Curriculum Vitae et Studiorum

Francesca Luisa Conforti

Professore Associato di Genetica Medica (MED/03)

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Istruzione e formazione

2021 **Abilitazione Scientifica Nazionale per Professore di I Fascia** per il Settore Concorsuale 06/N1 Scienze Delle Professioni Sanitarie e delle Tecnologie Mediche Applicate (Bando D.D. 553/2021).

2014 **Abilitazione Scientifica Nazionale per Professore di II Fascia** per il Settore Concorsuale 06/A1 - GENETICA MEDICA

29/10/ 1997 **Specializzazione in Patologia Clinica** presso l'Università degli Studi di Roma "La Sapienza" con votazione di 70/70 e Lode. Titolo della tesi: "Studio genetico e clinico di famiglie SCA1 e SCA2 provenienti dall'Italia meridionale".

1993-1995 Borsa di Studio Biennale CNR "Studio delle forme delezionali e non delezionali delle mutazioni nelle sindromi α-thalassemiche". Istituto di Scienze Neurologiche (già Istituto di Medicina Sperimentale e Biotecnologie), CNR – Mangone (CS); prot. 067567 del 26/06/96

1992 **Abilitazione alla professione di Biologo.** Università degli Studi della Calabria, Rende (CS); prot. N.258 del 3/03/1993. Iscrizione all'Albo Professionale dei Biologi 26/03/1993, matr EA_016163 .

20/03/ 1991 **Laurea in Scienze Biologiche** con 110/110 e Lode. Tesi: "Effetto del fattore di crescita epidermico (EGF) sul sistema di trasporto per aminoacidi neutri in coltura". Università degli Studi "La Sapienza" di Roma

Attività professionale ed esperienza lavorativa

Da gennaio 2019 **Responsabile del Laboratorio di Genetica Medica** C/o Centro Sanitario, Dipartimento di Farmacia e Scienze della Salute e della Nutrizione, Università della Calabria, Rende (CS)

Dal 31/12/2018 **Professore Associato in Genetica Medica** presso il Dipartimento di Farmacia e Scienze della Salute e della Nutrizione, Università della Calabria, Rende (CS)

2001- 2018 **Ricercatore III livello professionale,** settore Scientifico-disciplinare: Med/03 Genetica Medica, presso l'Istituto di Scienze Neurologiche (ISN) del CNR di Mangone (CS).

2001-2018 **Responsabile d**ello studio molecolare di patologie neurodegenerative quali la Sclerosi laterale Amiotrofica, la Sindrome di Rett presso l'ISN, CNR di Mangone (CS). Attestato CNR prot. 2083 del 11/11/2013.

1998-2001 **Collaboratore Tecnico Enti di Ricerca** a tempo indeterminate presso l' ISN (già Istituto di Medicina Sperimentale e Biotecnologie, IMSEB), CNR. Ambito lavorativo: Genetica molecolare delle Malattie ereditarie del Sistema Nervoso

1998 Incarico di consulenza professionale esterna. Responsabile progetto "Screening nel territorio di soggetti affetti da patologie neurodegenerative" ISN (già IMSEB), CNR; Contratto del 02/01/1998.

1996-1997 Incarico di consulenza professionale. Responsabile progetto "Creazione di una Banca Dati Genetica Calabrese" Facoltà di Medicina, Dipartimento di Medicina Sperimentale e Clinica, Università degli studi di Catanzaro; Contratto del 15/02/1996.

Consorzi o gruppi scientifici, ruoli ricoperti

Responsabile del Laboratorio di Genetica Medica, Università della Calabria, Rende (CS)
Membro del gruppo di studio MND (Motor Neuron Disease) della Società Italianadi Neurologia (SIN).
Membro della Commissione tecnico-scientifico AISLA (Associazione italianaSclerosi Laterale Amiotrofica)
Membro del consorzio ITALSgen (Consorzio Nazionale Centri Ricerca per laSclerosi Laterale Amiotrofica)
Responsabile sezione di genetica SLA afferente all'Istituto di ScienzeNeurologiche del CNR di Mangone (CS).

Collaborazioni Scientifiche e Accordi di collaborazione

- National Institute on Aging (NIH), Laboratory of Neurogenetics, Bethesda, MD, United States
- Department of Neuromuscular Diseases, UCL Queen Square Institute of Neurology
- ALS Clinical Research Center and Neurochemistry Laboratory, BioNeC, University of Palermo, Italy
- Dipartimento di Scienze Biomediche, Chirurgiche, Neurologiche, Metaboliche e dell'Invecchiamento, Università degli Studi della Campania, Napoli, Italy
- 'Rita Levi Montalcini' Department of Neuroscience, University of Turin, Turin, Italy
- Accordo di collaborazione con l'Azienda Ospedaliero Universitaria Policlinico "G. Rodolico – S. Marco" – U O.C. di Clinica Neurologica – Dipartimento ad attività integrata delle Scienze Neurologiche e della Ricerca Neurobiologica, Catania.

Elenco Pubblicazioni

1. Muglia M., Leone O., Annesi G., Gabriele A.L., Imbrogno E., Grandinetti C., Conforti F.L., Naso F., Brancati C. "A new non-isotopic method for accurate detection of (CAG)n repeats causing Huntington's disease". Clin Chem. 1996; 42 (10): 1601-03.
2. Leone O., Muglia M., Gabriele AL., Annesi G., Conforti F.L., Imbrogno E., Imbrogno L., Brancati C. "Analysis of the (CAG)n repeat at IT15 locus in a population from Calabria. Hum. Biol. 1996; 69/5: 653-662.
3. Annesi G., Muglia M., Conforti F.L., Leone O., Grandinetti C., Imbrogno E., Gabriele A.L., Naso F., Brancati C. "A simple and rapid non-isotopic method for sizing CAG repeats in the SCA1 gene". Human Heredity, 1997;47 (1): 47-51.
4. Muglia M., Leone O., Annesi G., Gabriele A.L., Imbrogno E., Grandinetti C., Conforti F.L., Naso F., Brancati C. To the Editor Clin Chem. 1997; 43(8): 1463.
5. Gambardella A., Annesi G., Bono F., Spadafora P., Valentino P., Pasqua A., Mazzei R., Montesanti R., Conforti F.L., Oliveri RL., Zappia M., Aguglia U. e Quattrone A. "CAG repeat lenght and clinical features in three italian families with spinocrebellar ataxia type 2 (SCA2)". J Neurol 1998; 245:647-652.
6. Gambardella A., Valentino P., Annesi G., Oliveri RL., Mazzei R., Conforti F.L., Bono F., Aguglia U., Zappia M., e Quattrone A. "Hyperekplexia in a patient with brainstem vascular anomaly". Acta Neurol Scand 1999; 98:1-5.

7. Bolino, E. R. Levy, M. Muglia, F.L. Conforti, E. LeGuern, M. A.M. Salih, D-M. Georgiou, R. K. Christodoulou, I. H.-Petrusewicz, P. Mandich, A. Gambardella, A. Quattrone, M. Devoto, A.P. Monaco. "Genetic refinement and physical mapping of the CMT4B gene on chromosome 11q22". Genomics 2000 63(2): 271-278.
8. M. Muglia, A. Toscano, A.L. Gabriele, A. Magariello, A. Patitucci, F.L. Conforti, R. Mazzei, C. Rodolico, A. Gambardella, A. Quattrone. "Identification of a new polymorphism in the exon 2 of the Myelin Protein Zero gene". Hum Mutat (Online) 2000 15(3):299.
9. Bolino, M. Muglia, F.L. Conforti, E. LeGuern, M. A.M. Salih, D-M. Georgiou, R. K. Christodoulou, I. H.-Petrusewicz, P. Mandich, Schenone A., Gambardella A., Bono F., Quattrone A., M. Devoto, A.P. Monaco. "Charcot-Marie-Tooth type 4B is caused by mutations in the gene encoding myotubularin-related protein-2." Nat Genet. 2000; 25(1):17-9.
10. M. Muglia, M. Zappia, V. Timmerman, P. Valentino, AL. Gabriele, F.L. Conforti, P. De Jonghe, M. Ragno, R. Mazzei, M Sabatelli, G. Nicoletti, AM. Patitucci, RL. Oliveri, F. Bono, A. Gambardella, and A. Quattrone. "Clinical and genetic study of a large Charcot-Marie-Tooth type 2A family from southern Italy". Neurology. 2001;56(1):100-103.
11. 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;58:1418-1422.
12. 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 Hungtington's disease presenting as progressive myoclonic epilepsy", Neurology, 2001;57:708-711.
13. 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; 51(6):794-5.
14. Conforti FL, Mazzei R, Magariello A, Bravaccio C, Militerni R, Gabriele AL, 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;249:1398-1400.
15. 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;249:1413-1416.
16. Fiumara A, Polizzi A, Mazzei R, Conforti L., Magariello A, Sorge G, Pavone L. "Rett syndrome phenotype following infantile acute encephalopathy." J Child Neurol. 2002;17(9):700-2
17. 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;117A(2):184-7.
18. Mazzei R, Conforti FL, Muglia M, Sprovieri T, Patitucci A, Magariello A, Gabriele AL, Quattrone A. "A simple method for diagnosis of autosomal recessive spinal muscular atrophy by denaturing high-performance liquid chromatography." J Child Neurol. 2003;18(4):269-71.
19. Guidetti D, Casali B, Mazzei RL, Cenacchi G, De Berti G, Zuccoli G, Nicoli D, Conforti FL, Sprovieri T, Pasquinelli G, Brini M "An Italian case of CADASIL with mutation CGC-TCG in codon 1006, exon 19 Notch3 gene." Neurol Sci. 2003;24(6):401-6.
20. 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 Scand. 2004;109(2):151-4.

21. 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. 2004;5(1):49-54.
22. Conforti FL, Magariello A, Mazzei R, Sprovieri T, Patitucci A, Crescibene L, Bastone L, Gabriele A, Scornaienchi M, Ferraro T, Muglia M, Quattrone A. "Abnormally high levels of SOD1 mRNA in a patient with amyotrophic lateral sclerosis." Muscle & Nerve. 2004;29(4):610-611.
23. 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, Cenacchi G, Santoro G, Muglia M, Quattrone A. A novel Notch3 gene mutation not involving a cysteine residue in an Italian family with CADASIL. Neurology. 2004;63(3):561-564.
24. F.L. Conforti, M. Muglia, R. Mazzei, A. Patitucci, P. Valentino, A. Magariello, T. Sprovieri, F. Bono, C. Bergmann, A.L. Gabriele, P. Peluso, R. Nisticò, J. Senderek, and A. Quattrone, "A new SBF2 mutation in a family with recessive demyelinating Charcot-Marie-Tooth (CMT4B2)". Neurology, 2004;63:1327-1328
25. L. Passamonti, M. Muglia, A. Magariello, M. Bellesi, F.L. Conforti, R. Mazzei, A. Patitucci, A.L. Gabriele, T. Sprovieri, G. Peluso, M. Caracciolo, E. Medici, F. Logullo, L. Provinciali, A. Quattrone. "Further evidence of genetic heterogeneity in autosomal dominant distal neuronopathy. Neuromuscular Disord, 2004, 14:705-710.
26. M. Mancuso, FL. Conforti, A. Rocchi, A. Tessitore, M. Muglia, G. Tedeschi, D. Panza, MR. Monsurrò, P. Sola, J. Mandrioli, A. DelCorona, M L. Manca, R. Mazzei, T. Sprovieri, M. Filosto, A. Salviati, P. Valentino, F. Bono, M. Caracciolo, I. Simone, V. La Bella, G. Majorana, G. Siciliano, L. Murri, A. Quattrone "Could mitochondrial haplogroups play a role in sporadic amyotrophic lateral sclerosis?". Neuroscience Letters, 2004, 371:158-162.
27. Dotti M.T., Federico A., Mazzei R., Bianchi S., Scali O., Conforti FL., Sprovieri T., Guidetti D., Aguglia U., Consoli D., Pantoni L., Sarti C., Inzitari D., Quattrone A. "The spectrum of Notch3 Mutations in 28 Italian CADASIL families". J Neurol Neurosurg Psychiatry. 2005;76(5):736-8
28. P. Valentino, F.L. Conforti, M. Zappia, D. Pirritano, A. Nisticò, R. Mazzei, A. Patitucci, T. Sprovieri, A.L. Gabriele, M. Muglia, A. Clodomiro, A. Gambardella, A. Quattrone. "Brachial Amyotrophic Diplegia associated with a novel Sod-1 mutation: a case report" . Neurology. 2005;64(8):1477-8.
29. Patitucci, M. Muglia, A. Magariello A. L. Gabriele, G. Peluso, T. Sprovieri, F. L. Conforti, R. Mazzei, C. Ungaro, F. Condino, P. Valentino, F. Bono, C. Rodolico, A. Mazzeo, A. Toscano, G. Vita, A. Quattrone. "Comparison of different techniques for detecting 17p12 duplication in CMT1A". Neuromuscul Disord. 2005;15(7):488-92.
30. Magariello, M. Muglia, A. Patitucci, R. Mazzei, F. L. Conforti, A.L. Gabriele, T. Sprovieri, C. Ungaro, A. Gambardella, M. Mancuso, G. Siciliano, D. Branca, U. Aguglia, M. V. de Angelis, K. Longo, A. Quattrone "Novel spastin (SPG4) mutations in Italian patients with hereditary spastic paraplegia". Neuromuscul Disord. 2006;16(6):387-390
31. A Tessitore, F Esposito, Mr Monsurrò, S Graziano, D Panza, A Russo, R Migliaccio, Fl Conforti, R Morrone, A Quattrone, F Di Salle, G Tedeschi." Subcortical Motor Plasticity In Patients With Sporadic Als: An Fmri Study". Brain Res Bull. 2006, 15;69(5):489-94.
32. Conforti FL, Sprovieri T, Mazzei R, Ungaro C, Tessitore A, Tedeschi G, Patitucci A, Magariello A, Gabriele A, Labella V, Simone IL, Majorana G, Monsurro MR, Valentino P, Muglia M, Quattrone A. " Sporadic ALS is not associated with VAPB gene mutations in Southern Italy" J Negat Results Biomed. 2006, 29;5:7.

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34. Conforti FL, Mazzei R., Sprovieri T, Ungaro C, Patitucci, Magariello A, , Gabriele AL, Bravaccio C., Muglia M, Quattrone A. "Gene symbol: MECP2". Disease: Rett syndrome. Hum Genet. 2006;119:676.
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". Neurosci Lett. 2007;418(1):1-3.
36. Mazzei R, Conforti FL, Ungaro C, Liguori M, Sprovieri T, Patitucci A, Magariello A, Gabriele AL, Muglia M, Quattrone A. "Gene symbol: NOTCH3". Hum Genet. 2007;121(2):296.
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39. Muglia M, Patitucci A, 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". J Neurol Sci. 2007;263(1-2):194-7.
40. 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, Monsurro MR, Muglia M, Quattrone A. "A novel Angiogenin gene mutation in a sporadic patient with amyotrophic lateral sclerosis from southern Italy". Neuromuscul Disord. 2008; 18(1):68-70
41. 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;79(1):108-10.
42. 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;73(5):486-91
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45. Sprovieri T, Mazzei R, Ungaro C, Citrigno L, Quattrone A, Conforti FL. "Gene symbol: MECP2", Hum Genet 2008; 123:555.
46. FL Conforti; T Sprovieri; R Mazzei; A Patitucci; C Ungaro; S Zoccolella; A Magariello; V La Bella; A Tessitore; G Tedeschi; IL Simone; G Majorana; P Valentino; L Citrigno; A Gabriele; F Bono; MR Monsurro'; M Muglia; A Quattrone Further evidence that D90A-SOD1 mutation is recessively inherited in ALS patients in Italy. Amyotroph Lateral Scler. 2009 Feb;10(1):58-60.
47. Liguori M, Mazzei R, Ungaro C, Simone IL, Gambardella A, Plasmati I, Fera F, Aguglia U, Lanza P, Bono F, Chiumarulo L, Conforti FL, Consoli D, Quattrone A. Conventional MRI and notch3 gene screening in sporadic CADASIL"Conventional MRI and NOTCH3 gene screening in sporadic CADASIL" Neurology. 2009;72(5):469-71.

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52. Mazzei R, Ungaro C, Garreffa G, Conforti FL, Mollo A, Sprovieri T, Servillo P, Blasi V, Gallo O, Cerasa A, Lanza PL, Quattrone A. Clinical, genetic and magnetic resonance findings in an Italian patient affected by L: -2-hydroxyglutaric aciduria. Neurol Sci. 2011; 32(1):95-9. Epub 2010 Sep 22.
53. 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; 27(4):635-8. Epub 2010 Oct 7
54. FL Conforti, W Sproviero, IL Simone, R Mazzei, P Valentino, C Ungaro, A Magariello, A Patitucci, V La Bella, T Sprovieri, G Tedeschi, L Citrigno, AL Gabriele, F Bono, M R Monsurrò, M Muglia, A Gambardella, A Quattrone TARDBP gene mutations in South Italian patients with Amyotrophic Lateral Sclerosis. J Neurol Neurosurg Psychiatry. 2011;82(5):587-8. Epub 2010 Oct 19.
55. Conforti FL, Barone R, Fermo SL, Giliberto C, Patti F, Gambardella A, Quattrone A, Zappia M. Sporadic motor neuron disease in a familial novel SOD1 mutation: incomplete penetrance or chance association? Amyotroph Lateral Scler. 2011;12(3):220-222.
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sporadic amyotrophic lateral sclerosis: clinical and genetic analysis. Neurobiol Aging. 2012 Apr;33(4):837.e1-5. epub 2011

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