit.edu (contiguous YAC)

http://insertion.stanford.edu/dhplc.html (methods for Dhplc)

http://genome-www2.stanford.edu/cgi-bin/SGD/web-primer (search for optimal primers)

Good competence of the following tools:

Windows Office 2013 operating system

Genemapper software and Sequencing analisys software

ADDITIONAL INFORMATION

Publications

1. A review of the antimicrobial potential of herbal drugs used in popular Italian medicine (1850s-1950s) to treat bacterial skin diseases. Mazzei R, Leonti M, Spadafora S, PATITUCCI A, Tagarelli G. J Ethnopharmacol. 2019 Nov 29;250:112443.

2. NeuroArray: A Customized aCGH for the Analysis of Copy Number Variations in Neurological Disorders. La Cognata V, Morello G, Gentile G, Cavalcanti F, Cittadella R, Conforti FL, De Marco EV, Magariello A, Muglia M, PATITUCCI A, Spadafora P, D'Agata V, Ruggieri M, Cavallaro S. Curr Genomics. 2018 Sep;19(6):431-443.

3. Cortese R, Zoccolella S, Muglia M, PATITUCCI A, Scarafino A, Paolicelli D, Simone IL. A rare association between multiple sclerosis and Charcot-Marie-Tooth type 1B. Brain Behav. 2016 Sep 25;6(12):e00580.

4. Gasparini S, Qualtieri A, Ferlazzo E, Cianci V, PATITUCCI A, Spadafora P, Aguglia U. Normal immunofluorescence pattern of skin basement membranes in a family with porencephaly due to COL4A1 G749S mutation. Neurol Sci. 2016 Mar;37(3):459-63.

5. Muglia M, Citrigno L, D'Errico E, Magariello A, Distaso E, Gasparro AA, Scarafino A, PATITUCCI A, Conforti FL, Mazzei R, Cortese R, Tortelli R, Simone IL. A novel KIF5A mutation in an Italian family marked by spastic paraparesis and congenital deafness. J Neurol Sci. 2014 Aug 15;343(1-2):218-20.

6. Magariello A, Citrigno L, Zuchner S, Gonzalez M, PATITUCCI A, Sofia V, Conforti FL, Pappalardo I, Mazzei R, Ungaro C, Zappia M, Muglia M. Further evidence that DDHD2 gene mutations cause autosomal recessive hereditary spastic paraplegia with thin corpus callosum. Eur J Neurol 2014 Mar;21(3):e25-6.

7. Magariello A, Tortorella C, PATITUCCI A, Tortelli R, Liguori M, Mazzei R, Conforti FL, Citrigno L, Ungaro C, Simone IL, Muglia M. First mutation in the nuclear localization signal sequence of spastin protein identified in a patient with hereditary spastic paraplegia. Eur J Neurol. 2013 Jan;20(1):e22-3.

8. Conforti FL, Spataro R, Sproviero W, Mazzei R, Cavalcanti F, Condino F, Simone IL, Logroscino G, PATITUCCI A, Magariello A, Muglia M, Rodolico C, Valentino P, Bono F, Colletti T, Monsurrò MR, Gambardella A, La Bella V. Ataxin-1 and ataxin-2 intermediate-length PolyQ expansions in amyotrophic lateral sclerosis. Neurology. 2012 Dec 11;79(24):2315-20.

9. PATITUCCI A, Magariello A, Ungaro C, Muglia M, Conforti FL, Gabriele AL, Citrigno L, Sproviero W, Mazzei R. SMN1 gene copy number analyses for SMA healthy carriers in Italian population. J Pediatr Genet. 2012 Jun;1(2):99-102.

10. Magariello A, Tortorella C, Citrigno L, PATITUCCI A, Tortelli R, Mazzei R, Conforti FL, Ungaro C, Sproviero W, Gambardella A, Muglia M. The p.Arg416Cys mutation in SPG3a gene associated with a pure form of spastic paraplegia. Muscle Nerve. 2012 Jun;45(6):919-20.

11. Sproviero W, La Bella V, Mazzei R, Valentino P, Rodolico C, Simone IL, Logroscino G, Ungaro C, Magariello A, PATITUCCI A, Tedeschi G, Spataro R, Condino F, Bono F, Citrigno L, Monsurrò MR, Muglia M, Gambardella A, Quattrone A, Conforti FL. FUS mutations in sporadic amyotrophic lateral sclerosis: clinical and genetic analysis. Neurobiol Aging. 2012 Apr;33(4):837.e1-5.

12. Conforti FL, Sproviero W, Simone IL, Mazzei R, Valentino P, Ungaro C, Magariello A, PATITUCCI A, La Bella V, Sprovieri T, Tedeschi G, Citrigno L, Gabriele AL, Bono F, Monsurrò MR, Muglia M, Gambardella A, Quattrone A. TARDBP gene mutations in south Italian patients with amyotrophic lateral sclerosis. J Neurol Neurosurg Psychiatry. 2011 May;82(5):587-8.

13. Gabriele AL, Ruggieri M, PATITUCCI A,Magariello A, Conforti FL, Mazzei R, Muglia M, Ungaro C, Di Palma G, Citrigno L, Sproviero W, Gambardella A, Quattrone A. A novel NF1 gene mutation in an Italian family with neurofibromatosis type 1. Childs Nerv Syst. 2011 Apr;27(4):635-8.

14. Magariello A, Muglia M, PATITUCCI A,Ungaro C, Mazzei R, Gabriele AL, Sprovieri T, Citrigno L, Conforti FL, Liguori M, Gambardella A, Bono F, Piccoli T, Patti F, Zappia M, Mancuso M, Iemolo F, Quattrone A. Mutation analysis of the SPG4 gene in Italian patients with pure and complicated forms of spastic paraplegia. J Neurol Sci. 2010 Jan 15;288(1-2):96-100. doi: 10.1016/j.jns.2009.09.025.

15. Cavalcanti F, Kidd T, PATITUCCI A, Valentino P, Bono F, Nisticò R, Quattrone A. An axon regeneration signature in a Charcot-Marie-Tooth disease type 2 patient. J Neurogenet. 2009;23(3):324-8.

16. Muglia M, Vazza G, PATITUCCI A,Milani M, Pareyson D, Taroni F, Quattrone A, Mostacciuolo ML A novel founder mutation in the MFN2 gene associated with variable Charcot-Marie-Tooth type 2 phenotype in two families from Southern Italy. BMJ Case Rep. 2009;2009.

17. Liguori M, Fera F, PATITUCCI A, Manna I, Condino F, Valentino P, Telarico P, Cerasa A, Gioia MC, di Palma G, Quattrone A. A longitudinal observation of brain-derived neurotrophic factor mRNA levels in patients with relapsing-remitting multiple sclerosis. Brain Res. 2009 Feb 23;1256:123-8.

18. Ungaro C, Mazzei R, Conforti FL, Sprovieri T, Servillo P, Liguori M, Citrigno L, Gabriele AL, Magariello A, PATITUCCI A, Muglia M, Quattrone A. CADASIL: extended polymorphisms and mutational analysis of the NOTCH3 gene. J Neurosci Res. 2009 Apr;87(5):1162-7.

19. Mazzeo A, Di Leo R, Toscano A, Muglia M, PATITUCCI A, Messina C, Vita G. Charcot-Marie-Tooth type X: unusual phenotype of a novel CX32 mutation. Eur J Neurol. 2008 Oct;15(10):1140-2.

20. Ungaro C, Mazzei R, Conforti FL, Sprovieri T, Servillo P, Liguori M, Citrigno L, Gabriele AL, Magariello A, PATITUCCI A, Muglia M, Quattrone A. CADASIL: extended polymorphisms and mutational analysis of the NOTCH3 gene. J Neurosci Res. 2009 Apr;87(5):1162-7.

21. A Liguori M, Fera F, PATITUCCI A, Manna I, Condino F, Valentino P, Telarico P, Cerasa A, Gioia MC, di Palma G, Quattrone A. A longitudinal observation of brain-derived neurotrophic factor mRNA levels in patients with relapsing-remitting multiple sclerosis. Brain Res. 2009 Feb 23;1256:123-8.

22. Luisa Conforti F, Sprovieri T, Mazzei R, PATITUCCI A, Ungaro C, Zoccolella S, Magariello A, Bella VL, Tessitore A, Tedeschi G, Simone IL, Majorana G, Valentino P, Citrigno L, Gabriele A, Bono F, Monsurrò MR, Muglia M, Quattrone A. Further evidence that D90A-SOD1 mutation is recessively inherited in ALS patients in Italy. Amyotroph Lateral Scler. 2009 Feb;10(1):58-60.

23. Cavalcanti F, Kidd T, PATITUCCI A, Valentino P, Bono F, Nisticò R, Quattrone A, An axon re generation signature in a Charcot-Marie- Tooth Disease type 2, Journal of Neurogenetics, 2009 Jan.

24. Mazzeo A, Muglia M, Rodolico C, Toscano A, PATITUCCI A, Quattrone A, Messina C, Vita G. Charcot-Marie-Tooth disease type 1B: marked phenotypic variation of the Ser78Leu mutation in five Italian families. Acta Neurol Scand. 2008 Nov;118(5):328-32.

25. Mazzeo A, Di Leo R, Toscano A, Muglia M, PATITUCCI A, Messina C, Vita G Charcot-Marie-Tooth type X: unusual phenotype of a novel CX32 mutation. Eur J Neurol. 2008 Oct;15(10):1140-2.

26. Muglia M, Magariello A, Citrigno L, Passamonti L, Sprovieri T, Conforti FL, Mazzei R, PATITUCCI A, Gabriele AL, Ungaro C, Bellesi M, Quattrone A. A novel locus for dHMN with pyramidal features maps to chromosome 4q34.3-q35.2. Clin Genet. 2008 May;73(5):486-91.

27. Malandrini A, Gambelli S, Muglia M, Berti G, Gaudiano C, PATITUCCI A, Sugie K, Umehara F, Quattrone A, Dotti MT, Federico A. Motor-sensory neuropathy without minifascicles in a patient with 46XY gonadal dysgenesis. Brain Dev. 2008 Apr;30(4):291-4.

28. Conforti FL, Sprovieri T, Mazzei R, Ungaro C, La Bella V, Tessitore A, PATITUCCI A, Magariello A, Gabriele AL, Tedeschi G, Simone IL, Majorana G, Valentino P, Condino F, Bono F, Monsurrò MR, Muglia M, Quattrone A A novel Angiogenin gene mutation in a sporadic patient with amyotrophic lateral sclerosis from southern Italy, Neuromuscul Disord. 2008 Jan;18(1):68-70.

29. Mazzei R, Guidetti D, Ungaro C, Conforti FL, Muglia M, Cenacchi G, Lanza PL, PATITUCCI A, Sprovieri T, Riguzzi P, Magariello A, Gabriele AL, Citrigno L, Preda P, Quattrone A. First evidence of a pathogenic insertion in the NOTCH3 gene causing CADASIL. J Neurol Neurosurg Psychiatry. 2008 Jan;79(1):108-10.

30. Muglia M, Vazza G, PATITUCCI A, Milani M, Pareyson D, Taroni F, Quattrone A, Mostacciuolo ML. A novel founder mutation in the MFN2 gene associated with variable Charcot-Marie-Tooth type 2 phenotype in two families from Southern Italy. J Neurol Neurosurg Psychiatry. 2007 Nov;78(11):1286-7

31. PATITUCCI A*, Muglia M*, Rizzi R, Ungaro C, Conforti FL, Gabriele AL, Magariello A, Mazzei R, Motti L, Saladini R, Sprovieri T, Marcello N, Quattrone A. A novel point mutation in PMP22 gene in an Italian family with hereditary neuropathy with liability to pressure palsies. Journal of Neurological Sciences, 2007, Dec 15;263(1-2):194-7. (*These authors equally contributed to this work).

32. Mazzei R, Conforti FL, Ungaro C, Liguori M, Magariello A, Gabriele AL, PATITUCCI A, Sprovieri T, Muglia M, Quattrone. Gene Symbol: NOTCH3. Disease: CADASIL. Hum Genet, 2007.

33. Mazzei R, Ungaro C, Conforti FL, Liguori M, Magariello A, Gabriele AL, Sprovieri T, PATITUCCI A, Muglia M, Quattrone. “Gene Symbol: NOTCH3”. Disease: CADASIL. Hum Genet, 2007

34. Mazzei R, Conforti FL, Ungaro C, Liguori M, Magariello A, Gabriele AL, PATITUCCI A, Sprovieri T, Muglia M, Quattrone. Gene Symbol: NOTCH3. Disease: CADASIL. Hum Genet, 2007.

35. Ungaro C, Sprovieri T, Conforti FL, Muglia M, PATITUCCI A, Magariello A, Gabriele AL, Quattrone A, Mazzei R. Putative role of specific JAG1 gene exons in modulating clinical features in patients with leucoencephalopathy. Neuroscience Letters, 2007 May 11;418(1):1-3.

36. Mazzei R, Conforti FL, Ungaro C, Liguori M, Magariello A, Gabriele AL, PATITUCCI A, Sprovieri T, Muglia M, Quattrone. Gene Symbol: NOTCH3. Disease: CADASIL. Hum Genet, 2007

Apr;121(2):296.

37. Mazzei R, Ungaro C, Conforti FL, Liguori M, Magariello A, Gabriele AL, Sprovieri T, PATITUCCI A, Muglia M, Quattrone. Gene Symbol: NOTCH3. Disease: CADASIL. Hum Genet, 2007 Apr;121(2):296.

38. Mazzei R, Conforti FL, Ungaro C, Liguori M, Magariello A, Gabriele AL, PATITUCCI A, Sprovieri T, Muglia M, Quattrone. Gene Symbol: NOTCH3. Disease: CADASIL. Hum Genet, 2007 Apr;121(2):295

39. Conforti FL, Mazzei R, Sprovieri T, Ungaro C, PATITUCCI A, Magariello A, Gabriele AL, Bravaccio C, Muglia M, Quattrone A. Gene symbol: MECP2. Disease: Rett syndrome. Hum Genet. 2006 Jul;119(6):676.

40. Conforti FL, Mazzei R, PATITUCCI A, Magariello A, Sprovieri T, Ungaro C, Gabriele AL, Muglia M, Del Giudice E, Quattrone A. Gene symbol: MECP2. Disease: Rett syndrome. Hum Genet. 2006 Jul;119(6):675.

41. Magariello A, Muglia M, PATITUCCI A, Mazzei R, Conforti FL, Gabriele AL, Sprovieri T, Ungaro C, Gambardella A, Mancuso M, Siciliano G, Branca D, Aguglia U, de Angelis MV, Longo K, Quattrone A. Novel spastin (SPG4) mutations in Italian patients with hereditary spastic paraplegia. Neuromuscul Disord. 2006 Jun;16(6):387-390.

42. Conforti FL, Sprovieri T, Mazzei R, Ungaro C, Tessitore A, Tedeschi G, PATITUCCI A, Magariello A, Gabriele AL, Labella V, Simone IL, Majorana G, Valentino P, Muglia M, Quattrone A. Sporadic ALS is not associated with VAPB gene in Southern Italy. J Negat Results Biomed. 2006 May 29;5:7.

43. Conforti FL, Mazzei R, Sprovieri T, Ungaro C, PATITUCCI A, Magariello A, Gabriele AL, Bravaccio C, Muglia M, Quattrone A. Gene symbol: MECP2. Disease: Rett syndrome. Hum Genet. 2006;119:676.

44. Conforti FL, Mazzei R, PATITUCCI A, Magariello A, Sprovieri T, Ungaro C, Gabriele AL, Muglia M, Del Giudice E, Quattrone A. Gene symbol: MECP2. Disease: Rett syndrome. Hum Genet. 2006;119:675.

45. Rodolico C, Toscano A, PATITUCCI A, Muglia M, Gaeta M, D'Arrigo G, Migliorato A, Messina S, Quattrone A, Messina C, Vita G, Clinical and muscle magnetic resonance imaging study of an Italian family with autosomal dominant inclusion body myopathy not linked to known genetic loci, Neurological Science, 2005 Dec;26(5):303-9.

46. Malandrini A, Gambelli S, Muglia M, Berti G, PATITUCCI A, Sugie K, Umehara F, Quattrone A, Dotti MT, Federico A, Motor-sensory neuropathy with minifascicle formation in a woman with normal karyotype, Neurology, 2005 Sep 13; 65(5):776.

47. PATITUCCI A, Muglia M, Gabriele AL, Magariello A, Peluso G, Sprovieri T, Conforti FL, Mazzei R, Ungaro C, Condino F, Valentino P, Bono F, Rodolico C, Mazzeo A, Toscano A, Quattrone A Comparison Of Different Techniques In Detecting CMT1A/HNPP Duplication/Deletion, Neuromuscul Disord. 2005 Jul;15(7):488-92.

48. Valentino P, Conforti FL, , Pirritano D, Nisticò R, Mazzei R, PATITUCCI A, Sprovieri T, Gabriele AL, Muglia M, Clodomiro A, Gambardella A, Zappia M and Quattrone A Brachial amyotrophic diplegia associated with a novel Sod1 mutation (L106P), Neurology. 2005 Apr 26;64(8):1477-8.

49. L. Passamonti, M. Muglia, A. Magariello, M. Bellesi, R. Mazzei, A. PATITUCCI, F. Conforti, AL. Gabriele, T. Sprovieri, G. Peluso, M. Caracciolo, E. Medici, F. Logullo, L. Provinciali and A. Quattrone. Further evidence of heterogeneity in autosomal dominant distal motor neuropathy. Neuromuscular Disorder, 2004 Nov; 14(11):705-10.

50. Conforti FL, Muglia M, Mazzei R, PATITUCCI A, Valentino P, Magariello A, Sprovieri T, Bono F, Bergmann C, Gabriele AL, Peluso P, Nisticò R, Senderek J, Quattrone A. A new SBF2 mutation in a family with recessive demyelinating Charcot-Marie-Tooth (CMT4B2). Neurology, 2004 Oct 12; 63(7):1327-8.

51. Mazzei R, Conforti FL, Lanza PL., Sprovieri T., Lupo MR., Gallo O., PATITUCCI A, Magariello A, Caracciolo M.,Gabriele AL, Fera F., Valentino P., Bono F.,Santoro G., Muglia M, Quattrone A. A novel Notch3 gene mutation not involving a cysteine residue in an italian family with Cadasil, Neurology, 2004 Aug 10;63(3):561-4.

52. Zuchner S, Mersiyanova IV, Muglia M, Bissar-Tadmouri N, Rochelle J, Dadali EL, Zappia M, Nelis E, PATITUCCI A, Senderek J, Parman Y, Evgrafov O, Jonghe PD, Takahashi Y, Tsuji S, Pericak-Vance MA, Quattrone A, Battologlu E, Polyakov AV, Timmerman V, Schroder JM, Vance JM. Mutations in the mitochondrial GTPase mitofusin 2 cause Charcot-Marie-Tooth neuropathy type 2A. Nat Genet. 2004 May; 36(5):449-51.

53. Conforti F.L., Magariello A., Mazzei R., Sprovieri T., PATITUCCI A., Gabriele A.L., Crescibene L., Bastone L., Muglia M., Quattrone A. Abnormally high levels of SOD1 mRNA in a patient with

Amyotrophic Lateral Sclerosis, Muscle & Nerve 2004; 29(4): 610-611.

54. Mazzei R, Gambardella A, Conforti FL, Magariello A, PATITUCCI A, Gabriele AL, Sprovieri T, Labate A, Valentino P, Bono F, Bonavita S, Zappia M, Muglia M, Quattrone A. Gene conversion events in adult-onset Spinal Muscular Atrophy, Acta Neurol Scan. 2004 Feb; 109(2): 151-4.

55. Muglia M, Criscuolo C, Magariello A, De Michele G, Scarano V, D'Adamo P, Ambrosio G, Gabriele AL, PATITUCCI A, Mazzei R, Conforti FL, Sprovieri T, Morgante L, Epifanio A, La Spina P, Valentino P, Gasparini P, Filla A, Quattrone A. Narrowing of the critical region in autosomal recessive spastic paraplegia linked to the SPG5 locus. Neurogenetics. 2003; 5(1): 49-54.

56. R. Mazzei, FL Conforti, M. Muglia, T. Sprovieri, A. PATITUCCI, A. Magariello, AL. Gabriele, A. Quattrone A Simple Method for Diagnosis of Autosomal Recessive Spinal Muscular Atrophy by DHPLC. J Child Neurol. 2003 Apr; 18(4):269-71.

57. Conforti FL, Mazzei R, Magariello A, PATITUCCI A, Gabriele AL, Muglia M, Quattrone A, Fiumara A, Barone R, Pavone L, Nistico R, Mangone L. Mutation analysis of the MECP2 gene in patients with Rett syndrome. Am J Med Genet 2003 Mar1;117A(2):184-7.

58. Muglia M, Magariello A, Nicoletti G, PATITUCCI A, Gabriele AL, Conforti FL, Mazzei R, Caracciolo M, Casari G, Ardito B, Lastilla M, Gambardella A, Quattrone A. A large family with pure autosomal dominant hereditary spastic paraplegia from southern Italy mapping to chromosome 14q11.2-q24.3. J Neurol 2002 Oct;249(10):1413-6.

59. Mazzei R, Conforti FL, Magariello A, Bravaccio C, Militerni R, GabrieleAL, Sampaolo S, PATITUCCI A, Di Iorio G, Muglia M, Quattrone A. A novel mutation in the CLN1 gene in a patient with juvenile neuronal ceroid lipofuscinosis. J Neurol 2002 Oct; 249(10):1398-400.

60. Muglia M., Magariello A., Nicoletti G., PATITUCCI A., Gabriele AL., Conforti FL., Mazzei R., Caracciolo M., Ardito B., Lastilla M., Tedeschi G., Quattrone A. Further evidence that SPG3A gene mutations cause autosomal dominant hereditary spastic paraplegia. Ann Neurol. 2002 Jun; 51(6):669-72.

61. Oliveri R.L., Muglia M., De Stefano N., Mazzei R., Labate A., Conforti F.L., PATITUCCI A., Gabriele A.L., Magariello A., Zappia M., Gambardella A., Federico A., Quattrone A. A novel mutation in the Notch3 gene in an italian family with CADASIL: genetic and MRI Spectroscopic findings, Archives of Neurology, 2001, 58(9):1418-22.

62. Gambardella A., Muglia M., Labate A., Magariello A., Gabriele A.L., Mazzei R., Pirritano D., Conforti F.L., PATITUCCI A., Valentino P., Zappia M., Quattrone A. Juvenile Huntington's disease presenting as progressive myoclonic epilepsy, Neurology, 2001, 57(4)708-11.

63. Muglia M, Zappia M, Timmerman V, Valentino P, Gabriele AL, Conforti FL, De Jonghe P, Ragno M, Mazzei R, Sabatelli M, Nicoletti G, PATITUCCI A, Oliveri RL, Bono F, Gambardella A, Quattrone A. Clinical and genetic study of a large Charcot-Marie-Tooth type 2A family from southern Italy. Neurology. 2001 Jan 9;56(1):100-3.

64. Muglia M., Toscano A., Gabriele A.L., Magariello A., PATITUCCI A., Conforti FL., Mazzei R., Rodolico C., Gambardella A., Quattrone A. Identification of a new polymorphism (c134G>A) in the exon 2 of the myelin protein zero gene. Hum Mutat. 2000 Mar; 15(3):299.

Conferences

• European Society of Human Genetics Conference 2010 Gothenburg, Sweden - June 12 - 15, 2010;

• Italian Society of Human Genetics 1999- Orvieto;

• International Symposium "The Human Genome", Naples 6-8 September 2000;

• European Society of Human Genetics Conference 2004, Munich, Germany, 12-15 June 2004;

• Oral communication at the XXXV Congress of the Italian Society of Neurology, Genoa 25-29 September 2004 with a presentation entitled "Comparison of different techniques in detecting CMT1A / HNPP duplication / deletion".

• Oral communication at the XXXVII Congress of the Italian Society of Neurology, Bari 14-18 October 2006 with a presentation entitled "HNPP two to a novel frameshift mutation of the PMP22 gene.

Courses

• Transgenomic DHPLC technical seminar: update, use and maintenance, 10 June 2009, Rome.

• MicroRNA profiling course and AB solution for gene expression and genotyping, 27 January 2009, Cosenza.

• Course on "Gene amplification in PCR end point and its evolution in qPCR", held at the Institute of Neurological Sciences - CNR, Piano Lago di Mangone (CS) on 17-18 June 2008.

• English language training course OISE School, London, 09-13 June 2008.

• Rna Intervention Seminar and integrated scientific solutions for genotyping and gene expression SNP, 18 April 2007, Arcavacata di Rende.

• ABI Prism 7900HT Course, in Real Time PCR, 16-17 April 2007, Cosenza.

• Focus on Biotechnology: PCR, Microarrays & Proteomics, Milan, Palazzo delle Stelline Congress Center, December 2004.

• Theoretical-practical course ABI PRISM 7900, Rome, 26-27 February 2002; • 2nd Quantitative PCR Course, Florence 8-10 May 200.

• Overview of the PCR, Arcavacata di Rende, 3 May 2001; • XI Residential Medical Genetics Course, San Giovanni Rotondo, 14-16 June 2001.

Memberships

• Member of the Commission of selection for the assignment of a collaboration assignment num 01/2018 / CS (prot. 2524/2018) - Appointment of the Commissionwith provision n. 22 of 11.05.2007 - "PUBLIC-PRIVATE LABORATORY FOR HIGH INNOVATIVE TECHNOLOGIES ENVIRONMENTAL SUSTAINABILITY IN THE FOREST-WOOD-ENERGY SUPPLY CHAIN"- PON03PE_00024_1 - CUP: B28C14000010005 at the CNR-ISAFoM- Rende (Cosenza).

• Member of the Commission of selection with provision n. 22 of 11.05.2007, note prot. 444 of 11.05.2007- No. 1 work contract entitled "Applications of molecular genetics to the study of nervous system diseases" at the CNR-ISN of Mangone (Cosenza).

• Member of the Commission of selection with provision n. 17 of 04.04.2007, note prot. No. 380 of 04.19.2007-No. 1 research grant to be used at the CNR-ISN of Mangone (Cosenza), part of the research program "Serious form of Charcot-Marie disease - tooth: a collaborative project for the extension of molecular analysis of new genes ".

Projects

• "Il paesaggio della Bonifica" - Ente finanziatore Fondazione CariCal (2019/2020)-**project participant**.

• "DNA on disk: platform and diagnostic kits for human health in oncology, neurology and infectious diseases and poverty-related diseases." Training program for technical and scientific personnel. Alisei Cluster Project. Code CTN01_00177_817708 (2014/2016)-**project participant**.

• "A multidisciplinary diagnostic approach for neurodegenerative diseases due to aging" (2014/2017)-**project participant**.

• "Advanced diagnostics of hereditary diseases of the nervous system" - Module: "Genetics of hereditary peripheral neuropathies" at the Institute of Neurological Sciences of Mangone (CS)-2005/2009-**project participant**.

• FIRB research project - 2007 code RBIP06PMF2_006 - “Study and treatment of tumors and degenerative diseases: development and production of a new analytical platform in DHPLC (Denaturing Hight Performance Liquid Chromatography) complete with diagnostic tests dedicated to the different application sectors in oncology and in degenerative diseases "-**project participant**.

• Telethon project n. GUP04009C - two-year 2006 call - Telehon multi-center project - "Severe Charcot-Marie-Tooth disease and related hereditary neuropathies: an Italian collaborative network for implementing the molecular analysis of rare forms and new genes"-**project participant**.

• Finalized Ministry of Health research - 2006 - "A national network for the study of cerebellar spino ataxias and hereditary spastic paraparesis in Italy"-**project participant**.

• Telethon project n. GGP05165 - 2005 two-year notice - Telehon multi-center project - “Genetics of primary late-onset dystonia”-**project participant**.

• Research project Fondazione Mariani - Grant R-05-44 - year 2005 - "Severe childhool-onset Charcot-Marie-Tooth disease: a collaborative project for implementing the molecular analysis of new genes"-**project participant**.

• From 2005 to 2017, he has collaborated on the following research projects:

- "Molecular genetics of Charcot-Marie-Tooth disease";

- "Molecular genetics of CADASIL";

- "Molecular genetics of hereditary peripheral neuropathies and hereditary spastic paraparesis".

• Scientific cooperation agreement CNR / ASA (Albania)-Peripheral hereditary neuropathies in the Albanian populatio-: genotype-phenotype correlation (2003-2004 biennium).

• Research aimed at the Ministry of Health - year 2002 "Clinical and genetic study of CADASIL patients".

• Linkage study in hereditary type 2 neuropathies and in hereditary spastic paraparesis and Molecular

genetics of hereditary peripheral neuropathies and hereditary spastic paraparesis.

• 2001-2003 to date: Linkage study in hereditary type 2 neuropathies and in hereditary spastic paraparesis and Molecular genetics of hereditary peripheral neuropathies and hereditary spastic paraparesis.

• 2000: Genetic-molecular analysis of the Notch3 gene in families affected by CADASIL;

• 2000: Genotype-phenotype correlations in type 1 demyelinating hereditary neuropathies (CMT1A);

• 2000: Genetic studies in hereditary neuropathies with focal myelin thickening;

• 2000: Linkage study in dominant hereditary neuropathies type 2 (CMT2);

• 2000: Genetic-molecular analysis of the Notch 3 gene in families affected by CADASIL;

• 1999: Genotype-phenotype correlations in type 1 demyelinating hereditary neuropathies (CMT1A);

• 1999: Linkage study in dominant hereditary neuropathies type 2 (CMT2);

• 1999: Genetic studies in hereditary neuropathies with focal myelin thickening;

• 1998: Genotype-phenotype correlations in type 1 demyelinating hereditary neuropathies;

• Project n. 1-1998: Linkage study in dominant hereditary neuropathies type 2 (CMT 2) and in recessive hereditary demyelinating neuropathies (CMT 4B);

• 1996: Molecular genetic study of subjects at risk for familial thrombophilia;

• 1995: Molecular-genetic study of subjects at risk for familial thrombophilia;

• 1994: Genetic-molecular characterization of hemophilia A in Calabrian subjects.

Seminars "The genetics of peripheral neuropathies" as part of the seminar activity for Participants in the Research Doctorate in Medical Biotechnology, Magna Graecia University of Catanzaro, PON 2000-2006 Scientific Research, Technological Development and Higher Education.