Curriculum vitae

PERSONAL INFORMATION

Angela Magariello

(italy)

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Sex Female | Nationality Italian

POSITION

Researcher

WORK EXPERIENCE

May 2018–Present

Permanent Researcher

Institute for Agricultural and Forest Systems in the Mediterranean (ISAFoM) of National Research Council (CNR), Rende (Italy)
www.isafom.cnr.it

Studies on the history of medicine and ethnomedicine with particular attention to the natural remedies used by Italian folk medicine for the treatment of different diseases.

Search for methods to protect biodiversity and safeguard rare species in Mediterranean ecosystems.

Business or sector Public Research

Feb 2010–May 2018

Permanent Researcher

Institute of Neurological Sciences (ISN) of National Research Council (CNR) Piano Lago di Mangone (CS), Italy
www.isn.cnr.it

Identification of genetic defects in subject with inherited diseases of the nervous system.

Business or sector Public Research

Sep 2007–Jan 2010

Term-based Researcher

Institute of Neurological Sciences (ISN) of National Research Council (CNR), Piano Lago di Mangone (CS), Italy

Molecular study of hereditary diseases of the nervous system. Mutational analysis and genotype-phenotype correlation study.

Business or sector Public Research

May 2007–Aug 2007

Research assistant (Research Grant)

Institute of Neurological Sciences (ISN) of National Research Council (CNR), Piano Lago di Mangone (CS), Italy

Molecular analysis of new genes in severe forms of Charcot-Marie-Tooth disease.

Business or sector Public Research

Apr 2005–Apr 2007

Term-based Researcher

Institute of Neurological Sciences (ISN) of National Research Council (CNR), Piano Lago di Mangone (CS), Italy

Molecular study of hereditary diseases of the nervous system with particular attention to: Hereditary Spastic Paraparesis, Peripheral Neuropaties, Huntington Disease, Neurofibromatosis, Amyotrophic Lateral Sclerosis,CADASIL and Spinal Muscular Atrophy.

Business or sector Public Research

2004–2005 **Laboratory tutor (25 hours)**

University of Calabria , Arcavacata di Rende (CS) Italy

Mar 2004–Mar 2005 **Research assistant (Research Grant)**

Institute of Neurological Sciences (ISN) of National Research Council (CNR), Piano Lago di Mangone (CS), Italy

Genetic of Neurofibramatosis.

Business or sector Public Research

EDUCATION AND TRAINING

21 Jan 2004 **PhD - Neuromuscular and Genetic Encephaloneuromyopathic Diseases (3 years)**

University of Messina, Italy

Experimental thesis entitled: *Clinical and genetic study in a family affected by a new form of hereditary dominant complicated spastic paraparesis.*

2000–2001 **Fellowship post-degree**

Institute of Experimental Medicine and Biotechnology (IMSEB) of the CNR, Piano Lago di Mangone (CS), Italy

Training in the use of different bio-molecular techniques for the identification of genetic defects in patients with neurological diseases

1998–1999 **Professional Collaboration**

Institute of Experimental Medicine and Biotechnology (IMSEB) of the CNR, Piano Lago di Mangone (CS), Italy

Screening in the territory of patients with hereditary neuropathy

1998 **Qualification for the profession of biologist**

University of Calabria , Arcavacata di Rende (CS) Italy

28 Feb 1997 **Graduated cum laude in Biological Sciences**

University of Calabria , Arcavacata di Rende (CS) Italy

Experimental thesis entitled: *Identification of an insertion / deletion polymorphism in the promoter of the gene responsible for Huntington's disease.*

PERSONAL SKILLS

Mother tongue(s) Italian

Foreign language(s)

	UNDERSTANDING		SPEAKING		WRITING
	Listening	Reading	Spoken interaction	Spoken production	
English	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

Communication skills

Good communication skills gained as scientific speaker in courses and conferences.

Organisational / managerial skills

Good organisational skills acquired as collaborator in the creation of scientific events.

Job-related skills

TECHNICAL SKILLS

Biomolecular skills

Molecular biology techniques: nucleic acid extractions (DNA and RNA) from biological materials of different origin (fresh tissue and cells); DNA amplification by PCR (polymerase chain reaction); digestion of genomic DNA with restriction endonucleases; horizontal and vertical electrophoresis with denaturing and non-denaturing gels; quantization of nucleic acids by spectrophotometry and fluorimetry.

Mutational analysis techniques: RFLP (Restriction Fragment Lenght Polymorphysm), SSCP (single strand conformation polymorphism), RDB (reverse dot blot), DGGE (denaturing gradient gel electrophoresis), PFGE (Pulsed Field Gel Electrophoresis), chromatographic techniques (HPLC, DHPLC), direct automatic sequencing (Sanger method and cycle Sequencing) and microsatellite (STR//VNTR) analysis. Southern blot for analysis of genomic deletions, methods for marking oligonucleotide probes, hybridization with radioactive and non-radioactive techniques, linkage analysis, in silico analysis of candidate genes, analysis of nucleotide sequences deposited in databases. MLPA analysis (Multiple Ligation/dependent Probe Amplification) for the identification of gene rearrangements (large deletions / duplications). Analysis of data derived from third-generation sequencing systems (Next Generation Sequencing)

Gene expression analysis techniques: Northern Blot, RT-PCR (reverse transcriptase-PCR), Nested RT-PCR, semi-quantitative RT-PCR, quantitative RT-PCR.

Protein analysis techniques: protein extraction from cells in suspension, tissue and biological fluids. Protein dosage, Western blot, tryptic digestion in gel, separation of proteins and peptides through chromatographic systems. Peptide analysis by mass spectrometry (Maldi Tof system).

Microbiological skills

Culture techniques**:** Growth and manipulation of cell cultures in suspension and "in vitro" in a sterile environment; growth curves, isolation and conservation of bacterial strains

Cloning techniques: screening and isolation of clones, preparation of competent cells, transformation by electroporation and by calcium chloride, construction of plasmid expression vectors. Preparation of small and large-scale plasmid DNA (Miniprep, Maxiprep), Ibridation colony, preparation of cell extracts.

Digital skills

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

Digital skills - Self-assessment grid

Good knowledge in the use of the following database:

http://www.ncbi.nih.gov

http://www.ensembl.org
http://www.genome.wi.mit.edu
http://insertion.stanford.edu/dhplc.html
http://genome-www2.stanford.edu/cgi-bin/SGD/web-primer
http://genetics.bwh.harvard.edu/pph/
http://sift.jcvi.org
http://www.fruitfly.org/seq_tools/splice.html
http://robetta.bakerlab.org/
http://genetics.bwh.harvard.edu/pph2/
http://snps-and-go.biocomp.unibo.it/snps-and-go/ http://www.pantherdb.org/tools/
http://www.genome.ucsc.edu.

Good knowledge in the use of the following informatics tools:

Microsoft Office™ tools (Word™, Excel™ and PowerPoint™)
Sequencing analysis software
Gene mapper analysis software

Other skills: I am passionate about music and cinema and I love travelling .

Driving licence: B

ADDITIONAL INFORMATION

Conferences

- XXXV National Congress of the Italian Association of Neuropathology, Lipari, 27-28 May 1999
- IV SIGU National Congress, Orvieto, 28-29 November 2001
- V SIGU National Congress, Verona, 24-27 September 2002
- VII SIGU National Congress, Pisa, 13-15 October 2004
- IX National Congress of the Italian Society of Human Genetics (S.I.G.U.), Palazzo del Cinema, Lido di Venezia, 8-10 November 2006.
- European Congress of Human Genetics 2007, Nice, 16-19 June 2007
- XXXVIII Congress of the Italian Society of Neurology, SIN, Florence, 13-17 October 2007
- XII National Congress of the Italian Society of Human Genetics (S.I.G.U.), Turin, 8-10 November 2009
- XL Congress of the Italian Society of Neurology, SIN, Padua, 23-24 November 2009
- Conference "A look at neurodegenerative diseases" Cosenza 24 June 2011.
- European Congress of Human Genetics 2013, Paris, 8-11 June 2013
- XLV Congress of the Italian Society of Neurology, SIN, Cagliari, 11-14 October 2014
- the 5th edition of the Convention of the International School of Functional Genomics. Catania 28-30 September 2015

Courses

- the XI Residential Medical Genetics Course, San Giovanni Rotondo, 14-16 June 2001

- the "Genetic Expression" course, Pisa, 16 October 2004
- the "ABI PRISM 3100 AVANT" Course Rome, 01-03 December 2004the ABI PRISM 7900 HT course. Cosenza, 16-17 April 2007
- the course "RNA Interference and integrated Sciences Solutions for SNP Genotyping and Gene Expression" Arcavata 18. April 2007
- the training seminar More recent developments in Gnomics and Proteomics, Mangone, 17 July 2007
- the OiSE school English language training course, London, 09-13 June 2008
- the theoretical-practical update course "Genetic amplification in end-point PCR and its evolution to qPCR. Mangone, 17-18 June 2008
- the microRNA profiling course and AB solution for Gene Expression and Genotyping, Cosenza, 27 January 2009
- the course Imaging of the sympathetic innervation of the heart in parkinson's and heart failure, Cosenza 18 June 2010

Projects

- " Il Paesaggio della Bonifica" Ente finanziatore Fondazione Carical (2019-2020). **Project Participant.**
- Specialist medical genetics and radiology services for the Calabria Region (2014-2017). **Project Participant.**
- Specialist medical genetics services outside the region (2014-2017)**. 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-scientific personnel". ALISEI Cluster Project-CTN01_00177_817708 (2014-2016). **Project Participant.**
- Aging project WP2.4: A multidisciplinary diagnostic approach for neurodegenerative diseases of aging (2014-2017). **Project Participant.**
- Financing Research Activity-ISN-CNR.Advanced diagnostics of hereditary diseases of the nervous system - Module: Genetics of hereditary peripheral neuropathies at (2005-2009). **Project Participant.**
- Finalized Ministry of Health research project - "A national network for the study of spino cerebellar ataxias and hereditary spastic paraparesis in Italy"(2007-2010). **Project Participant.**
- Telethon project - Telethon 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"(2006-2008). **Project Participant.**
- Financing Research Activity-ISN-CNR-Genetic of Peripheral Hereditary Neuropathy (2005-2011). **Project Participant.**
- Financing Research Activity-ISN-CNR-Molecular Genetic of Hereditary Spastic Paraparesis (2004-2006). **Project Participant.**
- Financing Research Activity-ISN-CNR-Linkage study in hereditary type 2 neuropathies and in hereditary spastic paraparesis and Molecular genetics of hereditary peripheral neuropathies and hereditary spastic paraparesis (2001). **Project Participant.**
- Financing Research Activity-ISN-CNR-Linkage study in dominant hereditary neuropathies type 2 (CMT2) (2000). **Project Participant.**

Publications

1. Kinesins in neurological inherited diseases: a novel motor-domain mutation in KIF5A gene in a patient from Southern Italy affected by hereditary spastic paraplegia. Citrigno L, **Magariello A**, Pugliese P, Di Palma G, Conforti FL, Petrone A, Muglia M.Acta Neurol Belg. 2018 Dec;118(4):643-646. doi: 10.1007/s13760-018-1039-0. Epub 2018 Nov

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. doi: 10.2174/1389202919666180404105451.

3. Exome sequencing reveals two FA2H mutations in a family with a complicated form of Hereditary Spastic Paraplegia and psychiatric impairments. **Magariello** A, Russo C, Citrigno L, Züchner S, Patitucci A, Mazzei R, Conforti FL, Ferlazzo E, Aguglia U, Muglia M. J Neurol Sci. 2017 Jan 15;372:347-349. doi: 10.1016/j.jns.2016.11.069

4. A novel KIF5A mutation in an Italian family marked by spastic paraparesis and congenital deafness.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.J Neurol Sci. 2014 Aug 15;343(1-2):218-20.

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

6. 5-HTTLPR, anxiety and gender interaction moderates right amygdala volume in healthy subjects.Cerasa A, Quattrone A, Piras F, Mangone G, **Magariello** A, Fagioli S, Girardi P, Muglia M, Caltagirone C, Spalletta G.Soc Cogn Affect Neurosci. 2014 Oct;9(10):1537-45.

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

8. Ataxin-1 and ataxin-2 intermediate-length PolyQ expansions in amyotrophic lateral sclerosis.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. Neurology. 2012 Dec 11;79(24):2315-20.

9. Amyotrophic lateral sclerosis: a new missense mutation in the SOD1 gene.Tortelli R, Conforti FL, Cortese R, D'Errico E, Distaso E, Mazzei R, Ungaro C, **Magariello** A, Gambardella A, Logroscino G, Simone IL.Neurobiol Aging. 2013 Jun;34(6):1709.e3-5.

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

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

12. FUS mutations in sporadic amyotrophic lateral sclerosis: clinical and genetic analysis.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.Neurobiol Aging. 2012 Apr;33(4):837.

13. MAO A VNTR polymorphism and amygdala volume in healthy subjects.Cerasa A, Quattrone A, Gioia MC, **Magariello** A, Muglia M, Assogna F, Bernardini S, Caltagirone C, Bossù P, Spalletta G.Psychiatry Res. 2011 Feb 28;191(2):87-91

14. TARDBP gene mutations in south Italian patients with amyotrophic lateral sclerosis.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. J Neurol Neurosurg Psychiatry. 2011 May;82(5):587-8. doi: 10.1136/jnnp.2009..

15. A novel NF1 gene mutation in an Italian family with neurofibromatosis type 1.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.Childs Nerv Syst. 2011 Apr;27(4):635-8.

16. Morphological correlates of MAO A VNTR polymorphism: new evidence from cortical thickness measurement.Cerasa A, Cherubini A, Quattrone A, Gioia MC, **Magariello** A, Muglia M, Manna I, Assogna F, Caltagirone C, Spalletta G.Behav Brain Res. 2010 Jul 29;211(1):118-24.

17. Mutation analysis of the SPG4 gene in Italian patients with pure and complicated forms of spastic paraplegia.**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.J Neurol Sci. 2010 Jan 15;288(1-2):96-100.

18. Spastic paraplegia with thinning of the corpus callosum and white matter abnormalities: further mutations and relative frequency in ZFYVE26/SPG15 in the Italian population.Denora PS, Muglia M, Casali C, Truchetto J, Silvestri G, Messina D, Boukrhis A, **Magariello** A, Modoni A, Masciullo M, Malandrini A, Morelli M, de Leva MF, Villanova M, Giugni E, Citrigno L, Rizza T, Federico A, Pierallini A, Quattrone A, Filla A, Brice A, Stevanin G, Santorelli FM.J Neurol Sci. 2009 Feb 15;277(1-2):22-5.

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

20. Further evidence that D90A-SOD1 mutation is recessively inherited in ALS patients in Italy.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.Amyotroph Lateral Scler. 2009 Feb;10(1):58-60.

21. MAO A VNTR polymorphism and variation in human morphology: a VBM study.Cerasa A, Gioia MC, Labate A, Lanza P, **Magariello** A, Muglia M, Quattrone A.Neuroreport. 2008 Jul 16;19(11):1107-10.

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

23. Ventro-lateral prefrontal activity during working memory is modulated by MAO A genetic variation.Cerasa A, Gioia MC, Fera F, Passamonti L, Liguori M, Lanza P, Muglia M, **Magariello** A, Quattrone A.Brain Res. 2008 Mar 27;1201:114-21.

24. Genetically dependent modulation of serotonergic inactivation in the human prefrontal cortex.Passamonti L, Cerasa A, Gioia MC, **Magariello** A, Muglia M, Quattrone A, Fera F.Neuroimage. 2008 Apr 15;40(3):1264-73.

25. First evidence of a pathogenic insertion in the NOTCH3 gene causing CADASIL.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.J Neurol Neurosurg Psychiatry. 2008 Jan;79(1):108-10.

26. A novel point mutation in PMP22 gene in an Italian family with hereditary neuropathy with liability to pressure palsies. Muglia M, Patitucci A, Rizzi R, Ungaro C, Conforti FL, Gabriele AL, **Magariello** A, Mazzei R, Motti L, Sabadini R, Sprovieri T, Marcello N, Quattrone A.J Neurol Sci. 2007 Dec 15;263(1-2):194-7

27. A novel Angiogenin gene mutation in a sporadic patient with amyotrophic lateral sclerosis from southern Italy.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**.** Neuromuscul Disord. 2008 Jan;18(1):68-70

28. Gene symbol: NOTCH3.Mazzei R, Conforti FL, Ungaro C, Liguori M, Sprovieri T, Patitucci A, **Magariello** A, Gabriele AL, Muglia M, Quattrone A.Hum Genet. 2007 Apr;121(2):296. No abstract available.

29. Gene symbol: NOTCH3.Mazzei R, Conforti FL, Ungaro C, Liguori M, **Magariello** A, Gabriele AL, Patitucci A, Sprovieri T, Muglia M, Quattrone A.Hum Genet. 2007 Apr;121(2):296. No abstract available.

30. Gene symbol: NOTCH3.Mazzei R, Conforti FL, Ungaro C, Liguori M, Sprovieri T, Patitucci A, **Magariello** A, Gabriele AL, Muglia M, Quattrone A.Hum Genet. 2007 Apr;121(2):295. No abstract available.

31. Putative role of specific JAG1 gene exons in modulating clinical features in patients with leukoencephalopathy.Ungaro C, Sprovieri T, Conforti FL, Muglia M, Patitucci A, **Magariello** A, Gabriele AL, Quattrone A, Mazzei R.Neurosci Lett. 2007 May 11;418(1):1-3

32. Gene symbol 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.

33. Gene symbol Conforti FL, Mazzei R, Patitucci, **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.

34. Sporadic ALS is not associated with VAPB gene mutations in Southern Italy F.L. Conforti, T. Sprovieri, R. Mazzei, C. Ungaro, A. Tessitore, G. Tedeschi, A. Patitucci, **A. Magariello**, A.L. Gabriele, V. Labella, I.L. Simone, G. Majorana, P. Valentino, M. Muglia, A. Quattrone Journal of Negat Results in BioMed. 2006 May 29; 5(1):7

35. Novel spastin (SPG4) mutations in Italian patients with hereditary spastic paraplegia **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. Neuromuscul Disord. 2006 Jun;16(6):387-90

36. Monoamine Oxidase-A Genetic Variations Influence Brain Activity Associated with Inhibitory Control: New Insight into the Neural Correlates of Impulsivity Passamonti L, Fera F, **Magariello A**, Cerasa A, Gioia MC, Muglia M, Nicoletti G, Gallo O, Provinciali L, Quattrone A. Biol Psychiatry. 2005 Sep 30**.**

37. Comparison Of Different Techniques In Detecting CMT1A/HNPP Duplication/Deletion,Alessandra Patitucci, PhD Maria Muglia, PhD, Anna Lia Gabriele, PhD, **Angela Magariello**, PhD, Giuseppina Peluso, PhD, Teresa Sprovieri, PhD, Francesca Luisa Conforti, PhD, Rosalucia Mazzei, PhD, Carmine Ungaro, PhD, Francesca Condino, Paola Valentino, MD, Franco Bono, MD, Carmelo Rodolico, MD, Anna Mazzeo, MD, Antonio Toscano, MD, Aldo Quattrone, MD, Neuromuscular Disorder, 15/7, pp. 488-492.

38. Furthern evidence of heterogeneity in autosomal in autosomal dominant distal motor neuropathy 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.*"*. Neuromuscular Disorder, 2004;14(11):705-10

39. A new SBF2 mutation in a familywith recessive demyelinating Charcot-Marie-Tooth (CMT4B2). 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. "Neurology, 2004,63(7):1327-8

40. A novel Notch3 gene mutation not involving a cysteine residue in an italian family with *cadasil.* 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. "Neurology , 2004; 63(3): 561-4

41. Abnormally high levels of SOD1 mRNA in a patient with Amyotrophic Lateral Sclerosis Conforti F.L., **Magariello A**., Mazzei R., Sprovieri T., Patitucci A., Gabriele A.L., Crescibene L., Bastone L., Muglia M., Quattrone A. Muscle & Nerve 2004; 29(4): 610-611

42. Gene conversion events in adult-onset Spinal Muscular Atrophy 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", Acta Neurol Scan. 2004 Feb; 109(2):151-4.

43. Narrowing of the critical region in autosomal recessive spastic paraplegia linked to the SPG5 locus 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.". Neurogenetics. 2003; 5(1): 49-54

44. A Simple Method for Diagnosis of Autosomal Recessive Spinal Muscular Atrophy by DHPLC. R Mazzei, PhD; Francesca Luisa Conforti, PhD; Maria Muglia, PhD; Teresa Sprovieri, PhD; Alessandra Patitucci, PhD; **Angela Magariello**, PhD; Anna Lia Gabriele, PhD; Aldo Quattrone MD. J Child Neurol. 2003 Apr;18(4):269-71

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

46. A large family with pure autosomal dominant hereditary spastic paraplegia from southern Italy mapping to chromosome 14q11.2-q24.3.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. J Neurol 2002 Oct;249(10):1413-6

47. Rett syndrome phenotype following infantile acute encephalopathy Fiumara A., Polizzi A:,Mazzei R Conforti L., **Magariello A**., Sorge G., Pavone L.. J Child Neurol 2002Sep;17(9):700-2

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