Curriculum Vitae

MARINA FRONTALI

Year of birth: 1941

Education

1968 Medical Doctor

1982 Doctor in Medical Genetics

Actual Positions:

Associate Director of research at Istituto di Neurobiologia e Medicina Molecolare CNR;

Teaching Professor of.'Clinical Genetics' at University Tor Vergata, Specialisation School of Medical Genetics, Rome;

Genetic Consultant at Policlinico Tor Vergata, Rome;

Genetic Consultant at Ospedale S. Andrea, Univ. La Sapienza Rome;

Coleader of the Working Group on Genetic Testing and Counseling of the European Huntington Disease Network (EHDN)

Previous Positions

1970-1985 Junior Researcher at the Psychology Institute of the CNR (National Research Council), Section of Comparative Psychology, Roma.

1980 (5 months) Visiting researcher at MRC Unit of Medical Genetics, London, U.K.

1984 (1 month) Visiting researcher at the Department of Medicine, Section of Medical Genetics dell'University of Wales College of Medicine Cardiff, U.K**.**

1985-2001 Senior Researcher at Istituto di Neurobiologia e Medicina Molecolare del CNR, Roma

1989-2002 Genetic Consultant at Centro di Diagnosi Prenatale , Rome University La Sapienza

1989-1995 Genetic Consultant at Istituto di Clinica delle Malattie Nervose e Mentali, Rome University La Sapienza

2001–2008 Director of Research at Istituto di Neurobiologia e Medicina Molecolare, CNR

Responsibility of Research projects :

1982 Principal Investigator of Project 'Contraccezione e fattori riproduttivi nelle coppie con rischio genetico' within Progetto Finalizzato di Medicina Preventiva e Riabilitativa del CNR (contratto n° 82.02176.56)

1987-89 & 1990-93 Italian partner of bilateral project CNR Italia-USA 'Study of RFLP on chromosome 4 in the Italian population and families segregting for Huntington's Disease' with Prof. J.F. Gusella, Neurogenetics Laboratory Massachusetts General Hospital, Boston, as USA partner.

1991-1995 Principal Investigator of Project 'Mappatura genetica del gene per la Atassia Spino-cerebellare tipo 1', within Progetto Finalizzato CNR 'Ingegneria genetica' Subproject 'Malattie ereditarie'

1992 Italian Partner of project EC PL9101033 'Molecular genetic prediction for Huntington's disease - analysis of ethical and social aspects and their implications for other late onset neurogenetic disorders', coordinated by Prof. P.S. Harper Cardiff University, UK.

1992-93 Principal Investigator of project 'Studio e realizzazione di un servizio modello di Consulenza Genetica' supported by USL RM24

1993-95 Principal Investigator of Telethon-Italia project (N°358) 'Molecular bases of late onset autosomal dominant ataxias'

1993-94 ItalianPartner of project EC 'Physical mapping of human chromosome 6 : detailed analysis of certain regions including the Major Histocompatibility Complex' (contract GENE-CT93-0075) coordinated by Prof. J. Trowsdale, Imperial Cancer Research Fund, London, UK

1994-95 Italian Partner of bilateral project CNR Italia-UK ‘Gene mapping of pure cerebellar ataxia’ with prof. A.E. Harding atInstitute of Neurology-National Hospital di London as UK partner

1995-96 ItalianPartner and member of the Steering Committee of EC project ‘Concerted Action to support research on the molecular genetic basis of the late onset autosomal dominant ataxias’, (contract BMH1 CT 941243), coordinated by Dr H. Cann, Centre d’Etude du Polymorphism Humain, Paris (France).

1996-97 Principal Investigator of Telethon-Italia project (N°E355) ‘Molecular basis of late onset autosomal dominant ataxias : refined mapping and identification of the gene for Episodic Ataxia type 2 and study of its relationship with Familial Hemiplegic Migraine and CADASIL genes’.

1996-99 Principal Investigator of one Operative Unit of Project ‘Eredoatassie in Calabria’ coordinated by Prof. Cristoforo Morocutti and supported by Regione Calabria, Italia

1997-98 Principal investigator of Telethon Project .Molecular basis of late onset autosomal dominant a taxias: refined mapping and identification of gene for Episodic Ataxia type 2 and study of its relationship with Familial Hemiplegic migraine and CADASIL genes';

1999-2000 Coordinator of Telethon multicentric Project: .Molecular basis of late onset autosomal dominant ataxias: Episodic Ataxia 2, Spinocerebellar Ataxia 6, and the role of CACNA1A gene mutations';

2003-2004 Principal Investigator of a Unit of Project MIUR-FISR .Neurotrofine e meccanismi relativi a malattie neurodegenerative';

2003-4 Coordinator of multicentric project 'Neurodegenerazione da misfolding delle proteine', supported by the Italian Ministry of Health

2003-5 Principal Invsetigator of a Unit of Project MIUR-FIRB 'Canalopatie genetiche: identificazione di markers moleculari e loro funzione, sviluppo di sistemi diagnostici avanzati, e identificazione di bersagli farmacologici';

2006-7 Principal Investigator of ATAXIA UK project :'Transcript mutation analysis and identification of new CACNA1A gene regions can improve mutation detection in Episodic Ataxia type 2 (EA2)'.

2008-2009 Subcontractor for the European Huntington Disease Network (EHDN) Registry

2007-2010 Principal Investigator of one Unit of the multicenter project:’Effects of tetrahydrobiopterine (BH4) on endothelium-dependent vasodilation in patient affected with CADASIL: a controlled, randomized study’ supported by the Drug Italian Agency AIFA (FARM659PTX AIFA Codice EudraCT 2007-004370-55**)**

Honours

1989 Invited speaker at 13° Workshop of the WFN Research Group on Huntington’s Chorea

1992-93 Expert of the Italian Ministry of Foreign Affairs in the project of Cooperation for Development entitled ‘ A laboratory for the diagnosis and prevention of hereditary disorders’ At Mansura University, El Mansura (Egipt

1993-today Member del ‘Research Group on Huntington’s Disease’, World Federation of Neurology (WFN).

1995-today Member of the Scientific Committee of the Italian Association for research on Dystonias (ARD)

1995-2000 Member of the ‘Committee for the study of Bioethics’ of the Italian National Research Council (CNR)

1997 Chairperson for the session ‘Genetic epidemiology, population genetics, mapping and cloning’ at the 1° International Symposium on Inherited Ataxias, Montreal (Canada) 29-5/1-6 1997

1997 Member of the 'Study Group for the genetic test guidelines' Italian National Committee for biosecurity and bitechnology and Istituto Superiore di Sanita' http://www.iss.it/binary/tege/cont/LG_TG_19980519.1151678979.PDF

1998 Invited speaker at the Royal Society Discussion Meeting 'Glutamine repeats and inherited neurodegenerative diseases: molecular aspects' London 7-8 Oct.1998

1999 Invited speaker on .Episodic Ataxia type 2 and Familial Hemiplegic Migraine' at the 18th International Conference on Biological membranes and "Channelopathies". Troina, 18-20 June 1999.

2002 Invited speaker on 'Behaviour Genetics: IQ and Schizofrenia as models'. International Conference on 'Juridical and Psychosocial Implications of Human Genetics. Roma 21-22 March 2002

2007-today Coleader of the Working Group on Genetic Testing and Counselling, European Huntington Disease Network (EHDN)

2010. Invited speaker at 'Brains in Dialogue on genetic testing' meeting organized by International School of Advanced Study SISSA, Trieste (Italy) 28th-29th January 2010

2010. Invited speaker on 'Genetic counselling: what to say and when' at the EHDN Plenary Meeting, Prague, September 3-5, 2010

2010 Member of the Technical-Scientific Commission, Subcommission Clinical Trials, of AIFA (Italian Drug Agency).

List of publications in International Journal with Impact Factor

1. Bignami G, Amorico L, **Frontali M**, Rosic N. Central cholinergic blockade and two-way avoidance acquisition: the role of response disinhibition. *Physiology and Behavior* 1971, 7, 461-470.

2. **Frontali M**, Bignami G. Go-no go discriminations in rats with simple go and compound no go signals: stimulus modality and stimulus intensity. *Animal Learning and Behavior* 1973, 1, 21-24.

3. **Frontali M**, Bignami G. Stimulus non-equivalences in go-no go avoidance discriminations: sensory, drive and response factors. *Animal Learning and Behavior* 1974, 2, 153-160.
4. **Frontali M**, Ramenghi M, Trabace S, Dallapiccola B. Microcyto-genetics and Langer-Giedion syndrome. *Journal of Medical Genetics* 1982, 19, 390-391.
5. Dallapiccola B, Chessa L, Brinchi V, **Frontali M**, Gandini E. Mating between balanced translocation carriers in two unrelated families. *Human Genetics* 1983, 65, 165-168.
6. **Frontali M**, Stomeo C, Dallapiccola B. Osteoporosis-Pseudoglioma syndrome: report of three sibs and overview. *American Journal of Medical Genetics* 1985, 22, 35-47.
7. **Frontali M**, Dallapiccola B. Osteoporosis-Pseudoglioma syndrome and the ocular form of Osteogenesis Imperfecta. *Clinical Genetics* 1986, 29, 262.
8. Vivona G, **Frontali M**, Di Nunzio ML, Vendemiati A: Aplasia cutis congenita and/or Epidermolysis Bullosa. *American Journal of Medical Genetics* 1987, 26, 497-502.
9. Grasso MG, **Frontali M**, Bernardi S, Pantano P, Fieschi C. Multifactorial inheritance and recurrence risks of multiple sclerosis in Italian patients. *Neuroepidemiology* 1989, 8, 300-307.
10. Novelli G, **Frontali M**, Baldini D, Bosman C, Dallapiccola B, Pachi A, Torcia F. Prenatal diagnosis of adult policystic kidney disease with DNA markers on chromosome 16 and the genetic heterogeneity problem. *Prenatal Diagnosis* 1989, 9, 759-767.
11. Brambilla C, **Frontali M**, Rossi C. On the estimation of the onset age distribution in Huntington's Chorea via the EM algorithm. *Annals of Human Genetics* 1990, 54, 225-233.
12. **Frontali M**, Malaspina P, Rossi C, Jacopini AG, Vivona G, Pergola MS, Palena A, Novelletto

A. Epidemiological and linkage studies on Huntington's disease in Italy. *Human Genetics* 1990, 85, 165-170**.**

13. Novelletto A, Mandich P, Bellone E, Malaspina P, Vivona G, Ajmar F, **Frontali M**. Non-random association between DNA markers and Huntington's disease locus in the Italian population. *American Journal of Medical Genetics* 1990, 40, 374-376.
14. **Frontali M**, Jodice C, Lulli P, Spadaro M, Cappellacci S, Giunti P, Malaspina P, Morellini M, Morocutti C, Novelletto A, Persichetti F, Trabace S, Anastasi R, Terrenato L. Spinocerebellar ataxia (SCA1) in two large kindreds: evidence in favour of a locus position distal to GLO1 and the HLA cluster. *Annals of Human Genetics* 1991, 55, 7-15
15. Zoghbi H, Jodice C, Sandkuijl LA, Kwiatowski TJ Jr, McCall AE, Huntoon SA, Lulli P, Spadaro M, Litt M, Cann H, **Frontali M**, Terrenato L. The gene for autosomal dominant ataxia (SCA1) maps telomeric to the HLA complex and is closely linked to the D6S89 locus in three large kindreds. *American Journal of Human Genetics* 1991, 49, 23-30.
16. **Frontali M**, Spadaro M, Giunti P, Bianco F, Jodice C, Persichetti F, Colazza GB, Lulli P, Terrenato L, Morocutti C. Autosomal dominant pure cerebellar ataxia: neurological and genetic study. *Brain* 1992, 115, 1647-1654
17. MacDonald ME, Novelletto A, Lin C, Tagle D, Barnes G, Bates G, Taylor S, Allitto B, Altherr M, Myers R, Lehrach H, Collins FS, Wasmuth JJ, **Frontali M**, Gusella JF. The Huntington's disease candidate region exhibits many different haplotypes. *Nature Genetics* 1992, 1, 99-103
18. Martino F, D'Eufemia P, Pergola MS, Finocchiaro R, Celli M, Giampà G, **Frontali M**, Giardini O. Child With Manifestations of Dermotrichic Syndrome and Ichthyosis Follicularis-Alopecia-Photophobia (IFAP) Syndrome. *American Journal of Medical Genetics* 1992, 44, 233-236.
19. Narod SA, Parry DM, Parboosingh J, Lenoir GM, Ruttledge M, Fischer G, Eldridge R, Martuza RL, **Frontali M**, Haines J, Gusella JF, Rouleau GA. Neurofibromatosis Type 2 Appears to Be a Genetically Homogeneous Disease. *American Journal of Human Genetics* 1992, 51, 486-492.
20. Spadaro M, Giunti P, Lulli P, **Frontali M**, Jodice C, Cappellacci S, Morellini M, Persichetti F, Trabace S, Anastasi R, Morocutti C. HLA-linked spinocerebellar ataxia: a clinical and genetic study of large Italian kindreds. *Acta Neurologica Scandinavica* 1992, 85, 257-265.
21. Duyao MP, Ambrose CM, Myers RH, Novelletto A, Persichetti F, **Frontali M**, Folstein SE, Ross C, Franz ML, Abbott M, Gray J, Conneally PM, Young A, Penney J, Hollingsworth Z, Shoulson I, Lazzarini AM, Falek A, Koroshetz W, Bird E, Vonsattel JP, Bonilla E, Alvir J, Bickham Conde J, Cha J-H, Dure L, Gomez F, Ramos M, Sanchez-Ramos J, Snodgrass SR, de Young M, Wexler NS, Barnes G, Srinishi J, MacDonald ME, Gusella JF. Trinucleotide repeat length: instability and age of onset in Huntington's disease. *Nature Genetics* 1993, 4, 387-392.
22. European Community Huntington's Disease Collaborative Study Group. Ball D, Harper PS, Tyler A, Tibben A, Vegter-van der Vlis M, Cassiman JJ, Evers-Kiebooms G, **Frontali M**, Jacopini AG. Ethical and social issues in presymptomatic testing for Huntington's Disease: a European Community collaborative study. *Journal of Medical Genetics* 1993, 30, 1028-1035.
23. Jodice C, **Frontali M (corresponding author)**, Persichetti F, Novelletto A, Pandolfo M, Spadaro M, Giunti P, Schinaia G, Lulli P, Malaspina P, Plasmati R, Tola R, Antonelli A, Di Donato S, Morocutti C, Weissenbach J, Cann HM and Terrenato L. The gene for spinal cerebellar ataxia 1 (SCA1) is flanked by two closely linked highly polymorphic microsatellite loci. *Human Molecular Genetics* 1993, 2, 1383-1387.
24. Kwiatkowski TJ, Ranum LPW, Banfi S, McCall AE, Jodice C, LeBorgne-DeMarquoy F, **Frontali M**, Beaudet AL, Terrenato L, Zoghbi HY, Orr HT. The gene for autosomal dominant spinocerebellar ataxia (SCA1) maps centromeric to D6S89 and shows no recombination in nine large kindreds with a dinucleotide repeat at the AM10 locus. *American Journal of Human Genetics* 1993, 53, 391-400.

25. Zoghbi HY, **Frontali M**, Orr HT, Sandkuijl L, Cann H, Sasaki H, Chamberlain S, Terrenato L, Rich SS. Linkage studies in dominantly inherited ataxias. *Advances in Neurology* 1993,61,133-7
26. Giunti P, Sweeney MG, Spadaro M, Jodice C, Novelletto A, Malaspina P, **Frontali M**, Harding AE. The trinucleotide repeat expansion on chromosome 6p (SCA1) in autosomal dominant cerebellar ataxias. *Brain* 1994, 117, 645-649.
27. Jodice C, Malaspina P, Persichetti F, Novelletto A, Spadaro M, Giunti P, Morocutti C, Terrenato L, Harding AE, **Frontali M**. Effect of trinucleotide repeat length and parental sex on phenotypic variation in spinocerebellar ataxia 1. *American Journal of Human Genetics* 1994, 54, 959-965.
28. Novelletto A, Persichetti F, Sabbadini G, Mandich P, Bellone E, Agruar F, Pergola M, Del Senno L, MacDonald ME, Gusella JF, **Frontali M**. Analysis of the trinucleotide repeat expansion in Italian families affected with Huntington disease. *Human Molecular Genetics* 1994, 3, 93-98
29. Novelletto A, Persichetti F, Sabbadini G, Mandich P, Bellone E, Ajmar F, Squitieri F, Campanella G, Bozza A, MacDonald ME, Gusella JF and **Frontali M**. Polymorphism analysis of the huntingtin gene in Italian families affected with Huntington disease. *Human Molecular Genetics* 1994, 3, 1129-1132. Sabbadini G, Francia A, Calandriello L, Di Biasi C, Trasimeni G, Gualdi GF, Palladini G, Manfredi M, **Frontali M**. Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL): clinical, neuroimaging, pathological and genetic study of a large Italian family. *Brain* 1995, 118, 207-215. **Frontali M**, Sabbadini G, Novelletto A, Jodice C, Naso F, Spadaro M, Giunti P, Jacopini Ag, Veneziano L, Mantuano E, Malaspina P, Ulizzi L, Brice A, Durr A, Terrenato L. Genetic fitness in Huntington's Disease and Spinocerebellar Ataxia 1 : a population genetics model for CAG repeat expansions. *Annals of Human Genetics* 1996 60, 423-435.
30. Malaspina P, Roetto A, Trettel F, Jodice C, Blasi P, **Frontali M**, Carella M, Franco B, Camaschella C, Novelletto A. Construction of a YAC Contig Covering Human Chromosome 6p22. *Genomics* 1996, 36, 399-407.
31. Rubinsztein DC, Leggo J, Coles R, Almqvist E, Biancalana V, Cassiman J-J, Chotai K, Connarty M, Crauford D, Curtis A, Curtis D, Davidson MJ, Differ A-M, Dode C, Dodge A, **Frontali M**, Ranen NG, Stine OC, Sherr M, Abbott MH, Franz ML, Graham CA, Harper PS, Hedreen JC, Jackson A, Kaplan J-C, Losekoot M, MacMillan JC, Morrison P, Trottier Y, Novelletto A, Simpson SA, Theilmann J, Whittaker JL, Folstein SE, Ross CA, Hayden MR. Phenotypic Characterization of Individuals with 30-40 CAG Repeats in the Huntington Disease (HD) Gene Reveals HD Cases with 36 Repeats and Apparently Normal Elderly Individuals with 36-39 Repeats. *American Journal of Human Genetics* 1996, 59, 16-22.
32. Trettel F, Malaspina P, Blasi P, Jodice C, Novelletto A, Sabbadini G, Veneziano L, **Frontali M**, Terrenato L. Ordering of 44 Genetic Markers in the 6p22 Cytogenetic Band. *DNA Sequence-The Journal of Sequencing and Mapping* 1996, 7, 51-52
33. Bentivoglio AR, Del Grosso N, Albanese A, Cassetta E, Tonali P, **Frontali M**. Non-DYT1 dystonia in a large Italian family. *Journal of Neurology, Neurosurgery & Psychiatry* 1997, 62, 357-360
34. Calandriello L, Veneziano L, Francia A, Sabbadini G, Colonnese C, Mantuano E, Jodice C, Trettel F, Viviani P, Manfredi M, **Frontali M**. Acetazolamide-responsive episodic ataxia in an Italian family refines gene mapping on chromosome 19p13. *Brain* 1997, 120, 805-812.
35. Giovannone B, Sabbadini G, Di Maio L, Calabrese O, Castaldo I, **Frontali M**, Novelletto A, Squitieri F. Analysis of (CAG)n size heterogeneity in somatic and sperm cell DNA from intermediate and expanded Huntington Disease gene carriers. *Human Mutation*, 1997, 10, 458-464
36. Jodice C, Mantuano E, Veneziano L, Trettel F, Sabbadini G, Calandriello L, Francia A, Spadaro M, Pierelli F, Salvi F, Ophoff RA, Frants RR, **Frontali M**. Episodic Ataxia type 2 (EA2) and

spiniocerebellar ataxia type 6 (SCA6) due to CAG repeat expansion in the CACNA1A gene on Chromosome 10p. *Human Molecular Genetics* 1997, 6, 1973-1978.

37. Giunti P, Sabbadini G, Sweeney MG, David MB, Veneziano L, Mantuano E, Federico A, Plasmati R, **Frontali M**, Wood NW. The role of the SCA2 trinucleotide repeat expansion in 89 ADCA families: Frequency, clinical and genetic correlates. *Brain* 1998, 121, 459-467.
38. Mandich P, Jacopini G, Di Maria E, Sabbadini G, Chimirri F, Bellone E, Novelletto A, Ajmar F, **Frontali M**. Predictive testing for Huntington's disease :ten years' experience in two Italian centres. *Italian Journal Neurolological Sciences* 1998,19, 68-74
39. Mantuano E, Trettel F, Olsen AS, Lennon G, **Frontali M**, Jodice C. Chromosomal localization and genomic structure of human deoxyhypusine syntase gene on chromosome 19p13.1. *Gene* 1998, 215, 153-157
40. Mascalchi M, Tosetti M, Plasmati R, Bianchi MC, Tessa C, Salvi F, **Frontali M**, Valzania F, Bartolozzi C, Tassinari CA. Proton magnetic resonance spettroscopy in an Italian family with spinocrebellar ataxia type 1. *Annals of Neurology* 1998,43,244-52.
41. Cassetta E, Del Grosso N, Bentivoglio AR, Valente EM, **Frontali M**, Albanese A. Italian family with cranial cervical dystonia: clinical and genetic study. *Movement Disorders* 1999, 14, 820-825.
42. **Frontali M**, Novelletto A, Annesi G, Jodice C. CAG repeat insatbility, cryptic sequenze variation and pathogeneicity: evidence from different loci. *Phylosophical Transaction of the. Royal Society B* 1999, 354,1089-1094.
43. Jarman PR, Del Grosso N, Valente EM, Leube B, Cassetta E, Bentivoglio AR, Waddy HM, Uitti RJ, Maraganore DM, Albanese A, **Frontali M**, Auburger G, Bressman SB, Wood NW, Nygaard TG. Primary torsion dystonia : the search for genes is not over. *Journal of Neurology Neursurgery & Psychiatry* 1999, 67, 395-397
44. Albanese A, Bentivoglio AR, Del Grosso N, Cassetta E, **Frontali M**, Valente EM, Tonali P. Phenotype variability of dystonia in monozygotic twins. *Journal of Neurology* 2000,247,148-150
45. Squitieri F, Sabbadini G, Mandich P, Gellera C, DiMaria E, Bellone E, Castellotti B, Nargi E, de Grazia U, **Frontali M**, Novelletto A. Family and molecular data for a fine analysis of age at onset in Huntington Disease. *American Journal of Medical Genetics* 2000, 95, 366-373.
46. Stevanin G, Herman A, Durr A, Jodice C, **Frontali M**, Agid Y, Brice A. Are (CTG)n expansions at the SCA8 locus rare polymorphisms? *Nature Genetics* 2000, 24, 213.
47. Trettel F, Mantuano E, Calabresi V, Veneziano L, Olsen AS, Georgescu A, Gordon L, Sabbadini G, **Frontali M**, Jodice C. A fine map of the gene CACNA1A region on 19p13.1-p13.2 chromosome. *Gene* 2000, 241, 45-50.
48. Frontali M. Spinocerebellar Ataxia type 6: channelopathy or glutamine repeat disorder? *Brain Research Bulletin* 2001, 56, 227-31.
49. Guida S, Trettel F, Pagnutti S, Mantuano E, Tottene A, Veneziano L, Fellin T, Spadaro M, Stauderman KA, Williams ME, Volsen S, Ophoff RA, Frants RR, Jodice C, **Frontali M (corresponding author),** Pietrobon D. Complete loss of P/Q calcium channel activity caused by a CACNA1A missense mutation carried by episodic ataxia type 2 patients. *American Journal of Human Genetics* 2001, 68, 759-764.
50. Valente EM, Bentivoglio AR, Cassetta E, Dixon PH, Davis MB, Ialongo T, Ferraris A, Del Grosso N, **Frontali M**, Wood NW, Albanese A. DYT13, a novel primary torsion dystonia locus, maps to chromosome 1p36.32-36.13 in an Italian family with cranial-cervical or upper limb onset. *Annals of Neurology* 2001, 49, 362-266
51. Valente EM, Bentivoglio AR, Cassetta E, Dixon PH, Davis MB, Ferraris A, Ialongo T, **Frontali M**, Wood NW, Albanese A. Identification of a novel primary torsion dystonia locus (DYT13) on chromosome 1p36 in an Italian family with cranial-cervical or upper limb onset. *Neurological Sciences* 2001, 22, 95-6.

52. Valente EM, Bentivoglio AR, Dixon PH, Ferrari A, Ialongo T, **Frontali M**, Albanese A, Wood NW Localization of a novel locus for autosomal recessive early-onset parkinsonism, PARK6, on human chromosome 1p35-p36. *American Journal of Human Genetics* 2001, 68, 895-900.
53. Djousse L, Knowlton B, Hayden M, Almqvist EW, Brinkman R, Ross C, Margolis R, Rosenblatt A, Durr A, Dode C, Morrison PJ, Novelletto A, **Frontali M**, Trent RJ, McCusker E, Gomez-Tortosa E, Mayo D, Jones R, Zanko A, Nance M, Abramson R, Suchowersky O, Paulsen J, Harrison M, Yang Q, Cupples LA, Gusella JF, MacDonald ME, Myers RH. Interaction of normal and expanded CAG repeat sizes influences age at onset of Huntington disease. *American Journal of Medical Genetics* 2003,119, 279-82
54. Li JL, Hayden MR, Almqvist EW, Brinkman RR, Durr A, Dode C, Morrison PJ, Suchowersky O, Ross CA, Margolis RL, Rosenblatt A, Gomez-Tortosa E, Cabrero DM, Novelletto A, **Frontali M**, Nance M, Trent RJ, McCusker E, Jones R, Paulsen JS, Harrison M, Zanko A, Abramson RK, Russ AL, Knowlton B, Djousse L, Mysore JS, Tariot S, Gusella MF, Wheeler VC, Atwood LD, Cupples LA, Saint-Hilaire M, Cha JH, Hersch SM, Koroshetz WJ, Gusella JF, MacDonald ME, Myers RH. A Genome Scan for Modifiers of Age at Onset in Huntington Disease: The HD MAPS Study. *American Journal of Human Genetics* 2003, 73, 682-7
55. Mantuano E, Veneziano L, Jodice C, **Frontali M**. Spinocerebellar Ataxia type 6 and Episodic Ataxia type 2: differences and similarities between two allelic disorders. *Cytogenetics and Genome Research* (ex Cytogenetics and Cell Genetics) 2003, 100, 147-153.
56. Djoussé L, Knowlton B, Hayden M, Almqvist EW, Brinkman R, Ross CA, Margolis R, Rosenblatt A, Durr A, Dode C, Morrison PJ, Novelletto A, **Frontali M**, Trent RJA, McCusker E, Gómez-Tortosa E, Mayo D, Jones R, Zanko A, Nance M, Abramson R, Suchowersky O, Paulsen J, Harrison M, Yang Q, Cupples LA, Mysore J, Gusella JF, MacDonald ME, Myers RH. Evidence for a Modifier of Onset Age in Huntington's Disease Linked to the HD Gene in 4p16. *Neurogenetics* . 2004;5:109-14.
57. Mantuano E, Veneziano L, Spadaro M, Giunti P, Guida S, Leggio MG, Verriello L, Wood N, Jodice C, **Frontali M**. Clusters of non-truncating mutations of P/Q type Ca^{2+} channel subunit $Ca_v2.1$ causing Episodic Ataxia 2. *Journal of Medical Genetics* 2004;41:e82 (http://jmg.bmjjournals.com/cgi/content/full/41/6/e82).
58. Spadaro M, Ursu S, Lehmann-Horn F, Veneziano L, Antonini G, Giunti P, **Frontali M**, Jurkat-Rott K. A G301R Na+/K+ -ATPase mutation causes familial hemiplegic migraine type 2 with cerebellar signs. *Neurogenetics*. 2004 Sep;5(3):177-85. Erratum in: Neurogenetics. 2005 Sep;6(3):169. Liana, Veneziano [corrected to Veneziano, Liana]; Giovanni, Antonini [corrected to Antonini, Giovanni]; Paola, Giunti [corrected to Giunti, Paola].
59. Li JL, Hayden MR, Warby SC, Durr A, Morrison PJ, Nance M, Ross CA, Margolis RL, Rosenblatt A, Squitieri F, Frati L, Gómez-Tortosa E, García CA, Suchowersky O, Klimek ML, Trent RJ, McCusker E, Novelletto A, **Frontali M,** Paulsen JS, Jones R, Ashizawa T, Lazzarini A, Wheeler VC, Prakash R, Xu G, Djoussé L, Mysore JS, Gillis T, Hakky M, Cupples LA, Saint-Hilaire MH, Cha JH, Hersch SM, Penney JB, Harrison MB, Perlman SL, Zanko A, Abramson RK, Lechich AJ, Duckett A, Marder K, Conneally PM, Gusella JF, MacDonald ME, Myers RH. Genome-wide significance for a modifier of age at neurological onset in Huntington's disease at 6q23-24: the HD MAPS study. *BMC Med Genet*. 2006,17;7:71.
60. Provenzano C, Veneziano L, Appleton R, **Frontali M**, Civitareale D. Functional characterization of a novel mutation in TITF-1 in a patient with benign hereditary chorea. *J Neurol Sci*. 2008 Jan 15;264(1-2):56-62. .
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