
Appendix 2 Coinvestigators

Name	Location	Role	Contribution
Eleonora Minacapilli	Fondazione IRCCS Istituto Neurologico Carlo Besta Developmental Neurology Unit, Milan, Italy	Data entry	Clinical support and coordination among the team
Riccardo Zanin	Fondazione IRCCS Istituto Neurologico Carlo Besta Developmental Neurology Unit, Milan, Italy	Clinical evaluator	Clinical support and coordination among the team
Maria Teresa Arnoldi	Fondazione IRCCS Istituto Neurologico Carlo Besta Developmental Neurology Unit, Milan, Italy	Clinical evaluator	Clinical support and coordination among the team
Stefano Scarparo	Institute of Experimental Neurology (INSPE), Division of Neuroscience, IRCCS San Raffaele Scientific Institute, Milan, Italy	Sub Investigator	Clinical support and coordination among the team
Giovanni Antonini	Department of Neuroscience, Mental Health and Sensory Organs (NESMOS), Sapienza University of Rome, Sant'Andrea Hospital, Rome, Italy	Sub Investigator	Clinical support and coordination among the team
Ilaria Bitetti	Department of Neurosciences, Pediatric Neurology, Santobono-Pausilipon Children's Hospital, Naples, Italy	Sub Investigator	Clinical support and coordination among the team

Luca Bello	Neurology Unit, Azienda Ospedale Padova, Padua, Italy	Sub Investigator	Clinical support and coordination among the team
Vittoria Zangaro	Neurology Unit, Azienda Ospedale Padova, Padua, Italy	Sub Investigator	Clinical support and coordination among the team
Maria Sframeli	Department of Clinical and Experimental Medicine, University of Messina, Messina, Italy	Sub Investigator	Clinical support and coordination among the team
Alessio Masaracchio	Department of Clinical and Experimental Medicine, University of Messina, Messina, Italy	Sub Investigator	Clinical support and coordination among the team
Adele Rocchetti	National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy	Sub Investigator	Clinical support and coordination among the team
Andrea Magnolato	Institute for Maternal and Child Health, IRCCS, Burlo Garofolo, Trieste, Italy	Sub Investigator	Clinical support and coordination among the team
Antonino Lupica	Section of Neurology, Department of Biomedicine, Neuroscience, and Advanced Diagnostics (BiND), University of Palermo, Palermo, Italy	Sub Investigator	Clinical support and coordination among the team
Vincenzo Di Stefano	Section of Neurology, Department of Biomedicine, Neuroscience, and Advanced Diagnostics (BiND), University of Palermo, Palermo, Italy	Sub Investigator	Clinical support and coordination among the team

Francesca Magri	Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Neurology Unit, Milan, Italy	Sub Investigator	Clinical support and coordination among the team
Megi Meneri	Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Neurology Unit, Milan, Italy	Sub Investigator	Clinical support and coordination among the team
Laura Antonaci	Centro Clinico Nemo, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy	Sub Investigator	Clinical support and coordination among the team
Giampaolo Cicala	Centro Clinico Nemo, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy	Sub Investigator	Clinical support and coordination among the team
Anna Lia Frongia	Centro Clinico Nemo, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy	Sub Investigator	Clinical support and coordination among the team
Costanza Cutrona	Centro Clinico Nemo, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy	Sub Investigator	Clinical support and coordination among the team
Chiara Bravetti	Centro Clinico Nemo, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy	Study nurse	Clinical support and coordination among the team
Beatrice Berti	Centro Clinico Nemo, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy	Sub Investigator	Clinical support and coordination among the team

Daniela Leone	Centro Clinico Nemo, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy	Sub Investigator	Clinical support and coordination among the team
Concetta Palermo	Centro Clinico Nemo, Fondazione Policlinico Universitario Agostino Gemelli IRCCS, Rome, Italy	Sub Investigator	Clinical support and coordination among the team
Enrico Bertini	Department of Neurosciences, Unit of Neuromuscular and Neurodegenerative Disorders, Bambino Gesù Children's Hospital, IRCCS, Rome, Italy	Sub Investigator	Clinical support and coordination among the team
Antonella Longo	Department of Neurosciences, Unit of Neuromuscular and Neurodegenerative Disorders, Bambino Gesù Children's Hospital, IRCCS, Rome, Italy	Data entry	Clinical support and coordination among the team
Michela Catteruccia	Department of Neurosciences, Unit of Neuromuscular and Neurodegenerative Disorders, Bambino Gesù Children's Hospital, IRCCS, Rome, Italy	Sub Investigator	Clinical support and coordination among the team
Claudia Dosi	Fondazione IRCCS Istituto Neurologico Carlo Besta Developmental Neurology Unit, Milan, Italy	Sub Investigator	Clinical support and coordination among the team
Silvia Bonanno	Neuroimmunology and Neuromuscular Disorders Unit,	Sub Investigator	Clinical support and coordination among the team

	Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy		
Emilio Albamonte	The NEMO Center in Milan, Neurorehabilitation Unit, University of Milan, ASST Niguarda Hospital, Milan, Italy	Sub Investigator	Clinical support and coordination among the team
Francesca Salmin	The NEMO Center in Milan, Neurorehabilitation Unit, University of Milan, ASST Niguarda Hospital, Milan, Italy	Clinical evaluator	Clinical support and coordination among the team
Simone Morando	Center of Translational and Experimental Myology, and Dept. of Neuroscience, Rehabilitation, Ophtalmology, Genetics, Maternal and Child Health, University of Genova, IRCCS Istituto Giannina Gaslini, Genova, Italy	Clinical evaluator	Clinical support and coordination among the team
Noemi Brolatti	Center of Translational and Experimental Myology, and Dept. of Neuroscience, Rehabilitation, Ophtalmology, Genetics, Maternal and Child Health, University of Genova, IRCCS Istituto Giannina Gaslini, Genova, Italy	Sub Investigator	Clinical support and coordination among the team

Chiara Brusa	AOU Città della Salute e della Scienza di Torino, presidio Molinette e OIRM (SS Malattie neuromuscolari e SC Neuropsichiatria Infantile), Turin, Italy	Sub Investigator	Clinical support and coordination among the team
Ilaria Cavallina	AOU Città della Salute e della Scienza di Torino, presidio Molinette e OIRM (SS Malattie neuromuscolari e SC Neuropsichiatria Infantile), Turin, Italy	Sub Investigator	Clinical support and coordination among the team
Giulio Gadaleta	AOU Città della Salute e della Scienza di Torino, presidio Molinette e OIRM (SS Malattie neuromuscolari e SC Neuropsichiatria Infantile), Turin, Italy	Sub Investigator	Clinical support and coordination among the team
Sara Bortolani	AOU Città della Salute e della Scienza di Torino, presidio Molinette e OIRM (SS Malattie neuromuscolari e SC Neuropsichiatria Infantile), Turin, Italy	Sub Investigator	Clinical support and coordination among the team
Michela Aringolo	Department of Neurological Sciences, AOU Ospedali Riuniti di Ancona, Ancona, Italy	Sub Investigator	Clinical support and coordination among the team
Alessandra Govoni	AOU Pisana (Department of Clinical and Experimental Medicine), Neurology Unit, Pisa, Italy	Sub Investigator	Clinical support and coordination among the team

Giulia Ricci	AOU Pisana (Department of Clinical and Experimental Medicine), Neurology Unit, Pisa, Italy	Sub Investigator	Clinical support and coordination among the team
Francesca Torri	AOU Pisana (Department of Clinical and Experimental Medicine), Neurology Unit, Pisa, Italy	Sub Investigator	Clinical support and coordination among the team
Simona Damioli	Department of Clinical and Experimental Sciences, University of Brescia (Italy); NeMO- Brescia Clinical Center for Neuromuscular Diseases, Brescia (Italy)	Sub Investigator	Clinical support and coordination among the team
Filomena Caria	Department of Clinical and Experimental Sciences, University of Brescia (Italy); NeMO- Brescia Clinical Center for Neuromuscular Diseases, Brescia (Italy)	Sub Investigator	Clinical support and coordination among the team
Stefano Cotti Piccinelli	Department of Clinical and Experimental Sciences, University of Brescia (Italy); NeMO- Brescia Clinical Center for Neuromuscular Diseases, Brescia (Italy)	Sub Investigator	Clinical support and coordination among the team
Annamaria Gallone	Neuroimmunology and Neuromuscular Disorders Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy	Sub Investigator	Clinical support and coordination among the team

Rocco Liguori	UOC Clinica Neurologica, IRCCS Institute of Neurological Sciences of Bologna, Bologna, Emilia-Romagna, Italy	Sub Investigator	Clinical support and coordination among the team
Matteo Sacchini	Metabolic Unit, A. Meyer Children's Hospital, 50139 Florence, Italy	Sub Investigator	Clinical support and coordination among the team
Chiara Ticci	Metabolic Unit, A. Meyer Children's Hospital, 50139 Florence, Italy	Sub Investigator	Clinical support and coordination among the team
Maria Ucci	Neurology unit, Azienda Ospedaliero-Universitaria, Policlinico Bari "Amaducci", Bari, Italy	Sub Investigator	Clinical support and coordination among the team
Maria Elena Pessa	Neurology Unit, Department of Neurosciences, University Hospital Santa Maria della Misericordia, Udine, Friuli-Venezia Giulia, Italy	Sub Investigator	Clinical support and coordination among the team
Stefano Parravicini	Department of Child Neuropsychiatry, Fondazione Istituto Neurologico Nazionale C Mondino Istituto di Ricovero e Cura a Carattere Scientifico, Pavia, Italy	Sub Investigator	Clinical support and coordination among the team
Eleonora Salomon	Dipartimento di Salute della Donna e del Bambino, Università di Padova, Padua, Italy;	Sub Investigator	Clinical support and coordination among the team
Elena Briganti	Neuroimmunology and Neuromuscular Disorders Unit,	Sub Investigator	Clinical support and coordination among the team

	Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy		
Gaia Scarpini	Neuroimmunology and Neuromuscular Disorders Unit, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy	Sub Investigator	Clinical support and coordination among the team
Luisa Politano	Cardiomyology and Medical Genetics Unit, Università degli Studi della Campania Luigi Vanvitelli Scuola di Medicina e Chirurgia, Napoli, Italy	Sub Investigator	Clinical support and coordination among the team
Esther Piccillo	Cardiomyology and Medical Genetics Unit, Università degli Studi della Campania Luigi Vanvitelli Scuola di Medicina e Chirurgia, Napoli, Italy	Sub Investigator	Clinical support and coordination among the team