CURRICULUM VITAE DR. FEDERICO ZARA

DATI PERSONALI

Nazionalità: Italiana

TITOLI DI STUDIO

1992:Laurea in biologia presso l'Università di Genova.

1997:Specializzazione in Genetica Medica presso l'Università di Genova.

ESPERIENZE PROFESSIONALI

- 1994-1995:Ricercatore presso il dipartimento di Neurologia del Baylor College of Medicine (Houston, USA).
- Dal gennaio 1996 all'ottobre 2002 presso il Laboratorio di Genetica Umana dell'Ospedale Galliera di Genova in qualità di specilializzando (1996-1997), borsista (1998-1999) e dirigente biologo di I livello (dal novembre 2000 all'ottobre 2002).
- Dal Novembre 2002 ad oggi presso il Laboratorio di Patologia Muscolare, Biochimica Muscolare e Neurogenetica dell'Istituto G. Gaslini di Genova in qualità di dirigente biologo di I livello.

RELAZIONI A MEETING NAZIONALI ED INTERNAZIONALI

- 21 Aprile 1996: "Mapping genes predisposing to epilepsy. A linkage study on Italian families". Genetic Analysis of the epilepsies, European Study Centre, Firenze - Hotel Villa le Rondini, 19-23 Aprile 1996.
- 22 Settembre 1997: "Genetica delle Epilessie idiopatiche". VI Congresso Nazionale Societa' Italiana di Neuroscienze, Brescia - Universita' degli studi di Brescia, 20-23 Settembre 1997.
- 2 Novembre 1999: " Genetica delle epilessie generalizzate: Aspetti molecolari . 13th Course in Clinical Epileptology, Gargnano (BS) – Palazzo Feltrinelli, 1-7 Novembre.
- 18 Novembre 1999: " Family and molecular study on Benign Infantile Familial Convulsions". VI Mediterranean Epilepsy Conference, Cairo – Hotel Sheraton, 17-19 Novembre 1999.

- 28 Novembre 2001: " Epilessie idiopatiche e canali ionici neuronali". IV Congresso Nazionale Società Italiana di Genetica Umana, Orvieto - Palazzo del Capitano del Popolo, 28-30 Novembre 2001.
- 3 Giugno 2002: "Convulsioni neonatali e infantili familiari ed epilessia mioclonica severa". Corso di aggiornamento sulla genetica dell'epilessia - Congresso Nazionale Lega Italiana contro l'Epilessia, Bologna - Palazzo Re Enzo, 2-5 Giugno 2002.
- 14 Giugno 2002: "Genetica delle Epilessie idiopatiche". XII Corso Residenziale di Genetica Medica, San Giovanni Rotondo 13-15 Giugno 2002.
- 26-28 Luglio 2002: "Genetics of Idiopathic Epilepsies". Advanced International Residential *Course Bridging basic with clinical epileptology*, Venezia 21 Luglio – 4 Agosto 2002.
- 5-6 Dicembre 2002 " Genetics of Severe Myoclonic Epilepsy" & "Autosomal Recessive Mioclonic Epilepsy of Infancy". International Symposium in honor of Charlotte Dravet *Myoclonic Epilepsies of infancy, childhood, adolescence & adulthood.* Seattle 5-6 Dicembtre 2002.

PUBBLICAZIONI

1. Zara F, Bianchi A, Avanzini A, Di Donato S, Castellotti B, Patel PI, Pandolfo M. Mapping of genes predisposing to idiopathic generalized epilepsy. Hum Mol Genet 1995; 4:1201-7.
2. Montermini L, Rodius F, Pianese L, Molto' MD, Cossee' M, Campuzano V, Cavalcanti F, Monticelli A, Palau F, Gyapay G, Wenhert M, Zara F, Patel PI, Cocozza S, Koenig M, Pandolfo M. The Friedreich ataxia critical region spans a 150 kb interval on chromosome 9q13. Am J Hum Genet 1995; 57:1061-7.
3. Campuzano V, Montermini L, Molto' MD, Pianese L, Cossee' M, Cavalcanti F, Monros E, Rodius F, Duclos F, Monticelli A, Zara F, Canizares J, Koutnikova H, Bidichindani SI, Gellera C, Brice A, Trouillas P, De Michele G, Filla A, De Frutos R, Palau F, Patel PI, Di Donato S, Mandel JL, Cocozza S, Koenig M, Pandolfo M. Friedreich's ataxia: autosomal recessive disease caused by an intronic GAA triplet repeat expansion. Science 1996; 271:1423-1427.
4. Juyal RC, Kuwano A, Kondo I, Zara F, Baldini A, Patel PI. Mosaicism for del(17)(p11.2p11.2) underlying the Smith-Magenis Syndrome. Am J Med Genet 1996; 66:193-196.
5. Minetti C, Sotgia F, Bruno C, Scartezzini P, Broda P, Bado M, Masetti E, Mazzocco M, Egeo A, Donati MA, Volonte' D, Galbiati F, Cordone G, Dagna Bricarelli F, Lisanti MP, Zara F. Mutations in the caveolin-3 gene cause autosomal dominant limb-girdle muscolar dystrophy. Nature Genetics 1998; 18:365-368.
6. Zara F, Carbone I, Dagna Bricarelli F. Genetics of epilepsies. Italian J Pediat 1998; 24:75-81.

7. Zara F, Labuda M, Garofalo PG, Durisotti C, Bianchi A, Castellotti B, Patel PI, Avanzini G, Pandolfo M. Unusual EEG pattern linked to chromosome 3p in a family with idiopathic generalized epilepsy. Neurology 1998; 51:493-498.
8. Giordano L, Accorsi P, Valseriati D, Tiberti A, Menegati E, Zara F, Vignoli A, Vigevano F. Benign infantile familial convulsions: natural history of a case and clinical characteristics of a large Italian family. Neuropediatrics 1998; 30:99-101.
9. Gennaro E, Malacarne M, Carbone I, Riggio MC, Bianchi A, Bonanni P, Boniver C, Dalla Bernardina B, De Marco P, Giordano L, Guerrini R, Santorum E, Sebastianelli R, Vecchi M, Veggiotti P, Vigevano F, Dagna Bricarelli F, Zara F. No evidence of a major locus for benign familial infantile convulsions on chromosome 19q12-q13.1. Epilepsia 1999; 40:1799-1803.
10. Carbone I, Bruno C, Sotgia F, Bado M, Broda P, Masetti E, Panella A, Zara F, Dagna Bricarelli F, Cordone G, Lisanti MP, Minetti C. Mutation in the CAV3 gene causes partial caveolin-3 deficiency and hyperckemia. Neurology 2000; 54:1373-1376
11. Zara F, Gennaro E, Stabile M, Carbone I, Malacarne M, Majello L, Santangelo R, de Falco FA, Dagna Bricarelli F. Mapping of a locus for a familial autosomal recessive idiopathic myoclonic epilepsy of infancy to chromsome 16p13. Am J Hum Genet 2000; 66:1552-1557
12. Sander T, Schulz H, Saar K, Gennaro E, Riggio MC, Bianchi A, Zara F, Luna D, Bulteau C, Kaminska A, Ville D, Cieuta C, Picard F, Prud'Homme JF, Bate L, Sundquist A, Gardiner RM, Janssen AMAJ, de Haan GJ, Kasteleijn-Nolst-Trenite' DGA, Bader A, Lindhout D, Riess O, Wienker TF, Janz D, Reis A. Genome search for susceptibility loci of common idiopathic generalised epilepsies. Hum Mol Genet 2000; 6:1465-1472.
13. Perfumo C, Cerruti Mainardi P, Cali A, Coucourde G, Zara F, Cavani S, Overhauser J, Dagna Bricarelli F, Pierluigi M. The first three mosaic cri du chat syndrome patients with two rearranged cell lines. J Med Genet 2000:37:967-72.
14. Cerruti Mainardi P, Perfumo C, Cali A, Coucorde G, Pastore G, Cavani S, Zara F, Overhauser J, Pierluigi M, Dagna Bricarelli F. Clinical and molecular characterisation of 80 patients with 5p deletion: genotype-phenotype correlation. J Med Genet 2001; 38:151-158.
15. Malacarne M, Gennaro E, Madia F, Pozzi S, Vacca D, Barone B, dalla Bernardina B, Bianchi A, Bonanni P, De Marco P, Gambardella A, Giordano L, Lispi ML, Romeo A, Santorum E, Vanadia F, Vecchi M, Veggiotti P, Vigevano F, Viri F, Dagna Bricarelli F, Zara F. Benign familial infantile convulsions: mapping of a novel locus on chromosome 2q24 and evidence for genetic heterogeneity. Am J Hum Genet 2001; 68:1521-1526

16. de Falco FA, Majello L, Santangelo R, Stabile, Dagna Bricarelli F, Zara F. Familial infantile myoclonic epilepsy clinical features in a large kindred with autosomal recessive inheritance. Epilepsia 2001; 42:1541-1548

17. Malacarne M, Madia M, Gennaro E, Vacca D, Güney AI, Buono S, Dalla Bernardina B, Gaggero R, Gobbi G, Lispi ML, Malamaci D, Melideo G, Roccella M, Sferro C, Tiberti A, Vanadia F, Vigevano F, Viri F, Vitali MR, Dagna Bricarelli F, Bianchi A, Zara F. Lack of SCN1A mutations in familial febrile seizures. Epilepsia 2002; 45:559-562

18. Sander T, Windemuth C, Schulz H, Saar K, Gennaro E, Bianchi A, Zara F, Prud'homme J-F, Dulac O, Bate L, Gardiner RM, de Haan J-G, Janssen GAMAJ, van Erp MG, Boezeman EHFJ, Lindhout D, Wienker TF, Janz D. No Evidence for a Susceptibility Locus for Idiopathic Generalized Epilepsy on Chromosome 18q21.1. Am J Med Genet 2002; 114:673-8

19. Windemuth C, Schulz H, Saar K, Gennaro E, Bianchi A, Zara F, Bulteau C, Kaminska A, Ville D, Cieuta C, Prud'homme J-F, Dulac O, Bate L, Gardiner RM, Lindhout D, Wienker TF, Janz D, Sander T. No evidence for a susceptibility locus for idiopathic generalized epilepsy on chromosome 5 in families with typical absence seizures. Epilepsy Res 2002; 51:23-29.

20. T. Sander T, Windemuth C, Schulz H, Saar K, Gennaro E, Riggio C, BianchiA, Zara F, Rudolf G, Picard F, Prud'homme J-F, Dulac O, Bate L, Robinson R, Gardiner RM, Covanis A, de Haan G-J, Janssen A M A J, van Erp MG, Boezeman EHFJ, Lindhout D, Heils A0, Nürnberg P, Janz D. Exploration of a Putative Susceptibility Locus for Idiopathic Generalized Epilepsy on Chromosome 8p12. Epilepsia 2003; 44:32-39.

21. Madia F, Gennaro E, Cecconi M, Buti D, Capovilla G, Dalla Bernardina B, Elia M, Ferrari A, Fontana E, Gaggero R, Giannotta M, Giordano L, Granata T, La Selva L, Lispi ML, Santucci M, Vanadia F, Veggiotti P, Vigliano P, Viri M, Dagna Bricarelli F, Bianchi A, **Zara F**. No evidence of GABRG2 mutations in severe myoclonic epilepsy of infancy. Epilepsy Research 2003; 53:196-200

22. Gennaro E, Veggiotti P, Malacarne M, Madia F, Cecconi M, Cardinali S, Cassetti A, Cecconi I, Bertini E, Bianchi A, Gobbi G, **Zara F**. Familial Severe Myoclonic Epilepsy of Infancy: truncation of Nav1.1 and genetic heterogeneity. Epileptic Disorders 2003; 5:21-25

23. de Falco FA, Striano P, de Falco A, Striano S, Santangelo R, Perretti A, Balbi P,Cecconi M, **Zara F**. Benign Adult Familial Myoclonic Epilepsy: genetic heterogeneity and allelelism with ADCME locus. Neurology 2003; 60:1381-1385

24. Nabbout R, Gennaro E, Dalla Bernardina B, Dulac O, Madia F, Bertini E, Capovilla G, Chiron C, Cristofori G, Elia M, Fontana E, Gaggero R, Granata T, Guerrini R, Loi M, La Selva L, Lispi ML, Matricardi A, Romeo A, Tzolas V, Valseriati D, Veggiotti P, Vigevano F, Vallée L, Dagna

Bricarelli F, Bianchi A, **Zara F**. Spectrum of SCN1A mutations in severe myoclonic epilepsy of infancy. Neurology 60:1961-1967.

25. Bonanni P, Malcarne M, Moro F, Veggiotti P, Buti D, Ferrari AR, Parrini E, Mei D, Volzone A, Zara F, Heron SE, Bordo L, Marini C, Guerrini R. Generalized epilepsy with febrile seizures plus (GEFS+): Clinical spectrum in 7 Italian families unrelated to SCN1A, SCN1B, GABRG2 gene mutations. Epilepsia *in press*
26. Berkovic SF, Heron SEB, Giordano L, Marini C, Guerrini R, Kaplan RE, Gambardella A, Steinlein OK, Grinton BE, Dean JT, Bordo L, Hodgson B, Yamamoto T, Mulley JC, Zara F, Scheffer IE. Benign familial neonatal-infantile seizures: Characterization of a new sodium channelopathy. Annals of Neurol *in press*
27. Baykan B, Madia F, Bebek N, Gianotti S, Güney AI, Cine N, Bianchi A, Gökyiğit A, Zara F. Autosomal recessive idiopathic epilepsy in an inbred family from Turkey: identification of a putative locus on chromosome 9q32-33.Epilepsia *in press*

Federico Zara CV:

Unità Operativa di Malattie Neuromuscolari dell'Ospedale Gaslini di Genova.

Si occupa di studi di genetica sulle epilessie idiomatiche, numerose pubblicazioni su riviste internazionali dal 1998 a oggi.

- **2009**: Erichello Luca; Perruolo Giuseppe; Pascarello Angelo; Formisano Pietro; Minetti Carlo; Striano Salvatore; Zara Federico; Striano Pasquale
  Autoantibodies to glutamic acid decarboxylase (GAD) in focal and generalized epilepsy: A study on 233 patients.
  Journal of neuroimmunology 2009;211(1-2):120-3.
- **2009**: Striano Pasquale; Caranci Ferdinando; Di Benedetto Raffaella; Tortora Fabio; Zara Federico; Striano Salvatore
  (1)H-MR spectroscopy indicates prominent cerebellar dysfunction in benign adult familial myoclonic epilepsy.
  Epilepsia 2009;50(6):1491-7.
- **2009**: Striano Pasquale; de Falco Fabrizio A; Minetti Carlo; Zara Federico
  Familial benign nonprogressive myoclonic epilepsies.
  Epilepsia 2009;50 Suppl 5(): 37-40.
- **2008**: Striano Pasquale; Zara Federico; Santorelli Filippo Maria; Striano Salvatore
  Topiramate-associated worsening symptoms in a patient with familial hemiplegic migraine.
  Journal of the neurological sciences 2008;272(1-2):194-5.
- **2008**: Vieusso V; Provitá S C; Schiatti E; Magnasi G; Minetti C; Zara F; Grasso M; Dagna-Bricarelli F; Di Maria E
  Inclusion body myopathy, Paget's disease of the bone and frontotemporal dementia: recurrence of the VCP R155H mutation in an Italian family and implications for genetic counselling.
  Clinical genetics 2008;74(1):54-60.
- **2008**: Madia Francesca; Striano Pasquale; Di Bonaventura Carlo; de Falco Arturo; de Falco Fabrizio A; Manfredi Mario; Casari Giorgio; Striano Salvatore; Minetti Carlo; Zara Federico
  Benign adult familial myoclonic epilepsy (BAFME): evidence of an extended founder haplotype on chromosome 2p11.1-q12.2 in five Italian families.
  Neurogenetics 2008;9(2):139-42.
- **2008**: Striano Pasquale; Striano Salvatore; Minetti Carlo; Zara Federico
  Refractory, life-threatening status epilepticus in a 3-year-old girl.
  Lancet neurology 2008;7(3):270-84.
- **2008**: Traverso Monica; Gazzerro Elisabetta; Assereto Stefania; Solgia Federica; Biancheri Roberta; Stringara Silvia; Giberti Laura; Pedemonte Marina; Wang Xiabo; Scapolan Sara; Pasquini Elisabetta; Donati Maria A; Zara Federico; Lisanti Michael P; Bruno Claudio; Minetti Carlo
  Caveolin-3 T78M and T78K missense mutations lead to different phenotypes in vivo and in vitro.
  Laboratory investigation; a journal of technical methods and pathology 2008;88(3):275-83.
- **2008**: Striano Pasquale; Soha Vito; Capovilla Giuseppe; Rubboli Guido; Di Bonaventura Carlo; Coppola Antonietta; Vitale Giuseppina; Fontanillas Luis; Gellonardo Anna Teresa; Bondi Roberto; Romeo Antonino; Vin Maurizio; Zara Federico; Striano Salvatore
  A pilot trial of levetiracetam in eyelid myoclonia with absences (Jeavons syndrome).
  Epilepsia 2008;49(3):425-30.
- **2008**: Striano Pasquale; Zara Federico; Turnbull Julie; Girard Jean-Marie; Ackerley Cameron A; Cervasio Mariarosaria; De Rosa Gaetano; Del Bassu-De Caro Maria Laura; Striano Salvatore; Minassian Berge A
  Typical progression of myoclonic epilepsy of the Lafora type: a case report.
  Nature clinical practice. Neurology 2008;4(2):106-11.
- **2007**: Biancheri Roberta; Falace Antonio; Tessa Alessandra; Pedemonte Marina; Scapolan Sara; Cassandrini Denise; Aiello Chiara; Rossi Andrea; Broda Paolo; Zara Federico; Santorelli Filippo Maria; Minetti Carlo; Bruno Claudio
  POMT2 gene mutation in limb-girdle muscular dystrophy with inflammatory changes.
  Biochemical and biophysical research communications 2007;363(4):1013-7.
- **2007**: Striano Pasquale; Coppola Antonietta; Madia Francesca; Pezzella Marianna; Ciampa Clotilde; Zara Federico; Striano Salvatore
  Life-threatening status epilepticus following gabapentin administration in a patient with benign adult familial myoclonic epilepsy.
  Epilepsia 2007;48(10):1999-8.
- **2007**: Striano Salvatore; Zara Federico; Striano Pasquale
  Comment to: overlap cases of eyelid myoclonia with absences and juvenile myoclonic epilepsy.
  Seizure : the journal of the British Epilepsy Association 2007;16(6):557-8.
- **2007**: Biancheri Roberta; Zara Federico; Bruno Claudio; Rossi Andrea; Bordo Laura; Gazzerro Elisabetta; Solgia Federica; Pedemonte Marina; Scapolan Sara; Bado Massimo; Uziel Graziella; Bugiani Marianna; Lamba Laura Doria; Costa Valeria; Schenone Angelo; Rozemuller Annemieke J M; Tortori-Donati Paolo; Lisanti Michael P; van der Knaap Marjo S; Minetti Carlo
  Phenotypic characterization of hypomyelination and congenital cataract.
  Annals of neurology 2007;62(2):121-7.

- **2007**: Striano Pasquale; Zara Federico; Coppola Antonietta; Ciampa Clotilde; Pezzella Manuela; Striano Salvatore
Epileptic myoclonus as ciprofloxacin-associated adverse effect.
Movement disorders : official journal of the Movement Disorder Society 2007;22(11):1675-6.

- **2007**: Striano P; Coppola A; Pezzella M; Ciampa C; Specchio N; Ragona F; Mancardi M M; Gennaro E; Beccaria F; Capovilla G; Rasmini P; Besana D; Coppola G G; Elia N; Granata T; Vecchi M; Vigevano F; Vini M; Gaggero R; Striano S; Zara F
An open-label trial of levetiracetam in severe myoclonic epilepsy of infancy.
Neurology 2007;69(3):250-4.

- **2007**: Mancardi Maria Margherita; Canrio Ubaldo; Schiaffino Maria Cristina; Baglietto Maria Giuseppina; Rossi Andrea; Battaglia Francesca Maria; Salomons Gajja Sophi; Jakobs Cornelis; Zara Federico; Veneselli Edvige; Gaggero Roberto
Severe epilepsy in X-linked creatine transporter defect (CRTR-D).
Epilepsia 2007;48(6):1211-3.

- **2007**: Striano Pasquale; Mancardi Maria Margherita; Biancheri Roberta; Madia Francesca; Gennaro Elena; Paravidino Roberta; Beccaria Francesca; Capovilla Giuseppe; Dalla Bernardina Bernardo; Diana Francesca; Elia Maurizio; Giordano Lucio; Gobbi Giuseppe; Granata Tiziana; Ragona Francesca; Guerrini Renzo; Marini Carla; Mei Davide; Longaretti Francesca; Romeo Antonino; Siri Laura; Specchio Nicola; Vigevano Federico; Striano Salvatore; Tortora Fabio; Rossi Andrea; Minetti Carlo; Dravet Charlotte; Gaggero Roberto; Zara Federico
Brain MRI findings in severe myoclonic epilepsy in infancy and genotype-phenotype correlations.
Epilepsia 2007;48(6):1092-6.

- **2007**: Striano Pasquale; Elefante Andrea; Coppola Antonietta; Tortora Fabio; Zara Federico; Minetti Carlo; Striano Salvatore
Dramatic response to levetiracetam in post-ischaemic Holmes' tremor.
Journal of neurology, neurosurgery, and psychiatry 2007;78(4):438-9.

- **2007**: Lohi H; Turnbull J; Zhao X C; Pullenayegum S; Tarazano L; Yahyaoui M; Mikati M A; Quinn N P; Franceschetti S; Zara F; Minassian B A
Genetic diagnosis in Lafora disease: genotype-phenotype correlations and diagnostic pitfalls.
Neurology 2007;68(13):996-1001.

- **2007**: Cannelli N; Nardocci N; Cassandrini D; Morbin M; Aiello C; Bugiani M; Criscuolo L; Zara F; Striano P; Granata T; Bertini E; Simonati A; Santorelli F M
Revelation of a novel CLN5 mutation in early juvenile neuronal ceroid lipofuscinosis.
Neuropediatrics 2007;38(1):46-9.

- **2007**: Striano Pasquale; Tortora Fabio; Evoli Amelia; Palmeri Giovannella; Elefante Andrea; Zara Federico; Tarr Philip E; Striano Salvatore
Periodic myoclonus due to cytomegalovirus encephalitis in a patient with good syndrome.
Archives of neurology 2007;64(2):277-9.

- **2007**: Striano Pasquale; Specchio Nicola; Biancheri Roberta; Cannelli Natalia; Simonati Alessandro; Cassandrini Denise; Rossi Andrea; Bruno Claudio; Fusco Lucia; Gaggero Roberto; Vigevano Federico; Bertini Enrico; Zara Federico; Santorelli Filippo M; Striano Salvatore
Clinical and electrophysiological features of epilepsy in Italian patients with CLN8 mutations.
Epilepsy & behavior : E&B 2007;10(1):187-91.

- **2007**: Falace Antonio; Striano Pasquale; Manganelli Fiore; Coppola Antonietta; Striano Salvatore; Minetti Carlo; Zara Federico
Inherited neuromyotonia: a clinical and genetic study of a family.
Neuromuscular disorders : NMD 2007;17(1):23-7.

- **2006**: Hempelmann Anne; Taylor Kirsten P; Heils Armin; Lorenz Susanne; Prud'homme Jean-Francois; Nabbout Rima; Dulac Olivier; Rudolf Gabrielle; Zara Federico; Bianchi Amedeo; Robinson Robert; Gardiner R Mark; Covanis Athanasios; Lindhout Dick; Stephani Ulrich; Elger Christian E; Weber Yvonne G; Lerche Holger; Nurnberg Peter; Kron Katherine L; Scheffer Ingrid E; Malley John C; Berkovic Samuel F; Sander Thomas
Exploration of the genetic architecture of idiopathic generalized epilepsies.
Epilepsia 2006;47(10):1682-90.

- **2006**: Mancardi Maria Margherita; Striano Pasquale; Gennaro Elena; Madia Francesca; Paravidino Roberta; Scapolan Sara; Dalla Bernardina Bernardo; Bertini Enrico; Bianchi Amedeo; Capovilla Giuseppe; Darra Francesca; Elia Maurizio; Freri Elena; Gobbi Giuseppe; Granata Tiziana; Guerrini Renzo; Pantaleoni Chiara; Parmeggiani Antonia; Romeo Antonino; Santucci Margherita; Vecchi Marilena; Veggiotti Pierangelo; Vigevano Federico; Pilzano Angela; Gaggero Roberto; Zara Federico
Familial occurrence of febrile seizures and epilepsy in severe myoclonic epilepsy of infancy (SMEI) patients with SCN1A mutations.
Epilepsia 2006;47(10):1629-35.

- **2006**: Striano Pascuale; Zara Federico; Minetti Carlo; Striano Salvatore
Epileptic seizures can follow high doses of oral vardenafil.
BMJ (Clinical research ed.) 2006;333(7572):785.

- **2006**: Madia F; Striano P; Gennaro E; Malacarne M; Paravidino R; Biancheri R; Budetta M; Cilio M R; Gaggero R; Pierluigi M; Minetti C; Zara F
Cryptic chromosome deletions involving SCN1A in severe myoclonic epilepsy of infancy.
Neurology 2006;67(7):1230-5.

- **2006**: Biancheri Roberta; Bertini Enrico; Falace Antonio; Pedemonte Marina; Rossi Andrea; D'Amico Adele; Scapolan Sara; Bergamino Laura; Petrini Stefania; Cassandrini Denise; Broda Paolo; Manfredi Mario; Zara Federico; Santorelli Filippo M; Minetti Carlo; Bruno Claudio
POMGnT1 mutations in congenital muscular dystrophy: genotype-phenotype correlation and expanded clinical spectrum.
Archives of neurology 2006;63(10):1491-5.

- **2006**: Zara Federico; Biancheri Roberta; Bruno Claudio; Bordo Laura; Assereto Stefania; Gazzerro Elisabetta; Solgia Federica; Wang Xiao Bo; Gianotti Stefania; Stringara Silvia; Pedemonte Marina; Uziel Graziella; Rossi Andrea; Schenone Angelo; Tortori-Donati Paolo; van der Knaap Marjo S; Lisanti Michael P; Minetti Carlo
Deficiency of hyccin, a newly identified membrane protein, causes hypomyelination and congenital cataract.
Nature genetics 2006;38(10):1111-3.

- **2006**: Striano Pasquale; Malacarne Michela; Cavani Simona; Pierluigi Mauro; Rinaldi Rosanna; Cavalieri Maria Luigia; Rinaldi Maria Michela; De Bernardo Carmelina; Coppola Antonietta; Pintaudi Maria; Gaggero Roberto; Grammatico Paola; Striano Salvatore; Dallapiccola Bruno; Zara Federico; Faravelli Francesca
Clinical phenotype and molecular characterization of 6q terminal deletion syndrome: Five new cases.
American journal of medical genetics. Part A 2006;140(18):1944-9.

- **2006**: Bianchini Roberta; Zara Federico; Striano Pasquale; Pedemonte Marina; Cassandrini Denise; Stringara Silvia; Margaretti Fiore; Sanforo Lucio; Schenone Angelo; Bellone Enrica; Minetti Carlo
GDAP1 mutation in autosomal recessive Charcot-Marie-Tooth with pyramidal features.
Journal of neurology 2006;253(9):1234-5.

- **2006**: Bruno Claudio; Cassandrini Denise; Martinuzzi Andrea; Toscano Antonio; Moggio Maurizio; Moraldi Lucia; Servidei Serena; Mongini Tiziana; Angelini Corrado; Musumeci Olimpia; Comi Giacomo P; Lamperti Costanza; Filosto Massimiliano; Zara Federico; Minetti Carlo
McArdle disease: the mutation spectrum of PYGM in a large Italian cohort.
Human mutation 2006;27(7):718.

- **2006**: Striano Pasquale; Lispi Maria Luisa; Gennaro Elena; Madia Francesca; Traverso Monica; Bordo Laura; Aridon Paolo; Boneschi Filippo Martinelli; Barone Baldassare; della Bernardina Bernardo; Bianchi Amedeo; Capovilla Giuseppe; De Marco Pasquale; Dulac Olivier; Gaggero Roberto; Gambardella Antonio; Nabbout Rima; Prud'homme Jean-François; Day Ruth; Vanadia Francesca; Vecchi Marilena; Veggiotti Pierangelo; Vigevano Federico; Van Maurice; Minetti Carlo; Zara Federico
Linkage analysis and disease models in benign familial infantile seizures: a study of 16 families.
Epilepsia 2006;47(6):1029-34.

- **2006**: Striano Pasquale; Coppola Antonietta; Vacca Giovanni; Zara Federico; Brescia Morra Vincenzo; Grefice Giuseppe; Striano Salvatore
Levetiracetam for cerebellar tremor in multiple sclerosis: an open-label pilot tolerability and efficacy study.
Journal of neurology 2006;253(6):762-6.

- **2006**: Elia Maurizio; Striano Pasquale; Fichera Marco; Gaggero Roberto; Castiglia Lucia; Galesi Ornella; Malacarne Michela; Pierluigi Mauro; Aurelio Carmelo; Musumeci Sebastiano A; Romano Corrado; Majorri Silvia; Grammatico Paola; Zara Federico; Striano Salvatore; Faravelli Francesca
6q terminal deletion syndrome associated with a distinctive EEG and clinical pattern: a report of five cases.
Epilepsia 2006;47(5):830-8.

- **2006**: Cannelli Natalia; Cassandrini Denise; Bertini Enrico; Striano Pasquale; Fusco Lucia; Gaggero Roberto; Specchio Nicola; Bianchini Roberta; Vigevano Federico; Bruno Claudio; Simonati Alessandro; Zara Federico; Santorelli Filippo M
Novel mutations in CLN8 in Italian variant late infantile neuronal ceroid lipofuscinosis: Another genetic hit in the Mediterranean.
Neurogenetics 2006;7(2):111-7.

- **2006**: Cassandrini Denise; Calevo Maria Grazia; Tessa Alessandra; Manfredi Giovanni; Fattori Fabiana; Meschini Maria Chiara; Carrozzo Rosalba; Tonoli Emmanuel; Pedemonte Marina; Minetti Carlo; Zara Federico; Santorelli Filippo M; Bruno Claudio
A new method for analysis of mitochondrial DNA point mutations and assess levels of heteroplasmy.
Biochemical and biophysical research communications 2006;342(2):387-93.

- **2006**: Franceschetti Silvana; Gambardella Antonio; Canafoglia Laura; Striano Pasquale; Lohi Hannes; Gennaro Elena; Tarzano Leonarda; Veggiotti Pierangelo; Sofia Vito; Biondi Roberto; Striano Salvatore; Gellera Cinzia; Arnesi Grazia; Madia Francesca; Civitelli Donata; Rocca Francesca E; Quattrone Aldo; Avanzini Giuliano; Minassian Berge; Zara Federico
Clinical and genetic findings in 26 Italian patients with Lafora disease.
Epilepsia 2006;47(3):640-3.

- **2006**: Gennaro Elena; Santorelli Filippo M; Bertini Enrico; Buti Daniela; Gaggero Roberto; Gobbi Giuseppe; Lini Marcella; Granata Tiziana; Freri Elena; Parmeggiani Antonia; Striano Pasquale; Veggiotti Pierangelo; Cardinali Simona; Briganti Franca Dagna; Minetti Carlo; Zara Federico
Somatic and germline mosaicisms in severe myoclonic epilepsy of infancy.
Biochemical and biophysical research communications 2006;341(2):489-93.

- **2006**: Assereto Stefania; Stringara Silvia; Sotgia Federica; Bonuccelli Gloria; Broccolini Aldobrando; Pedemonte Marina; Traverso Monica; Bianchini Roberta; Zara Federico; Bruno Claudio; Lisanti Michael P; Minetti Carlo
Pharmacological rescue of the dystrophin-glycoprotein complex in Duchenne and Becker skeletal muscle explants by proteasome inhibitor treatment.
American journal of physiology. Cell physiology 2006;290(2):C577-82.

- **2006**: Striano Pasquale; Bordo Laura; Lispi Maria Luisa; Specchio Nicola; Minetti Carlo; Vigevano Federico; Zara Federico
A novel SCN2A mutation in family with benign familial infantile seizures.
Epilepsia 2006;47(1):218-20.

- **2006**: Traverso Monica; Malnati Mauro; Minetti Carlo; Regis Stefano; Tedeschi Silvana; Pedemonte Marina; Bruno Claudio; Bassoni Roberto; Zara Federico
Multiplex real-time PCR for detection of deletions and duplications in dystrophin gene.
Biochemical and biophysical research communications 2006;339(1):145-50.

- **2005**: Striano Pasquale; Madia Francesca; Minetti Carlo; Striano Salvatore; Zara Federico
Electroclinical and genetic findings in a family with cortical tremor, myoclonus, and epilepsy.
Epilepsia 2005;46(12):1993-5.

- **2005**: Martinelli Boneschi Filippo; Aridon Paolo; Zara Federico; Guerrini Renzo; Marini Carla; De Fusco Maurizio; Comi Giancarlo; Casari Giorgio
No evidence of ATP1A2 involvement in 12 multiplex Italian families with benign familial infantile seizures.
Neuroscience letters 2005;388(2):71-4.

- **2005**: Baykan Betul; Striano Pasquale; Gianotti Stefania; Bebek Nerses; Gennaro Elena; Gurses Candan; Zara Federico
Late-onset and slow-progressing Lafora disease in four siblings with EPM2B mutation.
Epilepsia 2005;46(10):1695-7.

- **2005**: Minetti Carlo; Gattorno Marco; Repetto Silvia; Gregorio Andrea; Pedemonte Marina; Assereto Stefania; Zara Federico; Bruno Claudio; Martini Alberto
Chemokine receptor CCR7 is expressed in muscle fibers in juvenile dermatomyositis.
Biochemical and biophysical research communications 2005;333(2):540-3.

- **2005**: Striano P; Zara F; Striano S
Autosomal dominant cortical tremor, myoclonus and epilepsy: many syndromes, one phenotype.
Acta neurologica Scandinavica 2006;113(4):211-7.

- **2005**: Zara Federico; De Falco Fabrizio A
Autosomal recessive benign myoclonic epilepsy of infancy.
Advances in neurology 2005;95():139-45.

- **2004**: Bruno C; van Diggelen O P; Cassandrini D; Gimpelev M; Gaffné B; Donati M A; Introvini P; Alegria A; Assereto S; Morandi L; Mora M; Tonoli E; Minetti C; Traverso M; Pasquini E; Bado M; Vilarinho L; van Noort G; Mosca F; Di Mauro S; Zara F; Minetti C
Clinical and genetic heterogeneity of branching enzyme deficiency (glycogenosis type IV).
Neurology 2004;63(6):1053-8.

- **2004**: Broccolini Aldobrando; Ricci Enzo; Cassandrini Denise; Giubizzi Carla; Bruno Claudio; Tonoli Emmanuel; Silvestri Gabriella; Pescatori Mario; Rodolico Carmelo; Sinicropi Stefano; Servidei Serenella; Zara Federico; Minetti Carlo; Tonoli Pietro A; Morabella Massimiliano
Novel GNE mutations in Italian families with autosomal recessive hereditary inclusion-body myopathy.
Human mutation 2004;23(6):632.

- **2004**: Baykan Betül; Madia Francesca; Bebek Nerses; Gianotti Stefania; Güney Ahmet Ilter; Cine Nazi; Bianchi Amedeo; Gökyigit Aysen; Zara Federico
Autosomal recessive idiopathic epilepsy in an inbred family from Turkey: identification of a putative locus on chromosome 9q32-33.
Epilepsia 2004;45(5):479-87.

- **2004**: Berkovic Samuel F; Heron Sarah E; Giordano Lucio; Marini Carla; Guerrini Renzo; Kaplan Robert E; Gambardella Antonio; Steinlein Ortrud K; Grinton Bronwyn E; Dean Joanne T; Bordo Laura; Hodgson Bree L; Yamamoto Toshiyuki; Mulley John C; Zara Federico; Scheffer Ingrid E
Benign familial neonatal-infantile seizures: characterization of a new sodium channelopathy.
Annals of neurology 2004;55(4):550-7.

- **2004**: Bonanni Paolo; Malacarne Michela; Moro Francesca; Veggiotti Pierangelo; Buti Daniela; Ferrari Anna Rita; Perrini Elena; Mei Davide; Volzone Anna; Zara Federico; Heron Sarah E; Bordo Laura; Marini Carla; Guerrini Renzo
Generalized epilepsy with febrile seizures plus (GEFS+): clinical spectrum in seven Italian families unrelated to SCN1A, SCN1B, and GABRG2 gene mutations.
Epilepsia 2004;45(2):149-58.

- **2003**: Boccella P; Striano P; Zara F; Barbieri F; Sarappa C; Vacca G; de Falco F A; Striano S
Bioptically demonstrated Lafora disease without EPM2A mutation: a clinical and neurophysiological study of two sisters.
Clinical neurology and neurosurgery 2003;106(1):55-9.

- **2003**: Nabbout R; Kuchenbuch A; Gennaro E; Bahi-Buisson N; Zara F; Cheron C; Bianchi A; Brice A; Leguern E; Dulac O
Absence of mutations in major GEFS+ genes in myoclonic astatic epilepsy.
Epilepsy research 2003;56(2-3):127-33.

- **2003**: Nabbout R; Gennaro E; Dalla Bernardina B; Dulac O; Madia F; Bertini E; Capovilla G; Chiron C; Cristofori G; Elia M; Fontana E; Gaggero R; Granata T; Guerrini R; Loi M; La Selva L; Lispi M L; Matricardi A; Romeo A; Tzolas V; Valseriati D; Veggiotti P; Vigevano F; Vallee L; Dagna Bricarelli F; Bianchi A; Zara F
Spectrum of SCN1A mutations in severe myoclonic epilepsy of infancy.
Neurology 2003;60(12):1961-7.

- **2003**: de Falco F A; Striano P; de Falco A; Striano S; Santangelo R; Perretti A; Balbi P; Cecconi M; Zara F
Benign adult familial myoclonic epilepsy: genetic heterogeneity and allelism with ADCME.
Neurology 2003;60(8):1381-5.

- **2003**: Gennaro Elena; Veggiotti Pierangelo; Malacarne Michele; Madia Francesca; Cecconi Massimiliano; Cardinali Simonetta; Cassetti Alessandra; Cecconi Ilaria; Bertini Enrico; Bianchi Amedeo; Gobbi Giuseppe; Zara Federico
Familial severe myoclonic epilepsy of infancy: truncation of Nav1.1 and genetic heterogeneity.
Epileptic disorders : international epilepsy journal with videotape 2003;5(1):21-5.

- **2003**: Madia Francesca; Gennaro Elena; Cecconi Massimiliano; Buti Daniela; Capovilla Giuseppe; Dalla Bernardina Bernardo; Elia Maurizio; Ferrari Annarita; Fontana Elena; Gaggero Roberto; Giannotta Melania; Giordano Lucio; Granata Tiziana; La Selva Loretta; Luisa Lispi Maria; Santucci Margherita; Vanadia Francesca; Veggiotti Pierangelo; Vigliano Piernarda; Viri Maurizio; Dagna Bricarelli Franca; Bianchi Amedeo; Zara Federico
No evidence of GABRG2 mutations in severe myoclonic epilepsy of infancy.
Epilepsy research 2003;53(3):196-200.

- **2003**: Sander Thomas; Windemuth Christine; Schulz Herbert; Saar Kathrin; Gennaro Elena; Riggio Concetta; Bianchi Amedeo; Zara Federico; Rudolf Gabrielle; Picard Fabienne; Bulteau Christine; Kaminska Anna; Cieuta Cécile; Prudhomme Jean-François; Dulac Olivier; Bate Louise; Robinson Robert; Gardiner R Mark; Covanis Athanasios; de Haan Gerrit-Jan; Janssen Guus A M A J; van Erp M Gerard; Boezeman Eduard H J F; Lindhout Dick; Hells Armin; Nürnberg Peter; Janz Dieter;
Exploration of a putative susceptibility locus for idiopathic generalized epilepsy on chromosome 8p13.
Epilepsia 2001;44(1):12-9.

- **2002**: Windemuth C; Schulz H; Saar K; Gennaro E; Bianchi A; Zara F; Bulteau C; Kaminska A; Wile D; Cieuta C; Nabbout-Tarantino R; Prud'homme J-F; Dulac O; Bate L; Gardiner R M; Lindhout D; Wienker T F; Janz D; Sander T;
No evidence for a susceptibility locus for idiopathic generalized epilepsy on chromosome 5 in families with typical absence seizures.
Epilepsy research 2002;51(1-2):23-9.

- **2002**: Sander Thomas; Windemuth Christine; Schulz Herbert; Saar Kathrin; Gennaro Elena; Bianchi Amedeo; Zara Federico; Bulteau Christine; Kaminska Anna; Ville Dorothée; Cieuta Cécile; Prudhomme Jean-François; Dulac Olivier; Bate Louise; Gardiner R Mark; de Haan Gerrit-Jan; Janssen Guus A M A J; Witte Jorine; Halley Dicky J J; Lindhout Dick; Wienker Thomas F; Janz Dieter;
  No evidence for a susceptibility locus for idiopathic generalized epilepsy on chromosome 18q21.1.
  American journal of medical genetics 2002;114(6):673-8.
- **2002**: Malacarne Michela; Madia Francesca; Gennaro Elena; Vacca Daniela; Güney A Ilter; Buono Salvatore; Bernardina Bernardo Dalla; Gaggero Roberto; Gobio Giuseppe; Lispi Maria Luisa; Malaventi Daniela; Meliciani Giustino; Roccella Maurizio; Sferro Caterina; Tiberti Alessandra; Vanadia Francesca; Vigevano Federico; Vin Franco; Vitali Maria Rosa; Bricarelli Franca Dagna; Bianchi Amedeo; Zara Federico
  Lack of SCN1A mutations in familial febrile seizures.
  Epilepsia 2002;43(5):559-62.
- **2001**: de Falco F A; Majello L; Santangelo R; Stabile M; Bricarelli F D; Zara F
  Familial infantile myoclonic epilepsy: clinical features in a large kindred with autosomal recessive inheritance.
  Epilepsia 2001;42(12):1541-8.
- **2001**: Malacarne M; Gennaro E; Madia F; Pozzi S; Vacca D; Barone B; dalla Bernardina B; Bianchi A; Bonanni P; De Marco P; Gambardella A; Giordano L; Lispi M L; Romeo A; Santorum E; Vanadia F; Vecchi M; Veggiotti P; Vigevano F; Viri F; Bricarelli F D; Zara F
  Benign familial infantile convulsions: mapping of a novel locus on chromosome 2q24 and evidence for genetic heterogeneity.
  American journal of human genetics 2001;68(6):1521-6.
- **2001**: Mainardi P C; Perfumo C; Cali A; Coucourde G; Pastore G; Cavani S; Zara F; Overhauser J; Pierluigi M; Bricarelli F D
  Clinical and molecular characterisation of 80 patients with 5p deletion: genotype-phenotype correlation.
  Journal of medical genetics 2001;38(3):151-8.
- **2000**: Perfumo C; Cerruti Mainardi P; Cali A; Coucourde G; Zara F; Cavani S; Overhauser J; Bricarelli F D; Pierluigi M
  The first three mosaic cri du chat syndrome patients with two rearranged cell lines.
  Journal of medical genetics 2000;37(12):967-72.
- **2000**: Sander T; Schulz H; Saar K; Gennaro E; Riggio M C; Bianchi A; Zara F; Luna D; Bulteau C; Kaminska A; Ville D; Cieuta C; Picard F; Prudhomme J F; Bate L; Sundquist A; Gardiner R M; Janssen G A; de Haan G J; Kasteleijn-Nolst-Trenite D G; Bader A; Lindhout D; Rees G; Wienker T F; Janz D; Reis A
  Genome search for susceptibility loci of common idiopathic generalised epilepsies.
  Human molecular genetics 2000;9(10):1465-72.
- **2000**: Zara F; Gennaro E; Stabile M; Carbone I; Malacarne M; Majello L; Santangelo R; de Falco F A; Bricarelli F D
  Mapping of a locus for a familial autosomal recessive idiopathic myoclonic epilepsy of infancy to chromosome 16p13.
  American journal of human genetics 2000;66(5):1552-7.
- **2000**: Carbone I; Bruno C; Sotgia F; Bado M; Broda P; Masetti E; Panella A; Zara F; Bricarelli F D; Cordone G; Lisanti M P; Minetti C
  Mutation in the CAV3 gene causes partial caveolin-3 deficiency and hyperCKemia.
  Neurology 2000;54(6):1373-6.
- **1999**: Gennaro E; Malacarne M; Carbone I; Riggio M C; Bianchi A; Bonanni P; Bonner C; Dalla Bernardina B; De Marco P; Giordano L; Guerrini R; Santorum E; Sebastianelli R; Vecchi M; Veggiotti P; Vigevano F; Bricarelli F D; Zara F
  No evidence of a major locus for benign familial infantile convulsions on chromosome 19q12-q13.1.
  Epilepsia 1999;40(12):1799-803.
- **1999**: Giordano L; Accorsi P; Valseriati D; Tiberti A; Meneghini E; Zara F; Vignoli A; Vigevano F
  Benign infantile familial convulsions: natural history of a case and clinical characteristics of a large Italian family.
  Neuropediatrics 1999;30(2):99-101.
- **1998**: Minetti C; Sotgia F; Bruno C; Scartezzini P; Broda P; Bado M; Masetti E; Mazzocco M; Egeo A; Donati M A; Volonte D; Galbiati F; Cordone G; Bricarelli F D; Lisanti M P; Zara F
  Mutations in the caveolin-3 gene cause autosomal dominant limb-girdle muscular dystrophy.
  Nature genetics 1998;18(4):365-8.