

GIAN MARIA FABRIZI

Born, [REDACTED] Italy.

CURRENT POSITIONS

Associate Professor, Department of Neuroscience, Biomedicine and Movement Sciences, Section of Clinical Neurology, University of Verona. University Hospital GB Rossi, Verona, Italy

Head, Clinical Neurobiology Unit, Neurology Institute, Department of Neuroscience, Azienda Ospedaliera Universitaria Integrata di Verona, University Hospital GB Rossi, Verona.

EDUCATION

1984 Medical School degree, University of Siena, Italy.

1987-1989 Post-Doctoral Research Fellowship, H. Houston Merritt Clinical Research Center for Muscular Dystrophy and Related Diseases (Director Prof. S. Di Mauro), Columbia University College of Physician and Surgeons, New York, New York USA

1990 Ph.D. in Anthropology, Anthropometry and Physiology of the Sport, University of Siena, Italy

1990 Specialization in Neurology, University of Siena, Italy

PROFESSIONAL APPOINTMENTS, ACADEMIC POSITIONS, ACHIEVEMENTS

1985-1986. Medical Officer in the Italian Army

1990-1996. Assistant Professor of Neurology, Department of Neurological Science, University Hospital Santa Maria alle Scotte, Siena

1996-2002. Assistant Professor of Neurology, Department of Neuroscience, Biomedicine and Movement Sciences, Section of Clinical Neurology, University of Verona. University Hospital GB Rossi, Verona.

2002- Associate Professor of Neurology, Department of Neuroscience, Biomedicine and Movement Sciences, Section of Clinical Neurology, University of Verona. University Hospital GB Rossi, Verona.

2017- Qualification for full professorship in Neurology ("Abilitazione Nazionale Prima Fascia" Settore concorsuale 06/D6 Neurologia)

INSTITUTIONAL OFFICES

2019 President of the Italian Peripheral Nerve Society ("Associazione italiana per lo studio del Sistema Nervoso Periferico")

MAIN RESEARCH FIELDS

Clinical research on inherited neuropathies (Charcot-Marie-Tooth disease and related neuropathies, inherited amyloid neuropathies): genotype-phenotype correlations, natural history studies, therapeutic trials.

Molecular genetics and pathology of inherited neuropathies.

PUBLICATIONS

Author of more than 140 papers on peer-reviewed journals indexed in PubMed

Bibliometric Indexes: Scopus h-index 33, citations 4405; Web-of-Science h-index 33, 4174 citations.

Publications 2016-2020

1. Grandis M, Obici L, Luigetti M, Briani C, Benedicenti F, Bisogni G, Canepa M, Cappelli F, Danesino C, Fabrizi GM, Fenu S, Ferrandes G, Gemelli C, Manganelli F, Mazzeo A, Melchiorri L, Perfetto F, Pradotto LG, Rimessi P, Tini G, Tozza S, Trevisan L, Pareyson D, Mandich P. Recommendations for pre-symptomatic genetic testing for hereditary transthyretin amyloidosis in the era of effective therapy: a multicenter Italian consensus. *Orphanet J Rare Dis.* 2020;15(1):348. doi: 10.1186/s13023-020-01633-z.
2. Manganelli F, Fabrizi GM, Luigetti M, Mandich P, Mazzeo A, Pareyson D. Hereditary transthyretin amyloidosis overview. *Neurol Sci.* 2020. doi: 10.1007/s10072-020-04889-2.
3. Filosto M, Cotti Piccinelli S, Gazzina S, Foresti C, Frigeni B, Servalli MC, Sessa M, Cosentino G, Marchioni E, Ravaglia S, Briani C, Castellani F, Zara G, Bianchi F, Del Carro U, Fazio R, Filippi M, Magni E, Natalini G, Palmerini F, Perotti AM, Bellomo A, Osio M, Scopelliti G, Carpo M, Rasera A, Squintani G, Doneddu PE, Bertasi V, Cotelli MS, Bertolasi L, Fabrizi GM, Ferrari S, Ranieri F, Caprioli F, Grappa E, Broglio L, De Maria G, Leggio U, Poli L, Rasulo F, Latronico N, Nobile-Orazio E, Padovani A, Uncini A. Guillain-Barré syndrome and COVID-19: an observational multicentre study from two Italian hotspot regions. *J Neurol Neurosurg Psychiatry.* 2020;jnnp-2020-324837. doi: 10.1136/jnnp-2020-324837.
4. Boso F, Zanette G, Baldinotti F, Bertelloni S, Taioli F, Monaco S, Fabrizi GM, Cavallaro T. Convergent pathological and ultrasound features in hereditary syndromic and non-syndromic minifascicular neuropathy related to DHH. *J Peripher Nerv Syst.* 2020;25(4):423-428. doi: 10.1111/jns.12417.
5. Luigetti M, Romozzi M, Bisogni G, Cardellini D, Cavallaro T, Di Paolantonio A, Fabrizi GM, Fenu S, Gentile L, Grandis M, Marucci G, Massucco S, Mazzeo A, Pareyson D, Romano A, Russo M, Schenone A, Tagliapietra M, Tozza S, Vita G, Sabatelli M. hATTR Pathology: Nerve Biopsy Results from Italian Referral Centers. *Brain Sci.* 2020;10(11):780. doi: 10.3390/brainsci10110780.
6. Thomas FP, Saporta M, Attarian S, Sevilla T, Mascaró RS, Fabrizi GM, Genovese F, Gray A, Bull S, Tanesse D, Rego M, Moore A, Hollett C, Monteiro K, Paoli X, Llewellyn S, Larkin M, Boutalbi Y. Patient-reported impact of Charcot-Marie-Tooth disease: protocol for a real-world digital lifestyle study. *Neurodegener Dis Manag.* 2021;11(1):21-33. doi: 10.2217/nmt-2020-0044.
7. Cardellini D, Zanette G, Taioli F, Bertolasi L, Ferrari S, Cavallaro T, Fabrizi GM. CIDP, CMT1B, or CMT1B plus CIDP? *Neurol Sci.* 2020. doi: 10.1007/s10072-020-04789-5.
8. Pisciotta C, Calabrese D, Santoro L, Tramacere I, Manganelli F, Fabrizi GM, Schenone A, Cavallaro T, Grandis M, Previtali SC, Allegri I, Padua L, Pazzaglia C, Saveri P, Quattrone A, Valentino P, Tozza S, Gentile L, Russo M, Mazzeo A, Trapasso MC, Parazzini F, Vita G, Pareyson D; Italian CMT Network. Pregnancy in Charcot-Marie-Tooth disease: Data from the Italian CMT national registry. *Neurology.* 2020;95(24):e3180-e3189. doi: 10.1212/WNL.00000000000010860.
9. Prada V, Benedetti L, Cocito D, Briani C, Nobile Orazio E, Gallia F, Antonini G, Manganelli F, Fabrizi GM, Germano F, Grandis M, Schenone A. High-dose immunoglobulin pulse therapy and risk of Covid19 infection. *J Neurol.* 2020:1-3. doi: 10.1007/s00415-020-10146-5.
10. Salvalaggio A, Coraci D, Cacciavillani M, Obici L, Mazzeo A, Luigetti M, Pastorelli F, Grandis M, Cavallaro T, Bisogni G, Lozza A, Gemelli C, Gentile L, Ermani M, Fabrizi GM, Plasmati R, Campagnolo M, Castellani F, Gasparotti R, Martinoli C, Padua L, Briani C. Nerve ultrasound in hereditary transthyretin amyloidosis: red flags and possible progression biomarkers. *J Neurol.* 2020. doi: 10.1007/s00415-020-10127-8.
11. Russo M, Obici L, Bartolomei I, Cappelli F, Luigetti M, Fenu S, Cavallaro T, Chiappini MG, Gemelli C, Pradotto LG, Manganelli F, Leonardi L, My F, Sampaolo S, Briani C, Gentile L, Stancanelli C, Di Buduo E, Pacciolla P, Salvi F, Casagrande S, Bisogni G, Calabrese D, Vanoli F, Di Iorio G, Antonini G, Santoro L, Mauro A, Grandis M, Di Girolamo M, Fabrizi GM, Pareyson D,

- Sabatelli M, Perfetto F, Rapezzi C, Merlini G, Mazzeo A, Vita G. ATTRv amyloidosis Italian Registry: clinical and epidemiological data. *Amyloid*. 2020;27(4):259-265. doi: 10.1080/13506129.2020.1794807.
12. Dubbioso R, Nobile-Orazio E, Manganelli F, Santoro L, Briani C, Cocito D, Tedeschi G, Di Lazzaro V, Fabrizi GM; SIN, SINC and ASNP. Dealing with immune-mediated neuropathies during COVID-19 outbreak: practical recommendations from the task force of the Italian Society of Neurology (SIN), the Italian Society of Clinical Neurophysiology (SINC) and the Italian Peripheral Nervous System Association (ASNP). *Neurol Sci*. 2020;41(6):1345-1348. doi: 10.1007/s10072-020-04448-9.
13. Marastoni D, Africa L, Peretti A, Bocci S, Insana L, Ferrari S, Ginanneschi F, Zanette G, Fabrizi GM, Giannini F. Sustained response to subcutaneous immunoglobulins in chronic ataxic neuropathy with anti-disialosyl IgM antibodies (CANDA): report of two cases and review of the literature. *J Neurol*. 2020;267(8):2353-2361. doi: 10.1007/s00415-020-09843-y.
14. Saveri P, De Luca M, Nisi V, Pisciotta C, Romano R, Piscosquito G, Reilly MM, Polke JM, Cavallaro T, Fabrizi GM, Fossa P, Cichero E, Lombardi R, Lauria G, Magri S, Taroni F, Pareyson D, Bucci C. Charcot-Marie-Tooth Type 2B: A New Phenotype Associated with a Novel RAB7A Mutation and Inhibited EGFR Degradation. *Cells*. 2020;9(4):1028. doi: 10.3390/cells9041028.
15. Magrinelli F, Fabrizi GM, Santoro L, Manganelli F, Zanette G, Cavallaro T, Tamburin S. Pharmacological treatment for familial amyloid polyneuropathy. *Cochrane Database Syst Rev*. 2020;4(4):CD012395. doi: 10.1002/14651858.CD012395.pub2.
16. Fabrizi GM, Høyer H, Taioli F, Cavallaro T, Hilmarsen HT, Squintani GM, Zanette G, Braathen GJ. Inherited motor-sensory neuropathy with upper limb predominance associated with the tropomyosin-receptor kinase fused gene. *Neuromuscul Disord*. 2020;30(3):227-231. doi: 10.1016/j.nmd.2019.12.007.
17. Campagnolo M, Taioli F, Cacciavillani M, Ruiz M, Luigetti M, Salvalaggio A, Castellani F, Testi S, Ferrarini M, Cavallaro T, Gasparotti R, Fabrizi GM, Briani C. Sporadic hereditary neuropathies misdiagnosed as chronic inflammatory demyelinating polyradiculoneuropathy: Pitfalls and red flags. *J Peripher Nerv Syst*. 2020;25(1):19-26. doi: 10.1111/jns.12362.
18. Gentile L, Russo M, Fabrizi GM, Taioli F, Ferrarini M, Testi S, Alfonzo A, Aguenouz M, Toscano A, Vita G, Mazzeo A. Charcot-Marie-Tooth disease: experience from a large Italian tertiary neuromuscular center. *Neurol Sci*. 2020;41(5):1239-1243. doi: 10.1007/s10072-019-04219-1.
19. Boso F, Armirotti A, Taioli F, Ferrarini M, Nobbio L, Cavallaro T, Fabrizi GM. Deoxysphingolipids as candidate biomarkers for a novel SPTLC1 mutation associated with HSAN-I. *Neurol Genet*. 2019;5(6):e365. doi: 10.1212/NXG.0000000000000365.
20. Mori L, Signori A, Prada V, Pareyson D, Piscosquito G, Padua L, Pazzaglia C, Fabrizi GM, Picelli A, Schenone A; TreSPE study group. Treadmill training in patients affected by Charcot-Marie-Tooth neuropathy: results of a multicenter, prospective, randomized, single-blind, controlled study. *Eur J Neurol*. 2020;27(2):280-287. doi: 10.1111/ene.14074.
21. Zanette G, Tamburin S, Taioli F, Lauriola MF, Badari A, Ferrarini M, Cavallaro T, Fabrizi GM. Nerve size correlates with clinical severity in Charcot-Marie-Tooth disease 1A. *Muscle Nerve*. 2019;60(6):744-748. doi: 10.1002/mus.26688.
22. Previtali SC, Zhao E, Lazarevic D, Pipitone GB, Fabrizi GM, Manganelli F, Mazzeo A, Pareyson D, Schenone A, Taroni F, Vita G, Bellone E, Ferrarini M, Garibaldi M, Magri S, Padua L, Pennisi E, Pisciotta C, Riva N, Scaioli V, Scarlato M, Tozza S, Geroldi A, Jordanova A, Ferrari M, Molineris I, Reilly MM, Comi G, Carrera P, Devoto M, Bolino A. Expanding the spectrum of genes responsible for hereditary motor neuropathies. *J Neurol Neurosurg Psychiatry*. 2019;90(10):1171-1179. doi: 10.1136/jnnp-2019-320717.

23. Pazzaglia C, Padua L, Pareyson D, Schenone A, Aiello A, Fabrizi GM, Cavallaro T, Santoro L, Manganelli F, Coraci D, Gemignani F, Vitetta F, Quattrone A, Mazzeo A, Russo M, Vita G; CMT-TRIAAL Group. Are novel outcome measures for Charcot-Marie-Tooth disease sensitive to change? The 6-minute walk test and StepWatch™ Activity Monitor in a 12-month longitudinal study. *Neuromuscul Disord*. 2019;29(4):310-316. doi: 10.1016/j.nmd.2019.01.009.
24. Zanette G, Fabrizi GM, Taioli F, Lauriola MF, Badari A, Ferrarini M, Cavallaro T, Tamburin S. Nerve ultrasound findings differentiate Charcot-Marie-Tooth disease (CMT) 1A from other demyelinating CMTs. *Clin Neurophysiol*. 2018;129(11):2259-2267. doi: 10.1016/j.clinph.2018.08.016.
25. Mori L, Prada V, Signori A, Pareyson D, Piscosquito G, Padua L, Pazzaglia C, Fabrizi GM, Smania N, Picelli A, Schenone A; TreSPE Study Group. Outcome measures in the clinical evaluation of ambulatory Charcot-Marie-Tooth 1A subjects. *Eur J Phys Rehabil Med*. 2019;55(1):47-55. doi: 10.23736/S1973-9087.18.05111-0.
26. Fabrizi GM, Tamburin S, Cavallaro T, Zanette G. Reply to "Relationship between age and nerve dimensions in Charcot-Marie-Tooth disease. Do we know the reality?". *Clin Neurophysiol*. 2018;129(6):1335-1336. doi: 10.1016/j.clinph.2018.03.003.
27. Baldinotti F, Cavallaro T, Dati E, Baroncelli GI, Bertini V, Valetto A, Massart F, Fabrizi GM, Zanette G, Peroni D, Bertelloni S. Novel Familial Variant of the Desert Hedgehog Gene: Clinical Findings in Two Sisters with 46,XY Gonadal Dysgenesis or 46,XX Karyotype and Literature Review. *Horm Res Paediatr*. 2018;89(3):141-149. doi: 10.1159/000485507.
28. Fabrizi GM, Tamburin S, Cavallaro T, Cabrini I, Ferrarini M, Taioli F, Magrinelli F, Zanette G. The spectrum of Charcot-Marie-Tooth disease due to myelin protein zero: An electrodiagnostic, nerve ultrasound and histological study. *Clin Neurophysiol*. 2018;129(1):21-32. doi: 10.1016/j.clinph.2017.09.117.
29. Bongianni M, Orrù C, Groveman BR, Sacchetto L, Fiorini M, Tonoli G, Triva G, Capaldi S, Testi S, Ferrari S, Cagnin A, Ladogana A, Poleggi A, Colaizzo E, Tiple D, Vaianella L, Castriciano S, Marchioni D, Hughson AG, Imperiale D, Cattaruzza T, Fabrizi GM, Pocchiari M, Monaco S, Caughey B, Zanusso G. Diagnosis of Human Prion Disease Using Real-Time Quaking-Induced Conversion Testing of Olfactory Mucosa and Cerebrospinal Fluid Samples. *JAMA Neurol*. 2017;74(2):155-162. doi: 10.1001/jamaneurol.2016.4614.
30. van der Zee J, Gijssels I, Van Mossevelde S, Perrone F, Dillen L, Heeman B, Bäumer V, Engelborghs S, De Bleecker J, Baets J, Gelpi E, Rojas-García R, Clarimón J, Lleó A, Diehl-Schmid J, Alexopoulos P, Perneczky R, Synofzik M, Just J, Schöls L, Graff C, Thonberg H, Borroni B, Padovani A, Jordanova A, Sarafov S, Tournev I, de Mendonça A, Miltenberger-Miltényi G, Simões do Couto F, Ramirez A, Jessen F, Heneka MT, Gómez-Tortosa E, Danek A, Cras P, Vandenberghe R, De Jonghe P, De Deyn PP, Sleegers K, Cruts M, Van Broeckhoven C, Goeman J, Nuytten D, Smets K, Robberecht W, Damme PV, Bleecker J, Santens P, Dermaut B, Versijpt J, Michotte A, Ivanoiu A, Deryck O, Bergmans B, Delbeck J, Bruylant M, Willems C, Salmon E, Pastor P, Ortega-Cubero S, Benussi L, Ghidoni R, Binetti G, Hernández I, Boada M, Ruiz A, Sorbi S, Nacmias B, Bagnoli S, Sorbi S, Sanchez-Valle R, Llado A, Santana I, Rosário Almeida M, Frisoni GB, Maetzler W, Matej R, Fraidakis MJ, Kovacs GG, Fabrizi GM, Testi S. TBK1 Mutation Spectrum in an Extended European Patient Cohort with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. *Hum Mutat*. 2017;38(3):297-309. doi: 10.1002/humu.23161.
31. Opri R, Fabrizi GM, Cantalupo G, Ferrarini M, Simonati A, Dalla Bernardina B, Darra F. Progressive Myoclonus Epilepsy in Congenital Generalized Lipodystrophy type 2: Report of 3 cases and literature review. *Seizure*. 2016;42:1-6. doi: 10.1016/j.seizure.2016.08.008.
32. Manganelli F, Pisciotto C, Reilly MM, Tozza S, Schenone A, Fabrizi GM, Cavallaro T, Vita G, Padua L, Gemignani F, Laurà M, Hughes RA, Solari A, Pareyson D, Santoro L; CMT-TRIAAL and CMT-TRAUK Group. Nerve conduction velocity in CMT1A: what else can we tell? *Eur J Neurol*. 2016;23(10):1566-71. doi: 10.1111/ene.13079.

33. Gregianin E, Pallafacchina G, Zanin S, Crippa V, Rusmini P, Poletti A, Fang M, Li Z, Diano L, Petrucci A, Lispi L, Cavallaro T, Fabrizi GM, Muglia M, Boaretto F, Vettori A, Rizzuto R, Mostacciuolo ML, Vazza G. Loss-of-function mutations in the SIGMAR1 gene cause distal hereditary motor neuropathy by impairing ER-mitochondria tethering and Ca²⁺ signalling. *Hum Mol Genet.* 2016;25(17):3741-3753. doi: 10.1093/hmg/ddw220.
34. Luigetti M, Padua L, Coraci D, Fabrizi GM, Romano A, Sabatelli M. Nerve ultrasound in CMT2E/CMT1F due to NEFL mutation: Confirmation of an axonal pathology. *Clin Neurophysiol.* 2016;127(9):2990-2991. doi: 10.1016/j.clinph.2016.06.024.
35. Luigetti M, Fabrizi GM, Bisogni G, Romano A, Taioli F, Ferrarini M, Bernardo D, Rossini PM, Sabatelli M. Erratum to "Charcot-Marie-Tooth type 2 and distal hereditary motor neuropathy: Clinical, neurophysiological and genetic findings from a single-centre experience" [*Clin. Neurol. Neurosurg.* 144 (2016) 67-71]. *Clin Neurol Neurosurg.* 2016:S0303-8467(16)30182-2. doi: 10.1016/j.clineuro.2016.05.011.
36. Padua L, Pazzaglia C, Pareyson D, Schenone A, Aiello A, Fabrizi GM, Cavallaro T, Santoro L, Manganelli F, Gemignani F, Vitetta F, Quattrone A, Mazzeo A, Russo M, Vita G; CMT-TRIAAL Group. Novel outcome measures for Charcot-Marie-Tooth disease: validation and reliability of the 6-min walk test and StepWatch(™) Activity Monitor and identification of the walking features related to higher quality of life. *Eur J Neurol.* 2016;23(8):1343-50. doi: 10.1111/ene.13033.
37. Cortese A, Vita G, Luigetti M, Russo M, Bisogni G, Sabatelli M, Manganelli F, Santoro L, Cavallaro T, Fabrizi GM, Schenone A, Grandis M, Gemelli C, Mauro A, Pradotto LG, Gentile L, Stancanelli C, Lozza A, Perlini S, Piscosquito G, Calabrese D, Mazzeo A, Obici L, Pareyson D. Erratum to: Monitoring effectiveness and safety of Tafamidis in transthyretin amyloidosis in Italy: a longitudinal multicenter study in a non-endemic area. *J Neurol.* 2016;263(5):925-926. doi: 10.1007/s00415-016-8116-1.
38. Cortese A, Vita G, Luigetti M, Russo M, Bisogni G, Sabatelli M, Manganelli F, Santoro L, Cavallaro T, Fabrizi GM, Schenone A, Grandis M, Gemelli C, Mauro A, Pradotto LG, Gentile L, Stancanelli C, Lozza A, Perlini S, Piscosquito G, Calabrese D, Mazzeo A, Obici L, Pareyson D. Monitoring effectiveness and safety of Tafamidis in transthyretin amyloidosis in Italy: a longitudinal multicenter study in a non-endemic area. *J Neurol.* 2016;263(5):916-924. doi: 10.1007/s00415-016-8064-9.
39. Luigetti M, Fabrizi GM, Bisogni G, Romano A, Taioli F, Ferrarini M, Bernardo D, Rossini PM, Sabatelli M. Charcot-Marie-Tooth type 2 and distal hereditary motor neuropathy: Clinical, neurophysiological and genetic findings from a single-centre experience. *Clin Neurol Neurosurg.* 2016;144:67-71. doi: 10.1016/j.clineuro.2016.03.007.
40. Biasini F, Portaro S, Mazzeo A, Vita G, Fabrizi GM, Taioli F, Toscano A, Rodolico C. TRPV4 related scapuloperoneal spinal muscular atrophy: Report of an Italian family and review of the literature. *Neuromuscul Disord.* 2016;26(4-5):312-5. doi: 10.1016/j.nmd.2016.02.010.
41. Vogrig A, Tonin P, Fabrizi GM, Fenzi F, Beltramello A, Sala F, Meglio M, Monaco S. Spinal arachnoid cyst as a cause of isolated, progressive, bilateral C5-C6 radiculopathy. *Spine J.* 2016;16(6):e367-8. doi: 10.1016/j.spinee.2015.11.061.

Verona, January 14th 2021


Gian Maria Fabrizi