CURRICULUM VITAE

of

Ferdinando Squitieri, *M.D., Ph.D. - NEUROLOGIST*

Date of Birth: Novembre 3rd, 1961
Current work address: (1) IRCCS Casa Sollievo della Sofferenza Hospital, Viale Cappuccini, 1 - 71013 San Giovanni Rotondo and CSS-Mendel Institute of Human Genetics, Viale Regina Margherita, 261 - 00198, Roma, Italy Tel.: +39 06 44160.536. (2) Italian Research League of Huntington and related diseases onlus (www.lirh.it), via dei Mille 41/A - 00185, Rome, Italy
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Nationality: Italian
Language: Italian and English
Current Positions: Head, Huntington and Rare Diseases Unit at IRCCS Casa Sollievo della Sofferenza (San Giovanni Rotondo and Rome) and of Neurology at CSS-Mendel Institute of Human Genetics (Rome), Italy; Research Associate of the Italian National Research Council (CNR), Rome; Co-founder and Chief Scientific Officer of LIRH Foundation (Italian League for Research on Huntington and related diseases - www.lirh.it).

JOB TITLES

Sept. 2018 – present: Research Associate of the Italian National Research Council (CNR), Rome
Jun 2016 – present: Head of Neurology at CSS-Mendel Institute of Human Genetics, Rome, Italy
July 2015 – present: Head Huntington and Rare Diseases Unit at IRCCS Ospedale Casa Sollievo della Sofferenza (San Giovanni Rotondo) and CSS-Mendel Institute of Human Genetics (Rome), Italy
2015: Qualified as Associate Professor, Italian Ministry of University (MIUR)
2014 (Feb) – **2016 (Oct)**: Visiting Consultant of Ministry of Health, Royal Hospital, Sultanate of Oman, Muscat, Oman.
1998 (Jan) – 2015 (Jul): Head of the Centre for Neurogenetics and Rare Diseases - Neurological Research Institute IRCCS Neuromed, Italy
2005 – **present**: Member of the European Huntington Disease Network
2008 – **present**: Principal Investigator and Member of the Huntington Study Group
2012 (Dec) – present: Co-founder and Chief Scientific Officer of LIRH Foundation (Italian League for Research on Huntington and related diseases)
2000 - 2003: Head of Laboratory for radioactivity, IRCCS Neuromed
2001 - 2012: Scientific Officer Associazione Italiana Corea di Huntington (AICH-Neuromed)

EDUCATION AND TRAINING

1997 - Post-doctoral Research grant for Post-Doctoral Fellows: Neurobiology; Faculty of Medicine University Federico II, Naples, Italy
1994 (June 29) - Degree for "Use of Radiolabelled isotopes", University of British Columbia, Dept. of Occupational Health and Safety, Vancouver, BC, Canada
1994 (April, stage of six month) - Post-doctoral Research Fellow: Focal Dystonias and Botulinum Toxin therapy; Neurodegenerative Disorders Centre, Division of Neurology, Faculty of Medicine, University Hospital of British Columbia (U.B.C.), Vancouver, BC, Canada (Dr. Joseph K.C. Tsui)

1993-1994 Post-doctoral Research Fellow (Grant Italian CNR-Canadian MRC, PhD grant): Medical Genetics; Dept. of Medical Genetics, University of British Columbia, Vancouver, BC, Canada (Professor Michael R. Hayden)
1991 (Two months: February-April) - Visiting Researcher (Grant CNR-MRC): Neurogenetics and Movement Disorders; Institute for Neurology and Neurosurgery, Queen Square, London, UK (Professor Anita Harding)
1990-1997 Expertise in Clinical Psychiatry. Epydemiological Commettee for Psychiatric Disorders, ASL SA1, Italy
1990-1994 Ph.D in Applied Neurobiology (curricula in Genetics and Epydemiology); Faculty of Medicine, University of Siena, Siena, Italy
1986-1989 Specialization in Neurology; Faculty of Medicine University Federico II, Naples, Italy
1979-1985 Degree in Medicine; Faculty of Medicine University Federico II, Naples, Italy

TEACHING

2014 – 2015 Acting professor of Biology and Genetics, Course for Biomedical Technicians of Medical Laboratories and for Technicians in Radiology, "Sapienza" University of Rome, Italy
2014 Acting professor of Psychology en Bioethics, Course for Biomedical Technicians of Medical Laboratories, "Sapienza" University of Rome, Italy.
1998 – 2014 Acting professor of Medical Genetics (Code F03), Course for Biomedical Technicians of Medical Laboratories, "Sapienza" University of Rome, Italy. Supervisor of more than 30 ungraduate students.
1998 – 2012 Board member of PhD program in Neurobiology, University of Catania, Italy. Supervisor of five PhD students.

MEMBERSHIP/BOARDS

2019: Novartis advisor and consultat for Huntington's disease
2019: UCB advisor and consultat for Huntington's disease
2018/2019: ROCHE advisor and consultat for Huntington's disease
2018: WAVE advisor and consultat for Huntington's disease
2015: TEVA Pharma's advisor and consultat for Huntington's disease
Euro-HD network Principal Investigator (PI) of the Juvenile onset Huntington's disease Working Group (2004-present).
1998-2015: Central Coordination for Rare Diseases - Molise Region, Italy.
2008: Huntington Study Group (2008-present**).** Italian delegate and PI. Juvenile onset Huntington's disease Working Group from 2018.

GRANTS, PROJECTS AND AWARDS:

2019: PI of the observational trial HDClarity (sponsored by CHDI) on behalf of LIRH Foundation.
2019: PI of the Italian coordination site for Generation-HD1 double blind, multicenter, global drug trial in HD (sponsored by IONIS-Roche).
2018: JPND call for proposals**: "Multinational research projects on Health and Social Care for Neurodegenerative Diseases"**: European eHealth care model for rare neurodegenerative diseases
2018: Ministry of Health: Ordinary/Progetti ordinari di Ricerca Finalizzata 2016 - Theory-enhancing: **RARE** in rarity: Advanced in vivo and in vitro technologies to **ST**udy **J**uvenile **H**untington **D**isease neuronal connectivity and its relationship with clinical and genetic factors. The

RAREST-JHD project.
2015-present: PI of Legato-HD, double blind, multicenter, global drug trial of *Laquinimod* in HD (sponsored by TEVA).

2015-present: PI of Open PRIDE-HD, global drug trial of *pridopidine* in HD (sponsored by TEVA).
2013-2015: Scientific coordinator of the IIF Marie Curie on Huntington disease.
2014: PI and Italian Coordinator of PRIDE-HD, double blind, multicenter, global drug trial of *pridopidine* in HD (sponsored by TEVA).
2014-present: PI of the observational clinical trial ENROLL-HD (sponsored by CHDI) on behalf of LIRH Foundation.
2013-2014: PI and Research coordinator of project. Grant of one year from Telethon: TGFbeta1in Huntington disease.
2011-2012: PI**,** Italian Coordinator for the double blind multicenter, European drug trial of sirtuin inhibitor *Selisistat* in HD (sponsored by Siena Biotech).
2009-2011: PI**,** Italian Coordinator for the double blind, multicenter, global drug trial of *DIM20* (Horizon) in HD (sponsored by Medivation Trial).
2008-2010: Grant. PI of two years project sponsored by the Italian Society of Hospital Neurologists (SNO) to study Huntington's disease.
2008: PI and Italian Coordinator, global, multicenter, double-blind drug trial MarmeiHD with *ACR-16* in Huntington's disease (sponsored by NeuroSearch)
2006-2007: PI and member of the Steering Committee for the double blind multicentre-European drug trial of *Ethyl-Epa* in HD (sponsored by Amarin).
2006-2008: Project PI. Grant from Italian Health Ministry and Siena Biotech: Molecular bases of Huntington's disease: new approaches to validate new therapeutic targets.
2005-2006: Project PI. Grant of one year from Telethon: Structural Brain Changes in Huntington's disease.
2004-2014: PI of the observational clinical trial REGISTRY (sponsored by Euro-HD Network) on behalf of LIRH Foundation
2004: Award from the Italian No Profit Association "Ricerca in Movimento", for best Italian publication on Huntington disease in 2003 (Squitieri et al., Brain).
2003-2005: Project PI. Grant of two years from Italian Health Ministry (progetto finalizzato): Cell dysfunction and death in Huntington's disease by human cell models and neuroimaging
2001-2004: PI and Italian Coordinator of double-blind multicentre-European trial with *Riluzole* in Huntington disease (sponsored by Aventis Pharma).
2002-2003: Project PI. Grant of one year from Telethon: A2A receptor binding in Huntington disease
2001-2003: Project PI. Grant of two years from Italian Health Ministry (progetto finalizzato): Function and dysfunction of huntingtin
2000: Project PI. Grant of three years from Italian Research Ministry, Cluster02: Research on genetics of movement disorders.
1997: Award from the Italian No Profit Association "Ricerca in Movimento" for best Italian publication on Huntington disease in 1997 (Giovannone et al., Hum Mut)
1997: Fellowship from IRCCS Neuromed
1996: Fellowship from IRCCS Neuromed

1996: Fellowship for post-doctoral researchers, Dept. of Neurological Sciences, University "Federico II" of Naples, Italy.
1990: Fellowship for PhD students, University of Siena, Italy.
1991: Fellowship from Italian National Research Council (CNR)/British Council
1992: Fellowship from CNR/British Coun**cil**
1993: Fellowship from CNR/Canadian Medical Research Council
1994: Fellowship from CNR/Canadian Medical Research Council
1995: Consultancy from CNR (Institute of Experimental Medicine, Rome.
1988-89: Award "Roberto Mannajuolo" from Italian Huntington Disease Association (AICH).
1989-90: Award "Pino Candalino" from University Federico II of Naples, Italy.

BOOKS AND EDITORIAL BOARDS

2012: Editorial board in the Journal of Huntington Disease.
2009(Feb): Editor - Juvenile Huntington's Disease and other trinucleotide repeat disorders Oxford University Press book. Edited by Oliver W.J. Quarrell, Helen M. Brewer, **Ferdinando Squitieri**, Roger A. Barker, Martha A. Nance and G. Bernhard Landwehrmeyer ISBN13: 9780199236121ISBN10: 0199236127 Hardcover, 200 pages
1997: SPECT in Neurology and Psychiatry, edited by PP De Deyn, RA Derckx, A Alavi, BA Pickut. 1997, John Libbey & Company Ltd. Chapter 5, pp. 33-38. Huntington's chorea: IBZM SPECT role in preclinical diagnosis. D. Giobbe, F. **Squitieri**, G.C. Castellano.

Reviewer for many indexed top ranked journals including: Lancet Neurology, Lancet Psychiatry, Neurology, Brain, JNNP, Mov Disord, Eur J Neurol, Am J Hum Genet, Hum Mol Genet, Clin Genet, Europ J Hum Genet, PloS One, etc…

PUBBLICATIONS

1. A Novel Triplet-Primed PCR Assay to Detect the Full Range of Trinucleotide CAG Repeats in the Huntingtin Gene (*HTT*). De Luca A, Morella A, Consoli F, Fanelli S, Thibert JR, Statt S, Latham GJ, **Squitieri F***. Int J Mol Sci. 2021 Feb 8;22(4):1689. doi: 10.3390/ijms22041689. PMID: 33567536 Free article.

2. Known Drugs Identified by Structure-Based Virtual Screening Are Able to Bind Sigma-1 Receptor and Increase Growth of Huntington Disease Patient-Derived Cells. Battista T, Pascarella G, Staid DS, Colotti G, Rosati J, Fiorillo A, Casamassa A, Vescovi AL, Giabbai B, Semrau MS, Fanelli S, Storici P, **Squitieri F**, Morea V, Ilari A. Int J Mol Sci. 2021 Jan 28;22(3):1293. doi: 10.3390/ijms22031293. PMID: 33525510 Free PMC article.

3. Abnormal visual scanning and impaired mental state recognition in pre-manifest Huntington disease. Olivetti Belardinelli M, Hünefeldt T, Meloni R, **Squitieri F**, Maffi S, Migliore S. Exp Brain Res. 2021 Jan;239(1):141-150. doi: 10.1007/s00221-020-05957-x. Epub 2020 Nov 1. PMID: 33130907.

4. Sorcin is an early marker of neurodegeneration, Ca^{2+} dysregulation and endoplasmic reticulum stress associated to neurodegenerative diseases. Genovese I, Giamogante F, Barazzuol L, Battista T, Fiorillo A, Vicario M, D'Alessandro G, Cipriani R, Limatola C, Rossi D, Sorrentino V, Poser E, Mosca L, **Squitieri F**, Perluigi M, Arena A, van Petegem F,

Tito C, Fazi F, Giorgi C, Calì T, Ilari A, Colotti G. Cell Death Dis. 2020 Oct 15;11(10):861. doi: 10.1038/s41419-020-03063-y. PMID: 33060591 Free PMC article.

5. Mitochondrial Respiration Changes in R6/2 Huntington's Disease Model Mice during Aging in a Brain Region Specific Manner. Burtscher J, Di Pardo A, Maglione V, Schwarzer C, **Squitieri F**. Int J Mol Sci. 2020 Jul 30;21(15):5412. doi: 10.3390/ijms21155412. PMID: 32751413

6. Frequency of the loss of CAA interruption in the HTT CAG tract and implications for Huntington disease in the reduced penetrance range. Findlay Black H, Wright GEB, Collins JA, Caron N, Kay C, Xia Q, Arning L, Bijlsma EK, **Squitieri F**, Nguyen HP, Hayden MR. Genet Med. 2020 Aug 3. doi: 10.1038/s41436-020-0917-z. Online ahead of print.

7. Incidence and prevalence of Huntington disease (HD) in the Sultanate of Oman: the first Middle East post-*HTT* service-based study. **Squitieri F***, Maffi S, Al Harasi S, Al Salmi Q, D'Alessio B, Capelli G, Mazza T. J Neurol Neurosurg Psychiatry. 2020 Jul 31:jnnp-2020-323241. doi: 10.1136/jnnp-2020-323241. Online ahead of print. PMID: 32737262 No abstract available.

8. Tracing the mutated HTT and haplotype of the African ancestor who spread Huntington disease into the Middle East. **Squitieri F***, Mazza T, Maffi S, De Luca A, AlSalmi Q, AlHarasi S, Collins JA, Kay C, Baine-Savanhu F, Landwhermeyer BG, Sabatini U, Hayden MR. Genet Med. 2020 Jul 14. doi: 10.1038/s41436-020-0895-1. Online ahead of print.

9. Utility of the Parkinson's disease-Cognitive Rating Scale for the screening of global cognitive status in Huntington's disease. Martinez-Horta S, Horta-Barba A, Perez-Perez J, Sampedro F, de Lucia N, De Michele G, Kehrer S, Priller J, Migliore S, **Squitieri F**, Castaldo A, Mariotti C, Mañanes V, Lopez-Sendon JL, Rodriguez N, Martinez-Descals A, Garcia-Ruiz P, Júlio F, Januário C, Delussi M, de Tommaso M, Noguera S, Ruiz-Idiago J, Sitek EJ, Nuzzi A, Pagonabarraga J, Kulisevsky J; Cognitive Phenotype Working Group of the European Huntington's Disease Network. J Neurol. 2020 May;267(5):1527-1535. doi: 10.1007/s00415-020-09730-6. Epub 2020 Feb 7.

10. Emotion recognition and inhibitory control in manifest and pre-manifest Huntington's disease: evidence from a new Stroop task. Hunefeldt T, Maffi S, Migliore S, **Squitieri F**, Belardinelli MO. Neural Regen Res. 2020 Aug;15(8):1518-1525. doi: 10.4103/1673-5374.274342.

11. Kay C, Collins JA, Caron NS, Agostinho LA, Findlay-Black H, Casal L, Sumathipala D, Dissanayake VHW, Cornejo-Olivas M, Baine F, Krause A, Greenberg JL, Paiva CLA, **Squitieri F**, Hayden MR. A Comprehensive Haplotype Targeting Strategy for Allele-Specific HTT Suppression in Huntington Disease. Am J Hum Genet. 2019 Nov 4. pii: S0002-9297(19)30399-4. doi: 10.1016/j.ajhg.2019.10.011. [Epub ahead of print] PubMed PMID: 31708117.

12. Bidollari E, Rotundo G, Altieri F, Amicucci M, Wiquel D, Ferrari D, Goldoni M, Bernardini L, Consoli F, De Luca A, Fanelli S, Lamorte G, D'Agruma L, Vescovi AL, **Squitieri F**, Rosati J. Generation of induced pluripotent stem cell line CSSi008-A(4698) from a patient affected by advanced stage of Dentato-Rubral-Pallidoluysian atrophy (DRPLA). Stem Cell Res. 2019 Aug 27;40:101551.

13. Bachoud-Lévi AC, Ferreira J, Massart R, Youssov K, Rosser A, Busse M, Craufurd D, Reilmann R, De Michele G, Rae D, **Squitieri F**, Seppi K, Perrine C, Scherer-Gagou C, Audrey O, Verny C, Burgunder JM. International Guidelines for the Treatment of Huntington's Disease. Front Neurol. 2019 Jul 3;10:710. doi: 10.3389/fneur.2019.00710. eCollection 2019. PubMed PMID: 31333565; PubMed Central PMCID: PMC6618900.

14. Migliore S, Jankovic J, **Squitieri F***. Genetic Counseling in Huntington's Disease: Potential New Challenges on Horizon? Front Neurol. 2019 Apr 30;10:453. doi: 10.3389/fneur.2019.00453. eCollection 2019. PubMed PMID: 31114543; PubMed Central PMCID: PMC6503085.

15. Wright GEB, Collins JA, Kay C, McDonald C, Dolzhenko E, Xia Q, Bečanović K,Drögemöller BI, Semaka A, Nguyen CM, Trost B, Richards F, Bijlsma EK, **Squitieri F**, Ross CJD, Scherer SW, Eberle MA, Yuen RKC, Hayden MR. Length of Uninterrupted CAG, Independent of Polyglutamine Size, Results in Increased Somatic Instability,Hastening Onset of Huntington Disease. Am J Hum Genet. 2019 Jun 6;104(6):1116-1126. doi: 10.1016/j.ajhg.2019.04.007. Epub 2019 May 16. PubMed PMID: 31104771; PubMed Central PMCID: PMC6556907.

16. D'Aurizio G, Migliore S, Curcio G, **Squitieri F**. Safer Attitude to Risky Decision-Making in Premanifest Huntington's Disease Subjects. Front Psychol. 2019 Apr 16;10:846. doi: 10.3389/fpsyg.2019.00846. eCollection 2019. PubMed PMID:31057466; PubMed Central PMCID: PMC6477566.

17. Horton MC, Nopoulos P, Nance M, Landwehrmyer GB, Barker RA, **Squitieri F**; REGISTRY Investigators of the European Huntington's Disease Network, Burgunder JM, Quarrell O. Assessment of the Performance of a Modified Motor Scale as Applied to Juvenile Onset Huntington's Disease. J Huntingtons Dis. 2019;8(2):181-193. doi: 10.3233/JHD-180306. PubMed PMID: 30856116.

18. Quarrell OWJ, Nance MA, Nopoulos P, Reilmann R, Oosterloo M, Tabrizi SJ, Furby H, Saft C, Roos RAC, **Squitieri F**, Landwehrmeyer GB, Burgunder JM; Juvenile Huntington Disease Working Group of the European Huntington Disease Network.Defining pediatric huntington disease: Time to abandon the term Juvenile Huntington Disease? Mov Disord. 2019 Apr;34(4):584-585. doi: 10.1002/mds.27640. Epub 2019 Feb 20. PubMed PMID: 30788860.

19. Marano M, Migliore S, **Squitieri F**, Insola A, Scarnati E, Mazzone P. CM-Pf deep brain stimulation and the long term management of motor and psychiatric symptoms in a case of Tourette syndrome. J Clin Neurosci. 2019 Jan 3. pii: S0967-5868(18)32058-7. doi: 10.1016/j.jocn.2018.12.029. [Epub ahead of print]

20. Reilmann R, McGarry A, Grachev ID, Savola JM, Borowsky B, Eyal E, Gross N, Langbehn D, Schubert R, Wickenberg AT, Papapetropoulos S, Hayden M, **Squitieri F**, Kieburtz K, Landwehrmeyer GB; European Huntington's Disease Network; Huntington Study Group investigators. Safety and efficacy of pridopidine in patients with Huntington's disease (PRIDE-HD): a phase 2, randomised, placebo-controlled, multicentre, dose-ranging study. Lancet Neurol. 2018 Dec 14. pii: S1474-4422(18)30391-0. doi: 10.1016/S1474-4422(18)30391-0. [Epub ahead of print]

21. Castaldo I, De Rosa M, Romano A, Zuchegna C, **Squitieri F**, Mechelli R, Peluso S, Borrelli C, Del Mondo A, Salvatore E, Vescovi LA, Migliore S, De Michele G, Ristori G, Romano S, Avvedimento EV, Porcellini A. DNA damage signatures in peripheral blood cells (PBMC) as biomarkers in prodromal Huntington's disease. Ann Neurol. 2018 Dec 14. doi: 10.1002/ana.25393. [Epub ahead of print]

22. Oosterloo M, Bijlsma EK, van Kuijk SM, Minkels F, de Die-Smulders CE; within REGISTRY Investigators of the European Huntington's Disease Network; Registry Steering committee; Language coordinators; EHDN's associate site in Singapore. Clinical and genetic characteristics of late-onset Huntington's disease. Parkinsonism Relat Disord. 2018 Nov 29. pii: S1353-8020(18)30490-5.

23. Belardinelli MO, Huenefeldt T, Maffi S, **Squitieri F***, Migliore S. Effects of Stimulus-Related Variables on Mental States Recognition in Huntington'S Disease. Int J Neurosci. 2018 Nov 27:1-21. doi: 10.1080/00207454.2018.1552691.

24. Migliore S, Curcio G, Porcaro C, Cottone C, Simonelli I, D'aurizio G, Landi D, Palmieri MG, Ghazaryan A, **Squitieri F**, Filippi MM, Vernieri F. Emotional processing in RRMS patients: Dissociation between behavioural and neurophysiological response. Mult Scler Relat Disord. 2018 Nov 19;27:344-349. doi: 10.1016/j.msard.2018.11.019. [Epub ahead of print].

25. Fusilli C, Migliore S, Mazza T, Consoli F, De Luca A, Barbagallo G, Ciammola A, Gatto EM, Cesarini M, Etcheverry JL, Parisi V, Al-Oraimi M, Al-Harrasi S, Al-Salmi Q, Marano M, Vonsattel JG, Sabatini U, Landwehrmeyer GB, **Squitieri F***. Biological and clinical manifestations of juvenile Huntington's disease: a retrospective analysis. Lancet Neurol. 2018 Nov;17(11):986-993.

26. Migliore S, D'Aurizio G, Curcio G, **Squitieri F**. Task-switching abilities in pre-manifest Huntington's disease subjects. Parkinsonism Relat Disord. 2018 Sep pii: S1353-8020(18)30392-4.

27. McNulty P, Pilcher R, Ramesh R, Necuiniate R, Hughes A, Farewell D, Holmans P,Jones L; (within REGISTRY Investigators of the European Huntington's Disease Network). Reduced Cancer Incidence in Huntington's Disease: Analysis in the Registry Study. J Huntingtons Dis. 2018;7(3):209-222.

28. Gatto E, Parisi V, Persi G, Rey EF, Cesarini M, Etcheverry JL, Rivera P, **Squitieri F**. Optical coherence tomography (OCT) study in Argentinean Huntington's disease patients. Int J Neurosci. 2018 Jun 18:1-18.

29. Rotundo G, Bidollari E, Ferrari D, Spasari I, Bernardini L, Consoli F, De Luca A, Santimone I, Lamorte G, Migliore S, **Squitieri F**, Vescovi AL, Rosati J.Generation of the induced pluripotent stem cell line CSSi006-A (3681) from a patient affected by advanced-stage Juvenile Onset Huntington's Disease. Stem Cell Res. 2018 May;29:174-178.

30. Zarotti N, Simpson J, Fletcher I, **Squitieri F**, Migliore S. Exploring emotion regulation and emotion recognition in people with presymptomatic Huntington's disease: The role of emotional awareness. Neuropsychologia. 2018 Apr;112:1-9.

31. Bidollari E, Rotundo G, Ferrari D, Candido O, Bernardini L, Consoli F, De Luca A, Santimone I, Lamorte G, Ilari A, **Squitieri F**, Vescovi AL, Rosati J. Generation of induced pluripotent stem cell line, CSSi004-A (2962), from a patient diagnosed with Huntington's disease at the presymptomatic stage. Stem Cell Res. 2018 Apr;28:145-148.

32. Kay C, Collins JA, Wright GEB, Baine F, Miedzybrodzka Z, Aminkeng F, Semaka AJ, McDonald C, Davidson M, Madore SJ, Gordon ES, Gerry NP, Cornejo-Olivas M, **Squitieri F**, Tishkoff S, Greenberg JL, Krause A, Hayden MR. The molecular epidemiology of Huntington disease is related to intermediate allele frequence and haplotype in the general population. Am J Med Genet B Neuropsychiatr Genet. 2018 Apr;177(3):346-357.

33. Rosati J, Bidollari E, Rotundo G, Ferrari D, Torres B, Bernardini L, Consoli F, De Luca A, Santimone I, Lamorte G, **Squitieri F**, Vescovi AL. Generation of induced pluripotent stem cell line, CSSi002-A (2851), from a patient with juvenile Huntington Disease. Stem Cell Res. 2018 Jan 9;27:86-89.

34. Zielonka D, Ren M, De Michele G, Roos RAC, **Squitieri F**, Bentivoglio AR, Marcinkowski JT, Landwehrmeyer GB. The contribution of gender differences in motor, behavioral and cognitive features to functional capacity, independence and quality of life in patients with Huntington's disease. Parkinsonism Relat Disord. 2018 Jan 5. pii: S1353-8020(18)30006-3.

35. Cariulo C, Azzollini L, Verani M, Martufi P, Boggio R, Chiki A, Deguire SM, Cherubini M, Gines S, Marsh JL, Conforti P, Cattaneo E, Santimone I, **Squitieri F**, Lashuel HA, Petricca L, Caricasole A. Phosphorylation of huntingtin at residue T3 is decreased in Huntington's disease and modulates mutant huntingtin protein conformation. Proc Natl Acad Sci U S A. 2017 Dec 12;114.

36. Migliore S, Ghazaryan A, Simonelli I, Pasqualetti P, **Squitieri F**, Curcio G, Landi D, Palmieri MG, Moffa F, Filippi MM, Vernieri F. Cognitive Impairment in Relapsing-Remitting Multiple Sclerosis Patients with Very Mild Clinical Disability. Behav Neurol.

37. Ashkenazi A, Bento CF, Ricketts T, Vicinanza M, Siddiqi F, Pavel M, **Squitieri F**, Hardenberg MC, Imarisio S, Menzies FM, Rubinsztein DC. Polyglutamine tracts regulate autophagy. Autophagy. 2017 Jul 19:1-2.

38. Daldin M, Fodale V, Cariulo C, Azzollini L, Verani M, Martufi P, Spiezia MC, Deguire SM, Cherubini M, Macdonald D, Weiss A, Bresciani A, Vonsattel JG, Petricca L, Marsh JL, Gines S, Santimone I, Marano M, Lashuel HA, **Squitieri F**, Caricasole A. Polyglutamine expansion affects huntingtin conformation in multiple Huntington's disease models. Sci Rep. 2017 Jul 11;7(1):5070.

39. Migliore S, Curcio G, Couyoumdjian A, Ghazaryan A, Landi D, Moffa F, Quintiliani L, **Squitieri F**, Palmieri MG, Filippi MM, Vernieri F. Executive functioning in relapsing-remitting multiple sclerosis patients without cognitive impairment: A task-switching protocol. Mult Scler. 2017 Jul 1:1352458517719149.

40. Moss DJH, Pardiñas AF, Langbehn D, Lo K, Leavitt BR, Roos R, Durr A, Mead S; TRACK-HD investigators; **within** REGISTRY investigators, Holmans P, Jones L, Tabrizi SJ. Identification of genetic variants associated with Huntington's disease progression: a genome-wide association study. Lancet Neurol. 2017 Sep;16(9):701-711.

41. Sánchez-Castañeda C, de Pasquale F, Caravasso CF, Marano M, Maffi S, Migliore S, Sabatini U, **Squitieri F***. Resting-state connectivity and modulated somatomotor and default-mode networks in Huntington disease. CNS Neurosci Ther. 2017 Jun;23(6):488-497.

42. Ashkenazi A, Bento CF, Ricketts T, Vicinanza M, Siddiqi F, Pavel M, **Squitieri F**, Hardenberg MC, Imarisio S, Menzies FM, Rubinsztein DC. Polyglutamine tracts regulate beclin 1-dependent autophagy. Nature. 2017 May 4;545(7652): 108-111.doi: 10.1038/nature22078. [Epub ahead of print] PubMed PMID: 28445460.

43. Morea V, Bidollari E, Colotti G, Fiorillo A, Rosati J, De Filippis L, **Squitieri F***, Ilari A. Glucose transportation in the brain and its impairment in Huntington disease: one more shade of the energetic metabolism failure? Amino Acids. 2017 Apr 10. doi: 10.1007/s00726-017-2417-2. [Epub ahead of print] Review.PubMed PMID: 28396959.

44. Jankovic J, **Squitieri F**. Letter re: Huntington disease reduced penetrance alleles occur at high frequency in the general population. Neurology. 2017 Jan 17;88(3):334. doi: 10.1212/WNL.0000000000003527. PubMed PMID: 28093514.

45. Sciacca S, Favellato M, Madonna M, Metro D, Marano M, **Squitieri F***. Early enteric neuron dysfunction in mouse and human Huntington disease. Parkinsonism Relat Disord. 2017 Jan;34:73-74. doi: 10.1016/j.parkreldis.2016.10.017. Epub 2016 Nov 1. PubMed PMID: 27836713.

46. Cubo E, Ramos-Arroyo MA, Martinez-Horta S, Martínez-Descalls A, Calvo S,Gil-Polo C; European HD Network.. Clinical manifestations of intermediate allele carriers in Huntington disease. Neurology. 2016 Aug 9;87(6):571-8. doi:10.1212/WNL.0000000000002944. Epub 2016 Jul 8. PubMed PMID: 27402890.

47. Phillips OR, Joshi SH, **Squitieri F**, Sanchez-Castaneda C, Narr K, Shattuck DW, Caltagirone C, Sabatini U, Di Paola M. Major Superficial White Matter Abnormalities in Huntington's Disease. Front Neurosci. 2016 May 23;10:197. doi: 10.3389/fnins.2016.00197. eCollection 2016. PubMed PMID: 27242403; PubMed Central PMCID: PMC4876130.

48. Luca G, Bellezza I, Arato I, Di Pardo A, Mancuso F, Calvitti M, Falabella G, Bartoli S, Maglione V, Amico E, Favellato M, Basta G, Bodo M, Minelli A, Calafiore R, Frati L, **Squitieri F***. Terapeutic Potential of Microencapsulated Sertoli Cells in Huntington Disease. CNS Neurosci Ther. 2016 Aug;22(8):686-90. doi: 10.1111/cns.12569. Epub 2016 May 26. PubMed PMID: 27225886.

49. **Squitieri F***, de Yebenes JG. Profile of pridopidine and its potential in the treatment of Huntington disease: the evidence to date. Drug Des Devel Ther. 2015 Oct 28;9:5827-33. doi: 10.2147/DDDT.S65738. eCollection 2015. PubMed PMID: 26604684; PubMed Central PMCID: PMC4629959.

50. Gatto EM, Parisi V, Etcheverry JL, Sanguinetti A, Cordi L, Binelli A, Persi G, **Squitieri F**. Juvenile Huntington disease in Argentina. Arq Neuropsiquiatr. 2016 Jan;74(1):50-4. doi: 10.1590/0004-282X20150192. Epub 2015 Nov 24. PubMed PMID: 26602194.

51. Tedroff J, Waters S, Barker RA, Roos R, **Squitieri F**; EHDN Registry Study Group. Antidopaminergic Medication is Associated with More Rapidly Progressive Huntington's Disease. J Huntingtons Dis. 2015;4(2):131-40. doi: 10.3233/JHD-150143. PubMed PMID: 26397894.

52. Kay C, Collins JA, Skotte NH, Southwell AL, Warby SC, Caron NS, Doty CN, Nguyen B, Griguoli A, Ross CJ, **Squitieri** F, Hayden MR. Huntingtin Haplotypes Provide Prioritized Target Panels for Allele-specific Silencing in Huntington Disease Patients of European Ancestry. Mol Ther. 2015 Jul 23. doi: 10.1038/mt.2015.128. [Epub ahead of print] PubMed PMID: 26201449.

53. Ferreira JJ, Rosser A, Craufurd D, **Squitieri** F, Mallard N, Landwehrmeyer B. Ethyl-eicosapentaenoic acid treatment in Huntington's disease: A placebo-controlled clinical trial. Mov Disord. 2015 Jul 14. doi: 10.1002/mds.26308. [Epub ahead of print] PubMed PMID: 26175332.

54. **Squitieri** F*, Di Pardo A, Favellato M, Amico E, Maglione V, Frati L. Pridopidine, a dopamine stabilizer, improves motor performance and shows neuroprotective effects in Huntington disease R6/2 mouse model. J Cell Mol Med. 2015 Jun 22. doi: 10.1111/jcmm.12604. [Epub ahead of print] PubMed PMID: 26094900.

55. **Squitieri F***, Griguoli A, Capelli G, Porcellini A, D'Alessio B. Epidemiology of Huntington disease: first post-HTT gene analysis of prevalence in Italy. Clin Genet. 2016 Mar;89(3):367-70. doi: 10.1111/cge.12574. Epub 2015 Mar 15. PubMed PMID: 25689972.

56. Sánchez-Castañeda C, **Squitieri** F, Di Paola M, Dayan M, Petrollini M, Sabatini U. The role of iron in gray matter degeneration in Huntington's disease: a magnetic resonance imaging study. Hum Brain Mapp. 2015 Jan;36(1):50-66. doi: 10.1002/hbm.22612. Epub 2014 Aug 21. PubMed PMID: 25145324.

57. Di Paola M, Phillips OR, Sanchez-Castaneda C, Di Pardo A, Maglione V, Caltagirone C, Sabatini U, **Squitieri F**. MRI measures of corpus callosum iron and myelin in early Huntington's disease. Hum Brain Mapp. 2014 Jul;35(7):3143-51. doi: 10.1002/hbm.22391. Epub 2013 Oct 15. PubMed PMID: 24895252.

58. Phillips O, **Squitieri** F*, Sanchez-Castaneda C, Elifani F, Caltagirone C, Sabatini U, Di Paola M. Deep white matter in Huntington's disease. PLoS One. 2014 Oct 23;9(10):e109676. doi: 10.1371/journal.pone.0109676. eCollection 2014. PubMed PMID: 25340651; PubMed Central PMCID: PMC4207674.

59. Phillips O, **Squitieri F**, Sanchez-Castaneda C, Elifani F, Griguoli A, Maglione V, Caltagirone C, Sabatini U, Di Paola M. The Corticospinal Tract in Huntington's Disease. Cereb Cortex. 2015 Sep;25(9):2670-82. doi: 1093/cercor/bhu065. PubMed PMID: 24706734.

60. Alibardi A, **Squitieri F**, Fattapposta F, Missori P, Pierelli F, Trompetto C, Currà A. Psychiatric onset and late chorea in a patient with 41 CAG repeats in the TATA-binding protein gene. Parkinsonism Relat Disord. 2014 Jun;20(6):678-9. doi: 10.1016/j.parkreldis.2014.03.007. Epub 2014 Mar 21. PubMed PMID: 24698056.

61. Carrizzo A, Di Pardo A, Maglione V, Damato A, Amico E, Formisano L, Vecchione C, **Squitieri F*.** Nitric oxide dysregulation in platelets from patients with advanced Huntington disease. PLoS One. 2014 Feb 25;9(2):e89745. doi: 10.1371/journal.pone.0089745. eCollection 2014. PubMed PMID: 24587005; PubMed Central PMCID: PMC3934931.

62. Di Pardo A, Alberti S, Maglione V, Amico E, Cortes EP, Elifani F, Battaglia G, Busceti CL, Nicoletti F, Vonsattel JP, **Squitieri F***. Changes of peripheral TGF-β1 depend on monocytes-derived macrophages in Huntington disease. Mol Brain. 2013 Dec 13;6:55. doi: 10.1186/1756-6606-6-55. PubMed PMID: 24330808; PubMed Central PMCID: PMC4029620.

63. Quarrell OW, Nance MA, Nopoulos P, Paulsen JS, Smith JA, **Squitieri F**. Managing juvenile Huntington's disease. Neurodegener Dis Manag. 2013 Jun 1;3(3). doi: 10.2217/nmt.13.18. PubMed PMID: 24416077; PubMed Central PMCID: PMC3883192.

64. Di Pardo A, Amico E, Favellato M, Castrataro R, Fucile S, **Squitieri F***, Maglione V. FTY720 (fingolimod) is a neuroprotective and disease-modifying agent in cellular and mouse models of Huntington disease. Hum Mol Genet. 2014 May 1;23(9):2251-65. doi: 10.1093/hmg/ddt615. Epub 2013 Dec 2. PubMed PMID: 24301680.

65. Phillips O, Sanchez-Castaneda C, Elifani F, Maglione V, Di Pardo A, Caltagirone C, **Squitieri F**, Sabatini U, Di Paola M. Tractography of the corpus callosum in Huntington's disease. PLoS One. 2013 Sep 3;8(9):e73280. doi: 10.1371/journal.pone.0073280. eCollection 2013. PubMed PMID: 24019913; PubMed Central PMCID: PMC3760905.

66. Metzger S, Walter C, Riess O, Roos RA, Nielsen JE, Craufurd D; within REGISTRY Investigators of the European Huntington's Disease Network, Nguyen HP. The V471A polymorphism in autophagy-related gene ATG7 modifies age at onset specifically in Italian Huntington disease patients. PLoS One. 2013 Jul 22;8(7):e68951. doi: 10.1371/journal.pone.0068951. Print 2013. PubMed PMID: 23894380; PubMed Central PMCID: PMC3718802.

67. Hubers AA, van Duijn E, Roos RA, Craufurd D, Rickards H, Bernhard Landwehrmeyer G, van der Mast RC, Giltay EJ; within REGISTRY investigators of the European Huntington's Disease Network. Suicidal ideation in a European Huntington's disease population. J Affect Disord. 2013 Oct;151(1):248-58. doi: 10.1016/j.jad.2013.06.001. Epub 2013 Jul 20. PubMed PMID: 23876196.

68. **Squitieri F**. Neurodegenerative disease: 'Fifty shades of grey' in the Huntington disease gene. Nat Rev Neurol. 2013 Aug;9(8):421-2. doi: 10.1038/nrneurol.2013.128. Epub 2013 Jun 25. PubMed PMID: 23797246.

69. Ramos EM, Latourelle JC, Gillis T, Mysore JS, **Squitieri** F, Di Pardo A, Di Donato S, Gellera C, Hayden MR, Morrison PJ, Nance M, Ross CA, Margolis RL, Gomez-Tortosa E, Ayuso C, Suchowersky O, Trent RJ, McCusker E, Novelletto A, Frontali M, Jones R, Ashizawa T, Frank S, Saint-Hilaire MH, Hersch SM, Rosas HD, Lucente D, Harrison MB, Zanko A, Abramson RK, Marder K, Gusella JF, Lee JM, Alonso I, Sequeiros J, Myers RH, Macdonald ME. Candidate glutamatergic and dopaminergic pathway gene variants do not influence Huntington's disease

motor onset. Neurogenetics. 2013 May 4. [Epub ahead of print] PubMed PMID: 23644918.

70. **Squitieri F***, Landwehrmeyer B, Reilmann R, Rosser A, de Yebenes JG, Prang A, Ivkovic J, Bright J, Rembratt A. One-year safety and tolerability profile of pridopidine in patients with Huntington disease. Neurology. 2013 Mar 19;80(12):1086-94. doi: 10.1212/WNL.0b013e3182886965. Epub 2013 Feb 27. PubMed PMID: 23446684.

71. Eatough V, Santini H, Eiser C, Goller ML, Krysa W, de Nicola A, Paduanello M, Petrollini M, Rakowicz M, **Squitieri** F, Tibben A, Lee Weille K, Landwehrmeyer B, Quarrell O, Smith JA. The personal experience of parenting a child with Juvenile Huntington's Disease: perceptions across Europe. Eur J Hum Genet. 2013 Feb 27. doi: 10.1038/ejhg.2013.15. [Epub ahead of print] PubMed PMID: 23443023.

72. Georgiou-Karistianis N, Scahill R, Tabrizi SJ, **Squitieri** F, Aylward E. Structural MRI in Huntington's disease and recommendations for its potential use in clinical trials. Neurosci Biobehav Rev. 2013 Mar;37(3):480-90. doi: 10.1016/j.neubiorev.2013.01.022. Epub 2013 Jan 29. PubMed PMID: 23376047.

73. HORIZON Investigators of the Huntington Study Group and European Huntington's Disease Network. A randomized, double-blind, placebo-controlled study of latrepirdine in patients with mild to moderate Huntington disease. JAMA Neurol. 2013 Jan;70(1):25-33. doi: 10.1001/2013.jamaneurol.382. PubMed PMID: 23108692.

74. Zielonka D, Marinus J, Roos RA, De Michele G, Di Donato S, Putter H, Marcinkowski J, **Squitieri** F, Bentivoglio AR, Landwehrmeyer GB. The influence of gender on phenotype and disease progression in patients with Huntington's disease. Parkinsonism Relat Disord. 2013 Feb;19(2):192-7. doi: 10.1016/j.parkreldis.2012.09.012. Epub 2012 Oct 25. PubMed PMID: 23102616.

75. **Squitieri F***, Jankovic J. Huntington's disease: how intermediate are intermediate repeat lengths? Mov Disord. 2012 Dec;27(14):1714-7. doi:10.1002/mds.25172. Epub 2012 Sep 24. PubMed PMID: 23008174.

76. Ramos EM, Latourelle JC, Lee JH, Gillis T, Mysore JS, **Squitieri** F, Di Pardo A, Di Donato S, Hayden MR, Morrison PJ, Nance M, Ross CA, Margolis RL, Gomez-Tortosa E, Ayuso C, Suchowersky O, Trent RJ, McCusker E, Novelletto A, Frontali M, Jones R, Ashizawa T, Frank S, Saint-Hilaire MH, Hersch SM, Rosas HD, Lucente D, Harrison MB, Zanko A, Marder K, Gusella JF, Lee JM, Alonso I, Sequeiros J, Myers RH, Macdonald ME. Population stratification may bias analysis of PGC-1α as a modifier of age at Huntington disease motor onset. Hum Genet. 2012 Dec;131(12):1833-40. doi: 10.1007/s00439-012-1205-z. Epub 2012 Jul 25. PubMed PMID: 22825315; PubMed Central PMCID: PMC3492689.

77. Aubeeluck A, Dorey J, **Squitieri** F, Clay E, Stupple EJ, De Nicola A, Buchanan H, Martino T, Toumi M. Further evidence of reliability and validity of the Huntington's disease quality of life battery for carers: Italian and French translations. Qual Life Res. 2012 Jul 21. [Epub ahead of print] PubMed PMID: 22821667.

78. Lee JH, Lee JM, Ramos EM, Gillis T, Mysore JS, Kishikawa S, Hadzi T, Hendricks AE, Hayden MR, Morrison PJ, Nance M, Ross CA, Margolis RL, **Squitieri** F, Gellera C, Gomez-Tortosa E, Ayuso C, Suchowersky O, Trent RJ, McCusker E, Novelletto A, Frontali M, Jones R, Ashizawa T, Frank S, Saint-Hilaire MH, Hersch SM, Rosas HD, Lucente D, Harrison MB, Zanko A, Abramson RK, Marder K, Sequeiros J, Landwehrmeyer GB; Registry Study of the European Huntington's Disease Network. Shoulson I; Huntington Study Group COHORT project, Myers RH, MacDonald ME, Gusella JF. TAA repeat variation in the GRIK2 gene does not influence age at onset in Huntington's disease. Biochem Biophys Res Commun. 2012 Aug 3;424(3):404-8. Epub 2012 Jul 3. PubMed PMID: 22771793.

79. MacLeod R, Tibben A, Frontali M, Evers-Kiebooms G, Jones A, Martinez-Descales A, Roos RA; Editorial Committee and Working Group 'Genetic Testing Counselling' of the European Huntington Disease Network. Recommendations for the predictive genetic test in Huntington's disease. Clin Genet. 2013 Mar;83(3):221-31. doi: 10.1111/j.1399-0004.2012.01900.x. Epub 2012 Jul 30. PubMed PMID: 22642570.

80. Ciarmiello A, Giovacchini G, Orobello S, Bruselli L, Elifani F, **Squitieri** F. 18F-FDG PET uptake in the pre-Huntington disease caudate affects the time-to-onset independently of CAG expansion size. Eur J Nucl Med Mol Imaging. 2012 Jun;39(6):1030-6. doi: 10.1007/s00259-012-2114-z. Epub 2012 Apr 12. PubMed PMID: 22526956.

81. Clay E, De Nicola A, Dorey J, **Squitieri** F, Aballéa S, Martino T, Tedroff J, Zielonka D, Auquier P, Verny C, Toumi M. Validation of the first quality-of-life measurement for patients with Huntington's disease: the Huntington Quality of Life Instrument. Int Clin Psychopharmacol. 2012 Jul;27(4):208-14. doi: 10.1097/YIC.0b013e3283534fa9. PubMed PMID: 22508443.

82. Robertson L, Santini H, O'Donovan KL, **Squitieri** F, Barker RA, Rakowicz M, Landwehrmeyer GB, Quarrell O. Current Pharmacological Management in Juvenile Huntington's Disease. Version 3. PLoS Curr. 2012 Feb 15 [revised 2012 Mar 26];4:RRN1304. PubMed PMID: 22474619; PubMed Central PMCID: PMC3283037.

83. Lee JM, Gillis T, Mysore JS, Ramos EM, Myers RH, Hayden MR, Morrison PJ, Nance M, Ross CA, Margolis RL, **Squitieri** F, Griguoli A, Di Donato S, Gomez-Tortosa E, Ayuso C, Suchowersky O, Trent RJ, McCusker E, Novelletto A, Frontali M, Jones R, Ashizawa T, Frank S, Saint-Hilaire MH, Hersch SM, Rosas HD, Lucente D, Harrison MB, Zanko A, Abramson RK, Marder K, Sequeiros J, MacDonald ME, Gusella JF. Common SNP-based haplotype analysis of the 4p16.3 Huntington disease gene region. Am J Hum Genet. 2012

Mar 9;90(3):434-44. doi: 10.1016/j.ajhg.2012.01.005. Epub 2012 Mar 1. PubMed PMID: 22387017; PubMed Central PMCID: PMC3309179.

84. Sánchez-Castañeda C, Cherubini A, Elifani F, Péran P, Orobello S, Capelli G, Sabatini U, **Squitieri** F. Seeking huntington disease biomarkers by multimodal, cross-sectional basal ganglia imaging. Hum Brain Mapp. 2012 Feb 22. doi: 10.1002/hbm.22019. [Epub ahead of print] PubMed PMID: 22359398.

85. Lee JM, Ramos EM, Lee JH, Gillis T, Mysore JS, Hayden MR, Warby SC, Morrison P, Nance M, Ross CA, Margolis RL, **Squitieri** F, Orobello S, Di Donato S, Gomez-Tortosa E, Ayuso C, Suchowersky O, Trent RJ, McCusker E, Novelletto A, Frontali M, Jones R, Ashizawa T, Frank S, Saint-Hilaire MH, Hersch SM, Rosas HD, Lucente D, Harrison MB, Zanko A, Abramson RK, Marder K, Sequeiros J, Paulsen JS; PREDICT-HD study of the Huntington Study Group (HSG), Landwehrmeyer GB; REGISTRY study of the European Huntington's Disease Network, Myers RH; HD-MAPS Study Group, MacDonald ME, Gusella JF; COHORT study of the HSG. CAG repeat expansion in Huntington disease determines age at onset in a fully dominant fashion. Neurology. 2012 Mar 6;78(10):690-5. doi: 10.1212/WNL.0b013e318249f683. Epub 2012 Feb 8. PubMed PMID: 22323755; PubMed Central PMCID: PMC3306163.

86. Rickards H, De Souza J, Crooks J, van Walsem MR, van Duijn E, Landwehrmeyer B, **Squitieri** F, Simpson SA; European Huntington's Disease Network. Discriminant analysis of Beck Depression Inventory and Hamilton Rating Scale for Depression in Huntington's disease. J Neuropsychiatry Clin Neurosci. 2011 Fall;23(4):399-402. doi: 10.1176/appi.neuropsych.23.4.399. PubMed PMID: 22231310.

87. Di Paola M, Luders E, Cherubini A, Sanchez-Castaneda C, Thompson PM, Toga AW, Caltagirone C, Orobello S, Elifani F, **Squitieri** F, Sabatini U. Multimodal MRI analysis of the corpus callosum reveals white matter differences in presymptomatic and early Huntington's disease. Cereb Cortex. 2012 Dec;22(12):2858-66. doi: 10.1093/cercor/bhr360. Epub 2012 Jan 5. PubMed PMID: 22223853.

88. Cubo E, González M, del Puerto I, de Yébenes JG, Arconada OF, Gabriel y Galán JM; European Huntington's Disease Initiative Study Group. Placebo effect characteristics observed in a single, international, longitudinal study in Huntington's disease. Mov Disord. 2012 Mar;27(3):439-42. doi: 10.1002/mds.24062. 2011 Dec 9. PubMed PMID: 22162184.

89. de Yebenes JG, Landwehrmeyer B, **Squitieri** F, Reilmann R, Rosser A, Barker RA, Saft C, Magnet MK, Sword A, Rembratt A, Tedroff J; MermaiHD study investigators. Pridopidine for the treatment of motor function in patients with Huntington's disease (MermaiHD): a phase 3, randomised, double-blind, placebo-controlled trial. Lancet Neurol. 2011 Dec;10(12):1049-57. doi:10.1016/S1474-4422(11)70233-2. Epub 2011 Nov 7. PubMed PMID: 22071279.

90. Saft C, Epplen JT, Wieczorek S, Landwehrmeyer GB, Roos RA, de Yebenes JG, Dose M, Tabrizi SJ, Craufurd D; REGISTRY Investigators of the European Huntington's Disease Network, Arning L. NMDA receptor gene variations as modifiers in Huntington disease: a replication study. PLoS Curr. 2011 Oct 4;3:RRN1247. doi: 10.1371/currents.RRN1247. PubMed PMID: 21989477; PubMed Central PMCID: PMC3186947.

91. Quarrell OW, Handley O, O'Donovan K, Dumoulin C, Ramos-Arroyo M, Biunno I, Bauer P, Kline M, Landwehrmeyer GB; European Huntington's Disease Network. Discrepancies in reporting the CAG repeat lengths for Huntington's disease. Eur J Hum Genet. 2012 Jan;20(1):20-6. doi: 10.1038/ejhg.2011.136. Epub 2011 Aug 3. PubMed PMID: 21811303; PubMed Central PMCID: PMC3234505.

92. Johri A, Starkov AA, Chandra A, Hennessey T, Sharma A, Orobello S, **Squitieri** F, Yang L, Beal MF. Truncated peroxisome proliferator-activated receptor-γ coactivator 1α splice variant is severely altered in Huntington's disease. Neurodegener Dis. 2011;8(6):496-503. doi: 10.1159/000327910. Epub 2011 Jul 15. PubMed PMID: 21757867; PubMed Central PMCID: PMC3186722.

93. **Squitieri** F, Maglione V, Orobello S, Fornai F. Genotype-, aging-dependent abnormal caspase activity in Huntington disease blood cells. J Neural Transm. 2011 Nov;118(11):1599-607. doi: 10.1007/s00702-011-0646-1. Epub 2011 Apr 26. PubMed PMID: 21519949.

94. López-Sendón JL, Royuela A, Trigo P, Orth M, Lange H, Reilmann R, Keylock J, Rickards H, Piacentini S, **Squitieri** F, Landwehrmeyer B, Witjes-Ane MN, Jurgens CK, Roos RA, Abraira V, de Yébenes JG; European HD Network. What is the impact of education on Huntington's disease? Mov Disord. 2011 Jul;26(8):1489-95. doi: 10.1002/mds.23385. Epub 2011 Mar 22. PubMed PMID: 21432905.

95. Leoni V, Mariotti C, Nanetti L, Salvatore E, **Squitieri** F, Bentivoglio AR, Bandettini di Poggio M, Piacentini S, Monza D, Valenza M, Cattaneo E, Di Donato S. Whole body cholesterol metabolism is impaired in Huntington's disease. Neurosci Lett. 2011 May 2;494(3):245-9. doi: 10.1016/j.neulet.2011.03.025. Epub 2011 Mar 22. Erratum in: Neurosci Lett. 2011 Jul 8;498(2):171. Bandettini Del Poggio, M [corrected to Bandettini di Poggio, M]. PubMed PMID: 21406216.

96. **Squitieri** F, Esmaeilzadeh M, Ciarmiello A, Jankovic J. Caudate glucose hypometabolism in a subject carrying an unstable allele of intermediate CAG(33) repeat length in the Huntington's disease gene. Mov Disord. 2011 Apr;26(5):925-7. doi: 10.1002/mds.23623. Epub 2011 Mar 2. PubMed PMID: 21370274.

97. Orth M; European Huntington's Disease Network, Handley OJ, Schwenke C, Dunnett S, Wild EJ, Tabrizi SJ, Landwehrmeyer GB. Observing Huntington's disease: the European Huntington's Disease Network's REGISTRY. J Neurol Neurosurg Psychiatry. 2011

Dec;82(12):1409-12. doi: 10.1136/jnnp.2010.209668. Epub 2010 Nov 19. PubMed PMID: 21097549.

98. Giovacchini G, **Squitieri** F, Esmaeilzadeh M, Milano A, Mansi L, Ciarmiello A. PET translates neurophysiology into images: A review to stimulate a network between neuroimaging and basic research. J Cell Physiol. 2011 Apr;226(4):948-61. doi: 10.1002/jcp.22451. Review. PubMed PMID: 20945377.

99. Orth M, Handley OJ, Schwenke C, Dunnett SB, Craufurd D, Ho AK, Wild E, Tabrizi SJ, Landwehrmeyer GB; Investigators of the European Huntington's Disease Network. Observing Huntington's Disease: the European Huntington's Diseas Network's REGISTRY. Version 2. PLoS Curr. 2010 Sep 28 [revised 2011 Apr 13];2:RRN1184. PubMed PMID: 20890398; PubMed Central PMCID: PMC2947793.

100. Esmaeilzadeh M, Ciarmiello A, **Squitieri** F. Seeking brain biomarkers for preventive therapy in Huntington disease. CNS Neurosci Ther. 2011 Oct;17(5):368-86. doi: 10.1111/j.1755-5949.2010.00157.x. Epub 2010 Jun 11. Review. PubMed PMID: 20553306.

101. Rickards H, De Souza J, van Walsem M, van Duijn E, Simpson SA, **Squitieri** F, Landwehrmeyer B; European Huntington's Disease Network. Factor analysis of behavioural symptoms in Huntington's disease. J Neurol Neurosurg Psychiatry. 2011 Apr;82(4):411-2. doi: 10.1136/jnnp.2009.181149. Epub 2010 Apr 14. PubMed PMID: 20392980.

102. **Squitieri** F, Ciarmiello A. Key role of nuclear medicine in seeking biomarkers of Huntington's disease. Eur J Nucl Med Mol Imaging. 2010 Jun;37(6):1124-7. doi: 10.1007/s00259-010-1439-8. PubMed PMID: 20358195.

103. Battaglia G, Cannella M, Riozzi B, Orobello S, Maat-Schieman ML, Aronica E, Busceti CL, Ciarmiello A, Alberti S, Amico E, Sassone J, Sipione S, Bruno V, Frati L, Nicoletti F, **Squitieri** F. Early defect of transforming growth factor β1 formation in Huntington's disease. J Cell Mol Med. 2011 Mar;15(3):555-71. doi: 10.1111/j.1582-4934.2010.01011.x. PubMed PMID: 20082658.

104. **Squitieri** F, Falleni A, Cannella M, Orobello S, Fulceri F, Lenzi P, Fornai F. Abnormal morphology of peripheral cell tissues from patients with Huntington disease. J Neural Transm. 2010 Jan;117(1):77-83. doi: 10.1007/s00702-009-0328-4. Epub 2009 Oct 16. PubMed PMID: 19834779.

105. Aziz NA, Jurgens CK, Landwehrmeyer GB; EHDN Registry Study Group, van Roon-Mom WM, van Ommen GJ, Stijnen T, Roos RA. Normal and mutant HTT interact to affect clinical severity and progression in Huntington disease. Neurology. 2009 Oct 20;73(16):1280-5. doi: 10.1212/WNL.0b013e3181bd1121. Epub 2009 Sep 23. Erratum in: Neurology. 2009 Nov 10;73(19):1608. Neurology. 2011 Jan 11;76(2):202. Ciarmielo, Andrea [corrected to Ciarmiello, Andrea]. PubMed PMID: 19776381.

106. Ghidoni R, Albertini V, Squitti R, Paterlini A, Bruno A, Bernardini S, Cassetta E, Rossini PM, **Squitieri** F, Benussi L, Binetti G. Novel T719P AbetaPP mutation unbalances the relative proportion of amyloid-beta peptides. J Alzheimers Dis. 2009;18(2):295-303. doi: 10.3233/JAD-2009-1142. PubMed PMID: 19584443.

107. Ciammola A, Sassone J, Colciago C, Mencacci NE, Poletti B, Ciarmiello A, **Squitieri** F, Silani V. Aripiprazole in the treatment of Huntington's disease: a case series. Neuropsychiatr Dis Treat. 2009;5:1-4. Epub 2009 Apr 8. PubMed PMID: 19557093; PubMed Central PMCID: PMC2695210.

108. Chaturvedi RK, Adhihetty P, Shukla S, Hennessy T, Calingasan N, Yang L, Starkov A, Kiaei M, Cannella M, Sassone J, Ciammola A, **Squitieri** F, Beal MF. Impaired PGC-1alpha function in muscle in Huntington's disease. Hum Mol Genet. 2009 Aug 15;18(16):3048-65. doi: 10.1093/hmg/ddp243. Epub 2009 May 21. PubMed PMID: 19460884; PubMed Central PMCID: PMC2733807.

109. Smith BN, Bevan S, Vance C, Renwick P, Wilkinson P, Proukakis C, **Squitieri** F, Berardelli A, Warner TT, Reid E, Shaw CE. Four novel SPG3A/atlastin mutations identified in autosomal dominant hereditary spastic paraplegia kindreds with intra-familial variability in age of onset and complex phenotype. Clin Genet. 2009 May;75(5):485-9. doi: 10.1111/j.1399-0004.2009.01184.x. PubMed PMID: 19459885.

110. **Squitieri** F, Orobello S, Cannella M, Martino T, Romanelli P, Giovacchini G, Frati L, Mansi L, Ciarmiello A. Riluzole protects Huntington disease patients from brain glucose hypometabolism and grey matter volume loss and increases production of neurotrophins. Eur J Nucl Med Mol Imaging. 2009 Jul;36(7):1113-20. doi: 10.1007/s00259-009-1103-3. Epub 2009 Mar 11. PubMed PMID: 19280185.

111. **Squitieri** F, Cannella M, Simonelli M, Sassone J, Martino T, Venditti E, Ciammola A, Colonnese C, Frati L, Ciarmiello A. Distinct brain volume changes correlating with clinical stage, disease progression rate, mutation size, and age at onset prediction as early biomarkers of brain atrophy in Huntington's disease. CNS Neurosci Ther. 2009 Winter;15(1):1-11. doi: 10.1111/j.1755-5949.2008.00068.x. PubMed PMID: 19228174.

112. Cannella M, Maglione V, Martino T, Ragona G, Frati L, Li GM, **Squitieri** F. DNA instability in replicating Huntington's disease lymphoblasts. BMC Med Genet. 2009 Feb 11;10:11. doi: 10.1186/1471-2350-10-11. PubMed PMID: 19210789; PubMed Central PMCID: PMC2645380.

113. Cannella M, Martino T, Simonelli M, Ciammola A, Gradini R, Ciarmiello A, Gianfrancesco F, **Squitieri** F. De novo seven extra repeat expanded mutation in the PRNP gene in an Italian patient with early onset dementia. BMJ Case Rep. 2009;2009. doi:pii: bcr08.2008.0711. 10.1136/bcr.08.2008.0711. Epub 2009 Feb 2. PubMed PMID: 21686668; PubMed Central PMCID: PMC3029391.

114. Herishanu YO, Parvari R, Pollack Y, Shelef I, Marom B, Martino T, Cannella M, **Squitieri** F. Huntington disease in subjects from an Israeli Karaite community carrying alleles of intermediate and expanded CAG repeats in the HTT gene: Huntington disease or phenocopy? J Neurol Sci. 2009 Feb 15;277(1-2):143-6. Doi 10.1016/j.jns.2008.11.005. Epub 2008 Dec 6. PubMed PMID: 19059613.

115. Aziz NA, van der Burg JM, Landwehrmeyer GB, Brundin P, Stijnen T; EHDI Study Group, Roos RA. Weight loss in Huntington disease increases with higher CAG repeat number. Neurology. 2008 Nov 4;71(19):1506-13. doi: 10.1212/01.wnl.0000334276.09729.0e. PubMed PMID: 18981372.

116. **Squitieri** F, Cannella M, Frati L. Molecular medicine: predicting and preventing Huntington's disease. Neurol Sci. 2008 Sep;29(4):205-7. doi: 10.1007/s10072-008-0969-3. Epub 2008 Sep 20. Review. PubMed PMID: 18810593.

117. Gianfrancesco F, Esposito T, Penco S, Maglione V, Liquori CL, Patrosso MC, Zuffardi O, Ciccodicola A, Marchuk DA, **Squitieri** F. ZPLD1 gene is disrupted in a patient with balanced translocation that exhibits cerebral cavernous malformations. Neuroscience. 2008 Aug 13;155(2):345-9. doi: 10.1016/j.neuroscience.2008.05.030. Epub 2008 Jul 16. PubMed PMID: 18632209.

118. Liquori CL, Penco S, Gault J, Leedom TP, Tassi L, Esposito T, Awad IA, Frati , Johnson EW, **Squitieri** F, Marchuk DA, Gianfrancesco F. Different spectra of genomic deletions within the CCM genes between Italian and American CCM patient cohorts. Neurogenetics. 2008 Feb;9(1):25-31. Epub 2007 Dec 1. PubMed PMID: 18060436.

119. Traficante A, Riozzi B, Cannella M, Rampello L, **Squitieri** F, Battaglia G. Reduced activity of cortico-striatal fibres in the R6/2 mouse model of Huntington's disease. Neuroreport. 2007 Dec 3;18(18):1997-2000. PubMed PMID: 18007201.

120. **Squitieri** F, Ciammola A, Colonnese C, Ciarmiello A. Neuroprotective effects of riluzole in Huntington's disease. Eur J Nucl Med Mol Imaging. 2008 Jan;35(1):221-2. Epub 2007 Nov 6. PubMed PMID: 17987289.

121. Cannella M, Martino T, Simonelli M, Ciammola A, Gradini R, Ciarmiello A, Gianfrancesco F, **Squitieri** F*. De novo seven extra repeat expanded mutation in the PRNP gene in an Italian patient with early onset dementia. J Neurol Neurosurg Psychiatry. 2007 Dec;78(12):1411-3. Epub 2007 Aug 6. PubMed PMID: 17682007; PubMed Central PMCID: PMC2095619.

122. Di Fonzo A, Chien HF, Socal M, Giraudo S, Tassorelli C, Iliceto G, Fabbrini G, Marconi R, Fincati E, Abbruzzese G, Marini P, **Squitieri** F, Horstink MW, Montagna P, Libera AD, Stocchi F, Goldwurm S, Ferreira JJ, Meco G, Martignoni E, Lopiano L, Jardim LB, Oostra BA, Barbosa ER; Italian Parkinson Genetics Network, Bonifati V. ATP13A2 missense

mutations in juvenile parkinsonism and young onset Parkinson disease. Neurology. 2007 May 8;68(19):1557-62. PubMed PMID: 17485642.

123. Gianfrancesco F, Cannella M, Martino T, Maglione V, Esposito T, Innocenzi G, Vitale E, Liquori CL, Marchuk DA, **Squitieri** F*. Highly variable penetrance in subjects affected with cavernous cerebral angiomas (CCM) carrying novel CCM1 and CCM2 mutations. Am J Med Genet B Neuropsychiatr Genet. 2007 Jul 5;144B(5):691-5. PubMed PMID: 17440989.

124. Ciammola A, Sassone J, Cannella M, Calza S, Poletti B, Frati L, **Squitieri** F, Silani V. Low brain-derived neurotrophic factor (BDNF) levels in serum of Huntington's disease patients. Am J Med Genet B Neuropsychiatr Genet. 2007 Jun 5;144B(4):574-7. PubMed PMID: 17427191.

125. Liquori CL, Berg MJ, **Squitieri** F, Leedom TP, Ptacek L, Johnson EW, Marchuk DA. Deletions in CCM2 are a common cause of cerebral cavernous malformations. Am J Hum Genet. 2007 Jan;80(1):69-75. Epub 2006 Nov 14. PubMed PMID: 17160895; PubMed Central PMCID: PMC1785317.

126. 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 Aug 17;7:71. PubMed PMID: 16914060; PubMed Central PMCID: PMC1586197.

127. Mormone E, Matarrese P, Tinari A, Cannella M, Maglione V, Farrace MG, Piacentini M, Frati L, Malorni W, **Squitieri** F. Genotype-dependent priming to self- and xeno-cannibalism in heterozygous and homozygous lymphoblasts from patients with Huntington's disease. J Neurochem. 2006 Aug;98(4):1090-9. PubMed PMID: 16895579.

128. Zhong N, Kim CY, Rizzu P, Geula C, Porter DR, Pothos EN, **Squitieri** F, Heutink P, Xu J. DJ-1 transcriptionally up-regulates the human tyrosine hydroxylase by inhibiting the sumoylation of pyrimidine tract-binding protein-associated splicing factor. J Biol Chem. 2006 Jul 28;281(30):20940-8. Epub 2006 May 26. PubMed PMID: 16731528.

129. Ciammola A, Sassone J, Alberti L, Meola G, Mancinelli E, Russo MA, **Squitieri** F, Silani V. Increased apoptosis, Huntingtin inclusions and altereddifferentiation in muscle cell cultures from Huntington's disease subjects. Cell Death Differ. 2006 Dec;13(12):2068-78. Epub 2006 May 26. PubMed PMID: 16729030.

130. **Squitieri** F*, Ciarmiello A, Di Donato S, Frati L. The search for cerebral biomarkers of Huntington's disease: a review of genetic models of age at onset prediction. Eur J Neurol. 2006 Apr;13(4):408-15. Review. PubMed PMID: 16643321.

131. Ciarmiello A, Cannella M, Lastoria S, Simonelli M, Frati L, Rubinsztein DC, **Squitieri** F. Brain white-matter volume loss and glucose hypometabolism precede the clinical symptoms of Huntington's disease. J Nucl Med. 2006 Feb;47(2):215-22. PubMed PMID: 16455626.

132. Liquori CL, Berg MJ, **Squitieri** F, Ottenbacher M, Sorlie M, Leedom TP, Cannella M, Maglione V, Ptacek L, Johnson EW, Marchuk DA. Low frequency of PDCD10 mutations in a panel of CCM3 probands: potential for a fourth CCM locus. Hum Mutat. 2006 Jan;27(1):118. PubMed PMID: 16329096.

133. Maglione V, Cannella M, Gradini R, Cislaghi G, **Squitieri** F*. Huntingtin fragmentation and increased caspase 3, 8 and 9 activities in lymphoblasts with heterozygous and homozygous Huntington's disease mutation. Mech Ageing Dev. 2006 Feb;127(2):213-6. Epub 2005 Nov 10. PubMed PMID: 16289252.

134. **Squitieri** F*, Cannella M, Sgarbi G, Maglione V, Falleni A, Lenzi P, Baracca A, Cislaghi G, Saft C, Ragona G, Russo MA, Thompson LM, Solaini G, Fornai F. Severe ultrastructural mitochondrial changes in lymphoblasts homozygous for Huntington disease mutation. Mech Ageing Dev. 2006 Feb;127(2):217-20. Epub 2005 Nov 9. PubMed PMID: 16289240.

135. **Squitieri** F*, Frati L, Ciarmiello A, Lastoria S, Quarrell O. Juvenile Huntington's disease: does a dosage-effect pathogenic mechanism differ from the classical adult disease? Mech Ageing Dev. 2006 Feb;127(2):208-12. Epub 2005 Nov 7. Review. PubMed PMID: 16274727.

136. Maglione V, Cannella M, Martino T, De Blasi A, Frati L, **Squitieri** F*. The platelet maximum number of A2A-receptor binding sites (Bmax) linearly correlates with age at onset and CAG repeat expansion in Huntington's disease patients with predominant chorea. Neurosci Lett. 2006 Jan 23;393(1):27-30. Epub 2005 Oct 10. PubMed PMID: 16221531.

137. Maglione V, Giallonardo P, Cannella M, Martino T, Frati L, **Squitieri** F*. Adenosine A2A receptor dysfunction correlates with age at onset anticipation inblood platelets of subjects with Huntington's disease. Am J Med Genet B Neuropsychiatr Genet. 2005 Nov 5;139B(1):101-5. PubMed PMID: 16184606.

138. Bertoli-Avella AM, Giroud-Benitez JL, Akyol A, Barbosa E, Schaap O, van der Linde HC, Martignoni E, Lopiano L, Lamberti P, Fincati E, Antonini A, Stocchi F, Montagna P, **Squitieri** F, Marini P, Abbruzzese G, Fabbrini G, Marconi R, Dalla Libera A, Trianni G, Guidi M, De Gaetano A, Boff Maegawa G, De Leo A, Gallai V, de Rosa G, Vanacore N, Meco G, van Duijn CM, Oostra BA, Heutink P, Bonifati V; Italian Parkinson Genetics

Network. Novel parkin mutations detected in patients with early-onset Parkinson's disease. Mov Disord. 2005 Apr;20(4):424-31. PubMed PMID: 15584030.

139. Cannella M, Maglione V, Martino T, Simonelli M, Ragona G, **Squitieri** F*. New Huntington disease mutation arising from a paternal CAG34 allele showing somatic length variation in serially passaged lymphoblasts. Am J Med Genet B Neuropsychiatr Genet. 2005 Feb 5;133B(1):127-30. PubMed PMID: 15546151.

140. Mariotti C, Gellera C, Rimoldi M, Mineri R, Uziel G, Zorzi G, Pareyson D, Piccolo G, Gambi D, Piacentini S, **Squitieri** F, Capra R, Castellotti B, Di Donato S. Ataxia with isolated vitamin E deficiency: neurological phenotype, clinical follow-up and novel mutations in TTPA gene in Italian families. Neurol Sci. 2004 Jul;25(3):130-7. PubMed PMID: 15300460.

141. **Squitieri** F*, Almqvist EW, Cannella M, Cislaghi G, Hayden MR. Predictive testing for persons at risk for homozygosity for CAG expansion in the Huntington disease gene. Clin Genet. 2003 Dec;64(6):524-5. PubMed PMID: 14986835.

142. Cannella M, Gellera C, Maglione V, Giallonardo P, Cislaghi G, Muglia M, Quattrone A, Pierelli F, Di Donato S, **Squitieri** F*. The gender effect in juvenile Huntington disease patients of Italian origin. Am J Med Genet B Neuropsychiatr Genet. 2004 Feb 15;125B(1):92-8. PubMed PMID: 14755452.

143. Liquori CL, Berg MJ, Siegel AM, Huang E, Zawistowski JS, Stoffer T, Verlaan D, Balogun F, Hughes L, Leedom TP, Plummer NW, Cannella M, Maglione V, **Squitieri** F, Johnson EW, Rouleau GA, Ptacek L, Marchuk DA. Mutations in a gene encoding anovel protein containing a phosphotyrosine-binding domain cause type 2 cerebralcavernous malformations. Am J Hum Genet. 2003 Dec;73(6):1459-64. Epub 2003 Nov17. PubMed PMID: 14624391; PubMed Central PMCID: PMC1180409.

144. **Squitieri F***, Cannella M, Gaudio L, Martino T, Maglione V, Giallonardo P, Simonelli M, Simonelli G, Mangeruga D, Ciarmiello A, Pierelli F. Italian Huntington disease patients--data and tissue bank. Neurol Sci. 2003 Oct;24(3):215-6. PubMed PMID: 14598094.

145. Bonifati V, Rizzu P, **Squitieri** F, Krieger E, Vanacore N, van Swieten JC, Brice A, van Duijn CM, Oostra B, Meco G, Heutink P. DJ-1(PARK7), a novel gene for autosomal recessive, early onset parkinsonism. Neurol Sci. 2003 Oct;24(3):159-60. PubMed PMID: 14598065.

146. Varani K, Abbracchio MP, Cannella M, Cislaghi G, Giallonardo P, Mariotti C, Cattabriga E, Cattabeni F, Borea PA, **Squitieri** F, Cattaneo E. Aberrant A2A receptor function in peripheral blood cells in Huntington's disease. FASEB J 2003 Nov;17(14):2148-50. Epub 2003 Sep 4. PubMed PMID: 12958155.

147. Macedo MG, Anar B, Bronner IF, Cannella M, **Squitieri** F, Bonifati V, Hoogeveen A, Heutink P, Rizzu P. The DJ-1L166P mutant protein associated with early onset Parkinson's disease is unstable and forms higher-order protein complexes. Hum Mol Genet. 2003 Nov 1;12(21):2807-16. Epub 2003 Sep 2. PubMed PMID: 12952867.

148. **Squitieri F*,** Pustorino G, Cannella M, Toscano A, Maglione V, Morgante L, Tortorella G. Highly disabling cerebellar presentation in Huntington disease. Eur J Neurol. 2003 Jul;10(4):443-4. PubMed PMID: 12823498.

149. **Squitieri F*,** Gellera C, Cannella M, Mariotti C, Cislaghi G, Rubinsztein DC, Almqvist EW, Turner D, Bachoud-Lévi AC, Simpson SA, Delatycki M, Maglione V, Hayden MR, Donato SD. Homozygosity for CAG mutation in Huntington disease is associated with a more severe clinical course. Brain. 2003 Apr;126(Pt 4):946-55. PubMed PMID: 12615650.

150. **Squitieri F***, Cannella M, Simonelli M. CAG mutation effect on rate of progression in Huntington's disease. Neurol Sci. 2002 Sep;23 Suppl 2:S107-8. PubMed PMID: 12548366.

151. Bonifati V, Dekker MC, Vanacore N, Fabbrini G, **Squitieri** F, Marconi R, Antonini A, Brustenghi P, Dalla Libera A, De Mari M, Stocchi F, Montagna P, Gallai V, Rizzu P, van Swieten JC, Oostra B, van Duijn CM, Meco G, Heutink P; Italian Parkinson Genetics Network. Autosomal recessive early onset parkinsonism is linked to three loci: PARK2, PARK6, and PARK7. Neurol Sci. 2002 Sep;23 Suppl 2:S59-60. PubMed PMID: 12548343.

152. Bonifati V, Rizzu P, van Baren MJ, Schaap O, Breedveld GJ, Krieger E, Dekker MC, **Squitieri** F, Ibanez P, Joosse M, van Dongen JW, Vanacore N, van Swieten JC, Brice A, Meco G, van Duijn CM, Oostra BA, Heutink P. Mutations in the DJ-1 gene associated with autosomal recessive early-onset parkinsonism. Science. 2003 Jan 10;299(5604):256-9. Epub 2002 Nov 21. PubMed PMID: 12446870.

153. Bonifati V, Breedveld GJ, **Squitieri** F, Vanacore N, Brustenghi P, Harhangi BS, Montagna P, Cannella M, Fabbrini G, Rizzu P, van Duijn CM, Oostra BA, Meco G, Heutink P. Localization of autosomal recessive early-onset parkinsonism to chromosome 1p36 (PARK7) in an independent dataset. Ann Neurol. 2002 Feb;51(2):253-6. PubMed PMID: 11835383.

154. **Squitieri F*,** Cannella M, Giallonardo P, Maglione V, Mariotti C, Hayden MR. Onset and pre-onset studies to define the Huntington's disease natural history. Brain Res Bull. 2001 Oct-Nov 1;56(3-4):233-8. Review. PubMed PMID: 11719256.

155. Ladogana A, Almonti S, Petraroli R, Giaccaglini E, Ciarmatori C, Liù QG, Bevivino S, **Squitieri** F, Pocchiari M. Mutation of the PRNP gene at codon 211 in familial Creutzfeldt-Jakob disease. Am J Med Genet. 2001 Oct 1;103(2):133-7. PubMed PMID: 11568919.

156. Cannella M, Simonelli M, D'Alessio C, Pierelli F, Ruggieri S, **Squitieri F***. Presymptomatic tests in Huntington's disease and dominant ataxias. Neurol Sci. 2001 Feb;22(1):55-6. PubMed PMID: 11487199.

157. Modugno N, Currà A, Giovannelli M, Priori A, **Squitieri** F, Ruggieri S, Manfredi M, Berardelli A. The prolonged cortical silent period in patients with Huntington's disease. Clin Neurophysiol. 2001 Aug;112(8):1470-4. PubMed PMID: 11459687.

158. **Squitieri F*,** Cannella M, Piorcellini A, Brusa L, Simonelli M, Ruggieri S. Short-term effects of olanzapine in Huntington disease. Neuropsychiatry Neuropsychol Behav Neurol. 2001 Jan;14(1):69-72. PubMed PMID: 11234911.

159. **Squitieri F*,** Sabbadini G, Mandich P, Gellera C, Di Maria 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. Am J Med Genet. 2000 Dec 11;95(4):366-73. PubMed PMID: 11186892.

160. Cattaneo E, Rigamonti D, Goffredo D, Zuccato C, **Squitieri** F, Sipione S. Loss of normal huntingtin function: new developments in Huntington's disease research. Trends Neurosci. 2001 Mar;24(3):182-8. Review. PubMed PMID: 11182459.

161. **Squitieri** F*, Maglione V, Buzzi MG, Nargi E, Novelletto A, Cannella M, Simonelli M, Colonnese C, Simonelli P, Innocenzi G, Gagliardi FM, Caruso R, Ragona G, Cantore GP. Cavernous angiomas of the nervous system in Italy: clinical and genetic study. Neurol Sci. 2000 Jun;21(3):129-34. PubMed PMID: 11076000.

162. **Squitieri** F*, Berardelli A, Nargi E, Castellotti B, Mariotti C, Cannella M, Lavitrano ML, de Grazia U, Gellera C, Ruggieri S. Atypical movement disorders in the early stages of Huntington's disease: clinical and genetic analysis. Clin Genet. 2000 Jul;58(1):50-6. PubMed PMID: 10945661.

163. Giovannone B, Sabbadini G, Di Maio L, Calabrese O, Castaldo I, Frontali M, Novelleto A, **Squitieri** F*. Analysis of (CAG)n size heterogeneity in somatic and sperm cell DNA from intermediate and expanded Huntington disease gene carriers. Hum Mutat. 1997;10(6):458-64. PubMed PMID: 9401009.

164. **Squitieri** F*, Campanella G, Hayden MR. Update on genetics of Huntington's disease: availability of direct and accurate predictive test. Ital J Neurol Sci. 1996 Jun;17(3):185-7. PubMed PMID: 8856407.

165. Telenius H, Almqvist E, Kremer B, Spence N, **Squitieri** F, Nichol K, Grandell U, Starr E, Benjamin C, Castaldo I, et al. Somatic mosaicism in sperm is associated with intergenerational (CAG)n changes in Huntington disease. Hum Mol Genet. 1995 Feb;4(2):189-95. Erratum in: Hum Mol Genet 1995 May;4(5):974. PubMed PMID: 7757066.

166. **Squitieri** F, Andrew SE, Goldberg YP, Kremer B, Spence N, Zeisler J, Nichol K, Theilmann J, Greenberg J, Goto J, et al. DNA haplotype analysis of Huntington disease reveals clues to the origins and mechanisms of CAG expansion and reasons for geographic variations of prevalence. Hum Mol Genet. 1994 Dec;3(12):2103-14. PubMed PMID: 7881406.

167. Benjamin CM, Adam S, Wiggins S, Theilmann JL, Copley TT, Bloch M, **Squitieri** F, McKellin W, Cox S, Brown SA, et al. Proceed with care: direct predictive testing for Huntington disease. Am J Hum Genet. 1994 Oct;55(4):606-17. Erratum in: Am J Hum Genet 1995 Apr;56(4):1015. PubMed PMID: 7942838; PubMed Central PMCID: PMC1918294.

168. Novelletto A, Persichetti F, Sabbadini G, Mandich P, Bellone E, Ajmar F, **Squitieri** F, Campanella G, Bozza A, MacDonald ME, et al. Polymorphism analysis of the huntingtin gene in Italian families affected with Huntington disease. Hum Mol Genet. 1994 Jul;3(7):1129-32. PubMed PMID: 7981682.

169. Kremer B, Goldberg P, Andrew SE, Theilmann J, Telenius H, Zeisler J, **Squitieri** F, Lin B, Bassett A, Almqvist E, et al. A worldwide study of the Huntington's disease mutation. The sensitivity and specificity of measuring CAG repeats. N Engl J Med. 1994 May 19;330(20):1401-6. PubMed PMID: 8159192.

170. Andrew SE, Goldberg YP, Kremer B, **Squitieri** F, Theilmann J, Zeisler J, Telenius H, Adam S, Almquist E, Anvret M, et al. Huntington disease without CAG expansion: phenocopies or errors in assignment? Am J Hum Genet. 1994 May;54(5):852-63. PubMed PMID: 8178825; PubMed Central PMCID: PMC1918249.

171. Kremer B, **Squitieri** F, Telenius H, Andrew SE, Theilmann J, Spence N, Goldberg YP, Hayden MR. Molecular analysis of late onset Huntington's disease. J Med Genet. 1993 Dec;30(12):991-5. PubMed PMID: 8133510; PubMed Central PMCID: PMC1016630.

172. Goldberg YP, Andrew SE, Theilmann J, Kremer B, **Squitieri** F, Telenius H, Brown JD, Hayden MR. Familial predisposition to recurrent mutations causing Huntington's disease: genetic risk to sibs of sporadic cases. J Med Genet. 1993 Dec;30(12):987-90. PubMed PMID: 8133509; PubMed Central PMCID: PMC1016629.

173. Telenius H, Kremer HP, Theilmann J, Andrew SE, Almqvist E, Anvret M, Greenberg C, Greenberg J, Lucotte G, **Squitieri** F, et al. Molecular analysis of juvenile Huntington disease: the major influence on (CAG)n repeat length is the sex of the affected parent. Hum Mol Genet. 1993 Oct;2(10):1535-40. PubMed PMID: 8268906.

174. Goldberg YP, Kremer B, Andrew SE, Theilmann J, Graham RK, **Squitieri** F, Telenius H, Adam S, Sajoo A, Starr E, et al. Molecular analysis of new mutations for Huntington's

disease: intermediate alleles and sex of origin effects. Nat Genet. 1993 Oct;5(2):174-9. PubMed PMID: 8252043.

175. Andrew SE, Goldberg YP, Kremer B, Telenius H, Theilmann J, Adam S, Starr E, **Squitieri** F, Lin B, Kalchman MA, et al. The relationship between trinucleotide (CAG) repeat length and clinical features of Huntington's disease. Nat Genet 1993 Aug;4(4):398-403. PubMed PMID: 8401589.

176. Di Maio L, **Squitieri** F, Napolitano G, Campanella G, Trofatter JA, Conneally PM. Suicide risk in Huntington's disease. J Med Genet. 1993 Apr;30(4):293-5. PubMed PMID: 8487273; PubMed Central PMCID: PMC1016335.

177. Di Maio L, **Squitieri** F, Napolitano G, Campanella G, Trofatter JA, Conneally PM. Onset symptoms in 510 patients with Huntington's disease. J Med Genet. 1993 Apr;30(4):289-92. PubMed PMID: 8487272; PubMed Central PMCID: PMC1016334.

178. Filla A, De Michele G, Orefice G, Santorelli F, Trombetta L, Banfi S, **Squitieri** F, Napolitano G, Puma D, Campanella G. A double-blind cross-over trial of amantadine hydrochloride in Friedreich's ataxia. Can J Neurol Sci. 1993 Feb;20(1):52-5. PubMed PMID: 8467430.

179. Di Maio L, Boiano S, **Squitieri** F, Napolitano G, Cocozza S, Campanella G, Battistuzzi G. Genetic linkage analysis and presymptomatic testing in Huntington's disease. First report in Italy. Acta Neurol (Napoli). 1992 Aug-Dec;14(4-6):524-9. PubMed PMID: 1363459.

180. Campanella G, Di Maio L, Mengano A, **Squitieri** F, De Michele G, Filla A. [An ambulatory regional center for extrapyramidal and cerebellar disorders. 10 years' experience in Campania]. Riv Neurol. 1990 May-Jun;60(3):125-9. Italian. PubMed PMID: 2084815.

181. Mengano A, Di Maio L, Maggio MA, **Squitieri** F, Di Donato M, Barbieri F, Campanella G. Benign essential tremor. A clinical survey of 82 patients from Campania, a region of southern Italy. Acta Neurol (Napoli). 1989 Aug;11(4):239-46. PubMed PMID: 2801257.

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