

Elena Abati

WORK EXPERIENCE

Basic Research Activity

[03/2015 – Current]

Neural Stem Cells Lab and Molecular Biology and Genetics Lab, Neuroscience Section, IRCCS Ospedale Maggiore Policlinico, University of Milan, Italy

Active participation to basic research projects focused on:

- development of *in vitro* and *in vivo* disease models (ALS, MSA, GBA2 and CMT2A) to study pathogenic mechanisms
- development of therapeutic approaches based on RNA-interference and gene replacement therapy for Charcot-Marie-Tooth type 2A, an inherited neuropathy caused by *MFN2* gene mutations, including assessment of construct safety and efficacy in *in vivo* and *in vitro* models

Clinical Research Activity

[03/2015 – Current]

Neurology Unit and Neuromuscular and Rare diseases Unit, IRCCS Ospedale Maggiore Policlinico, Milan

Participation to clinical trials, involvement in clinical evaluation of patients, patient's data collection and analysis, creation of databases (neuromuscular diseases, ataxias, hereditary neuropathies), literature review.

Currently working on the following clinical projects:

- phenotypical and genetic characterization of a cohort of patients with known and novel *MFN2* gene mutations
- natural history study of patients with *MFN2* mutations
- description of families with inherited neuropathies and atypical genetic and phenotypic features
- clinical trials of novel therapeutic strategies for Spinal Muscular Atrophy (SMA)
- CSF and serum analysis of neurofilaments in SMA patients treated with antisense oligonucleotides

Sub-Investigator

[10/2020 – Current]

Monocentric prospective observational trial "Natural History of *MFN2* gene-related diseases", sponsored by Italian Ministry of Health "Ricerca Corrente 2020".

Status: Recruiting.

Sub-investigator

[11/2018 – Current]

Multicentric clinical trial "A Study to Investigate the Safety, Tolerability, Pharmacokinetics, Pharmacodynamics and Efficacy of Risdiplam (RO7034067) in Type 2 and 3 Spinal Muscular Atrophy (SMA) Participants (SUNFISH)", NCT02908685, sponsored by Hoffman-La Roche.

Status: Active, not recruiting.

Sub-Investigator

[11/2018 – Current]

Multicentric clinical trial "A Study of Risdiplam (RO7034067) in Adult and Pediatric Participants With Spinal Muscular Atrophy (Jewelfish)", NCT03032172, sponsored by Hoffman-La Roche.

Status: Recruiting.

Volunteer Emergency Medical Technician

[01/2012 – 07/2016]

Volunteer work as EMT with the Association "Croce D'Oro Milano".

Completion of the Anpas Training Course of Regione Lombardia.

EDUCATION AND TRAINING

Residency in Neurology

Neurology Department, IRCCS Foundation "Ospedale Maggiore Policlinico", University of Milan [29/12/2017 – Current]

Currently attending the fourth year

Habilitation to the profession of Medical Doctor – State Exam

Università degli Studi di Milano, Milan, Italy [31/07/2017]

Member of the Medical Board of Milano (Ordine dei Medici Chirurghi ed Odontoiatri di Milano)

Participation in the "MDS-ES Winter School for Young Neurologists" organized by the International Parkinson and Movement Disorders Society (MDS)

University of Catania [01/02/2017 – 04/02/2017]

Address: (Italy)

Selected to participate in a 4-days intensive course aimed at gaining an in-depth knowledge of movement disorders and their differential diagnosis; the course involved the presentation of a clinical case which ranked within the 10 best cases presented.

Clinical Neurology Internship

Neurology Department, Foundation I.R.C.C.S. "Ospedale Maggiore Policlinico", University of Milan [03/2015 – 12/2017]

Clinical activity in Neurology Inpatient and Outpatient Clinic, focused on Movement Disorders, Neurodegenerative and Neuromuscular Diseases. Actively taking part in clinical rounds, patient diagnosis, treatment and management, clinical cases discussion, Magnetic Resonance Imaging interpretation and practical skills.

Research Internship

Neural Stem Cells Lab, , IRCCS Foundation "Ospedale Maggiore Policlinico ", University of Milan [03/2015 – 12/2017]

Active participation to basic research projects focused on the development of *in vitro* and *in vivo* disease models

Master's Degree in Medicine and Surgery

University of Milan [27/07/2016]

Address: Milan (Italy)

Mark: summa cum laude

Title of dissertation: "Generation of oligodendrocytes from iPSCs of patients with Multiple System Atrophy"

Participation in the MD/PhD Program of the University of Milan

[09/2012 – 07/2016]

Tutor: Prof. Stefania Corti, Neural Stem Cells Lab, IRCCS Foundation "Ospedale Maggiore Policlinico ", University of Milan

Selected participant of the MD/PhD program, with the aim of completing the first year of PhD during Medical School (60/180 ECTS credits).

Total ECTS credits obtained: 60 (1 year of the PhD program).

Winner of a Research Scholarship

[07/2015 – 09/2015]

Recipient of a travel grant of the University of Milan for a Research Internship at the Herscot Centre for Tuberous Sclerosis Complex, Massachusetts General Hospital, with production of an abstract titled "Cannabidiol treatment of refractory epileptic spasms: an open label study", presented at the "69th Annual Meeting of American Epilepsy Society"

Research Internship

[10/2012 – 02/2015]

Neuropharmacology Lab, "San Paolo" Hospital, University of Milan

Participation in projects aimed at the development of a neural stem cell-based therapeutic approach for traumatic spinal injuries, under the supervision of Prof. Alfredo Gorio

Research Internship

Molecular Medicine Lab, IFOM/IEO Hospital, Milan [05/2012 – 09/2012]

Research work focused on the characterization of pathogenic genes in malignant breast tumours, under the supervision of Prof. Salvatore Pece

High School Graduation

[06/2010]

Classical studies Lyceum (graduation score: 100/100)

LANGUAGE SKILLS

Other language(s):

English

LISTENING C2 READING C2 WRITING C2

SPOKEN PRODUCTION C2 SPOKEN INTERACTION C2

INSTITUTIONAL ACTIVITY

Chief Resident

[10/2019 – Current]

Residents' Representative at the Board of Medical and non-Medical Residents of the University of Milan

[10/2020 – Current]

HONOURS AND AWARDS

Winner of the 2021 Neurology Resident & Fellow Writing Award

"Neurology" Journal - Resident & Fellow Section [02/2021]

Awarded for the manuscript "Education Research: Effect of the COVID-19 pandemic on neurology trainees in Italy: A resident-driven survey", Authors: Elena Abati, Gianluca Costamagna, First published September 16, 2020, DOI: <https://doi.org/10.1212/WNL.0000000000010878>

Best Investigator Award

EAN Investigator Award Committee, European Academy of Neurology [05/2020]

For Oral Presentation at the 6th Congress of the European Academy of Neurology - 1st Virtual Congress 2020

Title: "Knockdown and Replacement of MFN2: a Gene Therapy to treat Dominantly Inherited Peripheral Neuropathy CMT2A"

Authors: **E. Abati, F. Rizzo, R. De Gioia, S. Bono, G. Citterio, N. Bresolin, G. Comi, M. Nizzardo, S. Corti**

Travel Grant for 16th International Congress of Neuromuscular Diseases 2021, Valencia, Spain

Selected as Plenary Speaker

Travel Grant for 7th Congress of the European Academy of Neurology, Vienna, Austria

European Academy of Neurology [2020]

Travel Grant for 5th Congress of the European Academy of Neurology, Oslo, Norway

European Academy of Neurology [2019]

Travel Grant for 4th Congress of the European Academy of Neurology, Lisbon, Portugal

European Academy of Neurology [2018]

Winner of a Research Scholarship

MD/PhD Program of the University of Milan [2015]

Travel grant for a Research Internship at the Herscot Centre for Tuberous Sclerosis Complex, Massachusetts General Hospital

CONFERENCES AND SEMINARS

Speaker at ICNMD Digital Congress

[11/09/2020 – 14/11/2020]

Talk Title: "Combination of RNA-Interference and Gene Therapy to Treat Charcot-Marie-Tooth Type 2A"

Invited Chairman to the BraYn Symposia, Mario Negri Institute, Milan, Italy

[14/11/2019 – 16/11/2019]

Session Neurodegeneration

Oral Presentation at the 6th Congress of the European Academy of Neurology - 1st Virtual Congress

[23/05/2020]

Title: "Knockdown and Replacement of MFN2: a Gene Therapy to treat Dominantly Inherited Peripheral Neuropathy CMT2A"

Authors: **E. Abati**, F. Rizzo, R. De Gioia, S. Bono, G. Citterio, N. Bresolin, G. Comi, M. Nizzardo, S. Corti

Oral Presentation at the 50th Congress of the Italian Neurological Society (SIN), Bologna, Italy

[13/10/2019]

Title: "Identification of two novel *MFN2* mutations in CMT2A patients"

Authors: **E. Abati**, A. Manini, R. Del Bo, S. Salani, F. Rizzo, G. Citterio, P. Mandich, M.T. Bassi, N. Bresolin, G.P. Comi, S. Corti

Oral Presentation at the 5th Congress of the European Academy of Neurology, Oslo, Norway

[01/07/2019]

Title: "Evaluation of RNA interference/gene therapy combination as potential therapeutic approach for Charcot-Marie-Tooth 2A"

Authors: **E. Abati**, F. Rizzo, S. Bono, S. Salani, A. Bordoni, V. Melzi, M. Ruepp, S. Pagliarani, K. Barbullushi, C. Cordigliero, N. Bresolin, G. Comi, M. Nizzardo, S. Corti

Oral Presentation at the 4th Congress of the European Academy of Neurology, Lisbon, Portugal

[19/07/2018]

Title: "Generation and characterization of iPSC-derived oligodendrocytes of patients with Multiple System Atrophy"

Authors: **E. Abati**, G. Monzio Compagnoni, N. Bresolin, G. Comi, S. Corti, A. Di Fonzo

EDITORIAL ACTIVITY

Member of the Editorial Board of Frontiers in Neurology

[02/2021 – Current]

Member of the Editorial Board of BMC Neurology

[11/2020 – Current]

Peer Reviewer for indexed journals

- Neural Regeneration Research (indexed in Scopus and Web of Science, IF 3.171)
- Journal of Multiple Sclerosis (indexed in EBSCO)

POSTER PRESENTATIONS

Selected poster presentation at the 6th Congress of the European Academy of Neurology - 1st Virtual Congress

[23/05/2020 – 26/05/2020]

Title: "The role of LRP10 mutations in Parkinson's disease and Dementia with Lewy Bodies"

Authors: A. Manini, L. Straniero, E. Monfrini, **E. Abati**, M. Vizziello, G. Franco, S. Duga, A. Di Fonzo

Selected poster presentation at the BraYn Symposia, Mario Negri Institute, Milan, Italy

[14/11/2019 – 16/11/2019]

Title: "Combined RNA interference and gene therapy targeting MFN2 for the treatment of Charcot-Marie-Tooth 2A (CMT2A)"

Authors: **E. Abati**, F. Rizzo, S. Bono, S. Salani, A. Bordoni, V. Melzi, M. Ruepp, S. Pagliarani, K. Barbullushi, C. Cordiglieri, N. Bresolin, G. Comi, M. Nizzardo, S. Corti

Selected poster presentations at the 50th Congress of the Italian Neurological Society (SIN), Bologna, Italy

[12/10/2019 – 15/10/2019]

1) Title: "Can cerebrospinal fluid neurofilaments represent a reliable pharmacodynamic biomarker for Spinal Muscular Atrophy?"

Authors: Faravelli I, Magri F, Meneri M, Velardo D, **Abati E**, Parente V, Saccomanno D, Ronchi D, Stocchetti N, Calderini E, D'Angelo MG, Albamonte E, Sansone V, Comi G, Corti S, Govoni A.

2) Title: "Pediatric anti-HMGCR necrotizing myopathy resembling limb-girdle muscular dystrophy"

Authors: Velardo D, **Abati E**, Brusa R, Magri F, Govoni A, Meneri M, Sciacco M, Cinnante C, Ciscato P, Corti S, Bresolin N, Moggio M, Comi G.

3) Title: "Central nervous system lesions in a young and immunocompetent patient: not only inflammation"

Authors: Gagliardi D, Brusa R, **Abati E**, Faravelli I, Mauri E, Meneri M, Velardo D, Triulzi F, Bresolin N, Comi G, Corti S.

Selected poster presentation at the 5th Congress of the European Academy of Neurology, Oslo, Norway

[28/06/2019 – 01/07/2019]

Title: "Distinguishing primary central nervous system lymphoma from demyelinating lesions in young patients"

Authors: Brusa R., Meneri M., Faravelli I., Gagliardi D., Velardo D., Mauri E., **Abati E.**, Triulzi F., Valcamonica G., Corti S., Bresolin N., Comi G. P.

Selected poster presentation at the XIX National Congress of the Italian Myology Association (AIM), Bergamo, Italy

[05/06/2019 – 08/06/2019]

1) Title: "Pediatric anti-HMGCR necrotizing myopathy resembling limb-girdle muscular dystrophy"

Authors: Velardo D., **Abati E.**, Brusa R., Magri F., Govoni A., Meneri M., Sciacco M., Peverelli L., Cinnante C., Triulzi F., Corti S., Bresolin N., Moggio M., Comi GP.

2) Title: "Founder mutation in Eastern Europe patients with GNE myopathy"

Authors: Brusa R., Del Bo R., Ronchi D., Peverelli L., Velardo D., Meneri M., Magri F., Govoni A., Faravelli I., Gagliardi D., Mauri E., **Abati E.**, Costamagna G., Cinnante C., Sciacco M., Moggio M., Corti S., Comi GP., Torrente Y.

Selected poster presentation at 71st AAN Annual Meeting, Philadelphia, USA

[04/05/2019 – 10/05/2019]

Title: "RNAi/gene therapy combined approach as therapeutic strategy for Charcot-Marie-Tooth 2A"

Authors: Rizzo F., Bono S., Salani S., Bordoni A., Melzi V., Ruepp M., Pagliarani S., Barbullushi K., **Abati E.**, Cordigliero C., Bresolin N., Comi GP., Nizzardo, M., Corti S.

Selected poster presentation at the 69° Annual Meeting of American Epilepsy Society, Philadelphia, USA

[02/12/2015]

Titolo: "Cannabidiol treatment of refractory epileptic spasms: an open label study"

Authors: **E. Abati**, E. Hess, A. Morgan, P.L. Bruno, E. Thiele

PUBLICATIONS ON PEER-REVIEWED JOURNAL

Education Research: Impact of SARS-CoV-2 pandemic on Neurology trainees in Italy,: A resident-driven survey

Abati E, Costamagna G. Neurology. 2020 Sep 16:10.1212/WNL.0000000000010878. Epub ahead of print.
PMID: 32938783. <https://doi.org/10.1212/WNL.0000000000010878>

Estimating the impact of COVID-19 pandemic on services provided by Italian Neuromuscular Centers: an Italian Association of Myology survey of the acute phase.

Mauri E* (equal contributor), **Abati E*** (equal contributor), Musumeci O, Rodolico C, D'Angelo MG, Mirabella M, Lucchini M, Bello L, Pegoraro E, Maggi L, Manneschi L, Gemelli C, Grandis M, Zuppa A, Massucco S, Benedetti L, Caponnetto C, Schenone A, Prelle A, Previtali SC, Scarlato M, D'Amico A, Bertini E, Pennisi EM, De Giglio L, Pane M, Mercuri E, Mongini T, Ricci F, Berardinelli A, Astrea G, Lenzi S, Battini R, Ricci G, Torri F, Siciliano G, Santorelli FM, Ariatti A, Filosto M, Passamano L, Politano L, Scutifero M, Tonin P, Fossati B, Panicucci C, Bruno C, Ravaglia S, Monforte M, Tasca G, Ricci E, Petrucci A, Santoro L, Ruggiero L, Barp A, Albamonte E, Sansone V, Gagliardi D, Costamagna G, Govoni A, Magri F, Brusa R, Velardo D, Meneri M, Sciacco M, Corti S, Bresolin N, Moroni I, Messina S, Di Muzio A, Nigro V, Liguori R, Antonini G, Toscano A, Minetti C, Comi GP; Italian Association of Myology. *Acta Myol.* 2020 Jun 1;39(2):57-66. PMID: 32904925; PMCID: PMC7460733. doi: [10.36185/2532-1900-008](https://doi.org/10.36185/2532-1900-008)

Management of patients with neuromuscular disorders at the time of the SARS-CoV-2 pandemic.

Costamagna G* (equal contributor), **Abati E*** (equal contributor), Bresolin N, Comi GP, Corti S. *J Neurol.* 2020 Aug 17;1-12. Epub ahead of print. PMID: 32804279; PMCID: PMC7429942. doi: [10.1007/s00415-020-10149-2](https://doi.org/10.1007/s00415-020-10149-2)

Juvenile cataract in association with tuberous sclerosis complex

Geffrey AL, Geenen KR, **Abati E**, Greenstein SH, VanderVeen DK, Levy RL, Davidson SL, McGarrey MP, Thiele EA, Aronow ME. *Ophthalmic Genet.* 2020 Aug;41(4):345-349. doi: 10.1080/13816810.2020.1755989. Epub 2020 Apr 27. Erratum in: *Ophthalmic Genet.* 2020 Aug;41(4):I. PMID: 32340510.

Glial cells involvement in spinal muscular atrophy: Could SMA be a neuroinflammatory disease?

Abati E, Citterio G, Bresolin N, Comi GP, Corti S. *Neurobiol Dis.* 2020 Jul;140:104870. doi: 10.1016/j.nbd.2020.104870. Epub 2020 Apr 12. PMID: 32294521.

Silence superoxide dismutase 1 (SOD1): a promising therapeutic target for amyotrophic lateral sclerosis (ALS)

Abati E, Bresolin N, Comi G, Corti S. *Expert Opin Ther Targets.* 2020 Apr;24(4):295-310. doi: 10.1080/14728222.2020.1738390. Epub 2020 Mar 14. PMID: 32125907

Nusinersen treatment and cerebrospinal fluid neurofilaments: An explorative study on Spinal Muscular Atrophy type 3 patients

Faravelli I, Meneri M, Saccomanno D, Velardo D, **Abati E**, Gagliardi D, Parente V, Petrozzi L, Ronchi D, Stocchetti N, Calderini E, D'Angelo G, Chidini G, Prandi E, Ricci G, Siciliano G, Bresolin N, Comi GP, Corti S, Magri F, Govoni A. *J Cell Mol Med.* 2020 Mar;24(5):3034-3039. doi: 10.1111/jcmm.14939. Epub 2020 Feb 7. PMID: 32032473; PMCID: PMC7077557.

Mitochondrial Dysregulation and Impaired Autophagy in iPSC-Derived Dopaminergic Neurons of Multiple System Atrophy

Monzio Compagnoni G, Kleiner G, Samarani M, Aureli M, Faustini G, Bellucci A, Ronchi D, Bordonì A, Garbellini M, Salani S, Fortunato F, Frattini E, **Abati E**, Bergamini C, Fato R, Tabano S, Miozzo M, Serratto G, Passafaro M, Deleidi M, Silipigni R, Nizzardo M, Bresolin N, Comi GP, Corti S, Quinzii CM, Di Fonzo A. *Stem Cell Reports.* 2018 Nov 13;11(5):1185-1198. doi: 10.1016/j.stemcr.2018.09.007. Epub 2018 Oct 18. PMID: 30344007; PMCID: PMC6234905.

Herpes Simplex virus type 2 myeloradiculitis with a pure motor presentation in a liver transplant recipient

Abati E, Gagliardi D, Velardo D, Meneri M, Conte G, Cinnante C, Bresolin N, Comi G, Corti S. *Transpl Infect Dis.* 2020 Feb;22(1):e13236. doi: 10.1111/tid.13236. Epub 2020 Jan 1. PMID: 31868290.

Central Nervous System Involvement in Common Variable Immunodeficiency: A Case of Acute Unilateral Optic Neuritis in a 26-Year-Old Italian Patient

Abati E, Faravelli I, Magri F, Govoni A, Velardo D, Gagliardi D, Mauri E, Brusa R, Bresolin N, Fabio G, Comi GP, Carrabba M, Corti S. *Front Neurol.* 2018 Nov 30;9:1031. doi: 10.3389/fneur.2018.01031. PMID: 30555409; PMCID: PMC6284006.

Advances, Challenges, and Perspectives in Translational Stem Cell Therapy for Amyotrophic Lateral Sclerosis

Abati E, Bresolin N, Comi G, Corti S. *Mol Neurobiol.* 2019 Oct;56(10):6703-6715. doi: 10.1007/s12035-019-1554-x. Epub 2019 Mar 26. PMID: 30911936.

Ophthalmoplegia Due to Miller Fisher Syndrome in a Patient With Myasthenia Gravis

Brusa R, Faravelli I, Gagliardi D, Magri F, Cogiamanian F, Saccomanno D, Cinnante C, Mauri E, **Abati E**, Bresolin N, Corti S, Comi GP. *Front Neurol.* 2019 Aug 13;10:823. doi: 10.3389/fneur.2019.00823. PMID: 31456730; PMCID: PMC6700242.

Hyperacute extensive spinal cord infarction and negative spine magnetic resonance imaging: a case report and review of the literature

Costamagna G, Meneri M, **Abati E**, Brusa R, Velardo D, Gagliardi D, Mauri E, Cinnante C, Bresolin N, Comi G, Corti S, Faravelli I. *Medicine (Baltimore).* 2020 Oct 23;99(43):e22900. doi: 10.1097/MD.00000000000022900. PMID: 33120840; PMCID: PMC7581089

Disease Modeling and Therapeutic Strategies in CMT2A: State of the Art

Barbullushi K, **Abati E**, Rizzo F, Bresolin N, Comi GP, Corti S. *Mol Neurobiol.* 2019 Sep;56(9):6460-6471. doi: 10.1007/s12035-019-1533-2. Epub 2019 Mar 4. PMID: 30830587.

Can Intestinal Pseudo-Obstruction Drive Recurrent Stroke-Like Episodes in Late-Onset MELAS Syndrome? A Case Report and Review of the Literature

Gagliardi D, Mauri E, Magri F, Velardo D, Meneri M, **Abati E**, Brusa R, Faravelli I, Piga D, Ronchi D, Triulzi F, Peverelli L, Sciacco M, Bresolin N, Comi GP, Corti S, Govoni A. *Front Neurol.* 2019 Jan 31;10:38. doi: 10.3389/fneur.2019.00038. PMID: 30766507; PMCID: PMC6365425.

Preconditioning and Cellular Engineering to Increase the Survival of Transplanted Neural Stem Cells for Motor Neuron Disease Therapy

Abati E, Bresolin N, Comi GP, Corti S. *Mol Neurobiol.* 2019 May;56(5):3356-3367. doi: 10.1007/s12035-018-1305-4. Epub 2018 Aug 17. PMID: 30120734.

Pregnancy outcomes in women with spinal muscular atrophy: A review

Abati E, Corti S. *J Neurol Sci.* 2018 May 15;388:50-60. doi: 10.1016/j.jns.2018.03.001. Epub 2018 Mar 2. PMID: 29627031.

In vitro models of multiple system atrophy from primary cells to induced pluripotent stem cells

Abati E, Di Fonzo A, Corti S. *J Cell Mol Med.* 2018 May;22(5):2536-2546. doi: 10.1111/jcmm.13563. Epub 2018 Mar 4. PMID: 29502349; PMCID: PMC5908105.

Animal Models of CMT2A: State-of-art and Therapeutic Implications

De Gioia R, Citterio G, **Abati E**, Nizzardo M, Bresolin N, Comi GP, Corti S, Rizzo F. *Mol Neurobiol.* 2020 Dec; 57(12):5121-5129. doi: 10.1007/s12035-020-02081-3. Epub 2020 Aug 27. PMID: 32856204; PMCID: PMC7541381.

Neural Stem Cell Transplantation for Neurodegenerative Diseases

De Gioia R, Biella F, Citterio G, Rizzo F, **Abati E**, Nizzardo M, Bresolin N, Comi GP, Corti S. Int J Mol Sci. 2020 Apr 28;21(9):3103. doi: 10.3390/ijms21093103. PMID: 32354178; PMCID: PMC7247151.

Fluid intake and chronic kidney disease: effect of coaching an increase in fluid intake on kidney function decline

Bozzano V, **Abati E**; Gruppo di Autoformazione Metodologica (GrAM). Intern Emerg Med. 2018 Dec;13(8): 1283-1285. doi: 10.1007/s11739-018-1952-5. Epub 2018 Sep 22. PMID: 30244426.

18/06/2021
