

# LORENZO FONTANELLI

## EDUCATION

- November 2023      **Diploma di Specializzazione in Neurologia**, *Università di Pisa*
- Final Dissertation:** “Fenotipizzazione clinica di un'ampia coorte di pazienti affetti da Distrofia Miotonica di tipo 1 e sviluppo di un modello cellulare basato su organoidi”
- Grade: 110/110 cum Laude.
- May 2023            **Fellow of the European Board of Neurology (FEBN)**, 15<sup>th</sup> European Board of Neurology Examination, *European Academy of Neurology & European Union of Medical Specialists - Joint Education Board*
- Grade: Excellent.
- May 2023 - September 2023      **Visiting Scholar**, Aguirre's Lab, *Institute for Quantitative Health Science & Engineering, Michigan State University (MI, USA)*
- Manipulation and analysis of induced pluripotent stem cells (iPSCs) and embryonic stem cells. Generation, manipulation and multidimensional analyses of cardiac organoids derived from iPSCs of patients affected with Myotonic Dystrophy type 1 as *in vitro* models of the disease.
- April 2023            **Premio di Studio “Fondazione Un Passo per te”**, *Fondazione Un Passo per Te, per la ricerca nelle malattie neuromuscolari, Pisa*
- Grant awarded for a project on complex cellular models for the study of Myotonic Dystrophy type 1 by means of cardiac organoids.
- March 2023            **Diploma di Perfezionamento in Ecografia Muscoloscheletrica**, *University of Pisa*
- Post-Graduate Diploma, Corso di Perfezionamento Universitario in Ecografia Muscoloscheletrica
- March 2023            **Attended XX Corso Residenziale di: Elettroencefalografia e Tecniche Correlate**, *University of Pisa and SINC, Società Italiana di Neurofisiologia Clinica*
- May 2022            **EAN Spring School**, *European Academy of Neurology*
- May 2022            **Gymnasium Ecografico 2022, Upgrade Course per giovani SIUMB**
- November 2019 – November 2023      **Neurology Resident**, *University of Pisa*
- Diagnosis, treatment and follow up of acute and chronic neurological diseases, with a special interest in neuromuscular disorders. Since my first year of residency, I have followed the clinical and research activities of the Neuromuscular department. I followed, as a sub-investigator, CHAMPION-ALS Phase III trial of ultomiris in ALS, discontinued on August, 2021.

Since October 2021 I have started to perform intrathecal infusions of nusinersen in patients affected with spinal muscular atrophy and tofersen in patients with amyotrophic lateral sclerosis. I have collaborated to write a multidisciplinary protocol aimed to recognize early respiratory involvement in ALS to allow better patients stratification by means of deep phenotyping and multiparametric investigations using electrophysiologic techniques (Motor Unit Number Estimation, MUNE) and diaphragmatic ultrasound assessment (Multiparametric Assessment of Early Respiratory Involvement in ALS, M-AERIALS). Since January 2022 I have started to follow patients undergoing Chimeric antigen receptor (CAR) T-cell therapy in order to develop predictive measures of neurological toxicity. In 2022 I enrolled in the advanced course on musculoskeletal ultrasound “Corso di Perfezionamento in Ecografia Muscolo-Scheletrica” of the University of Pisa in collaboration with Società Italiana di Ultrasonologia in Medicina e Biologia, SIUMB.

May 2019 –  
November 2023

**Locum tenens, General Practitioner**

February 2019 -  
May 2019

**Erasmus Traineeship, School of Pharmacy and Biomedical Sciences, University of Portsmouth**

**Title:** Laboratory methods in embryology and mesodermal precursor/stem cell biology. *In ovo* electroporation.

**Final evaluation (Dr. Susanne Dietrich):** Lorenzo very quickly acquired the background to plan and carry out experiments independently. He acquired the dexterity to perform delicate manipulations of embryos including microdissections and *in ovo* electroporation, and he acquired the practical skills to carry out basic methods in molecular biology, microbiology, in situ hybridisation, immunofluorescence and photo-microscopy. He made excellent progress throughout, and was able to work independently and unsupervised. He very much contributed to progress in the lab, and it was a pleasure to work with him.

September 2019

**Diploma Nazionale SIUMB di competenza in Ecografia Clinica**

February 2019

**Abilitazione all'esercizio della Professione di Medico Chirurgo** (License to Practice Medicine) *Italian Ministry of Education, University & Research.*

**Final evaluation:** 270/270

License to Practice was awarded after a three-months internship (held from November, 2018 and January, 2019) and a theoretical test in February 2019.

October 2012 -  
October 2018

**Degree in Medicine and Surgery, Università di Pisa**

Graduation grade: 110/110 cum laude and publishing honors.

**Final Dissertation:** “PD-L1 and IFN- $\gamma$  exosomal mRNA expression in patients with non small cell lung cancer: correlation with clinical response with anti-PD-1 antibodies.”

July 2018

**Course: Gene expression data analysis using R: How to make sense out of your RNA-Seq/microarray data, Erasmus MC, University Medical Center Rotterdam**

Advanced statistical tools in order to analyze gene expression, especially using RNA microarray data.

July 2016 -  
September 2016

**Erasmus Traineeship**, *Sexual Health at The Hatersage Center, Central Manchester University Hospital*

Experience in management of HIV and sexually transmitted infections. The programme included: attending regular sexual health clinics; Attending regular HIV clinics; attending HIV ward rounds; performing a Service Review on HIV management; performing a results review on Tenofovir Alafenamide; giving a presentation on the departmental teaching programme; learning the role of HIV specialist nurses and health advisers Knowledge

**Final evaluation: (Dr. Gabriel Schembri)** Lorenzo has been an exemplary trainee who got to grips with the systems we use to review and treat patients very quickly, and also how we manage people with sexually transmitted infections, including HIV. He was enthusiastic and keen to improve his medical knowledge. His presentation on sexual health services in Italy was amongst the most entertaining presentations I've ever heard. He was well liked by the staff here and integrated well within the department.

1-14 July 2015

**Giovanni Mazzotti Italian-American Conference on Human Anatomy, D'Youville College, Buffalo:** Dissection of the human body.

**Final evaluation:** 3<sup>rd</sup> classified in the final test; Laude for the expertise in the dissection.

## LAB EXPERIENCES

May 2023 -  
September 2023

**Institute for Quantitative Health Science & Engineering**, *Michigan State University (USA)*  
Experience in handling and manipulating human induced pluripotent stem cells and in generation and analysis of cardiac organoids by means of confocal microscopy and quantitative PCR.

October 2020 -  
ongoing

**Physiology Department**, *University of Pisa*

Experience in neural and cardiac correlates of hypnosis. Training in advanced EEG analysis, heart-rate variability analysis.

May 2019 & March  
2022

**Anatomy Tutor in Cadaver Lab, Dissection Course**, *University of Pisa*

February 2019 -  
May 2019

**School of Pharmacy and Biomedical Sciences**, *University of Portsmouth*

Experience in skeletal and cardiac development and the associated progenitor/stem cells. Training in molecular, cell biological and embryological, procedures including embryo culture and micromanipulation, generation and transduction of molecular constructs, in situ hybridisation, immunohistochemistry, various microscopical techniques.

September 2017 -  
present

**Farmacogenetics, U.O. Farmacogenetica, *University of Pisa***

I started my project for my final dissertation with Professor R. Danesi to determine whether concentration of neoplastic RNA in circulating exosomes extracted from peripheral blood correlates with response to immunotherapy treatment.

July 2016 -  
September 2016

**The Hatersage Center, *Central Manchester University Hospital*** Diagnosis of the most common agents of sexual transmitted diseases including dark field microscopy.

## CERTIFICATIONS

March 2021

**Advanced Cardiac Life Support (ACLS) Provider, *American Heart Association***

July 2016

**Cambridge English: Advanced (CAE) – CEFR level C1**  
**Final Evaluation:** Grade B

September 2009

**European Computer Driving Licence (ECDL) – Syllabus 4.0**

## RESEARCH ARTICLES AND REVIEWS

**Clinical reasoning: When the eyes flutter - an acute case of oscillopsia** Lorenzo Fontanelli, Eleonora Del Prete, Gianmichele Migaleddu, Francesca Bianchi, Gabriele Bellini, Lucrezia Becattini, Nicola Giannini, Mirco Cosottini, Gabriele Siciliano (*under review*)

**Cardiac comorbidities in McArdle disease: case report and systematic review** D Hoxhaj, G Vadi, L Bianchi, Lorenzo Fontanelli, F Torri, G Siciliano, G Ricci, Neurological Sciences (2024)

**Prokineticin-2 Is Highly Expressed in Colonic Mucosa of Early Parkinson's Disease Patients** Gabriele Bellini, Francesco Rettura, Giovanni Palermo, Chiara Ippolito, Cristina Segnani, Clarissa Pierucci, Lorenzo Fontanelli, Daniela Frosini, Vincenzo Nardini, Christian Lambiase, Nunzia Bernardini, Carolina Pellegrini, Roberto Ceravolo Movement Disorders (2024)

**Is hypnotic induction necessary to experience hypnosis and responsible for changes in brain activity?** Alejandro Callara, Zan Zelic, Lorenzo Fontanelli, Alberto Greco, Enrica Laura Santarcangelo, Laura Sebastiani, (2023)

**Pre-existing frontal lobe dysfunction signs as predictors of subsequent neurotoxicity in CAR T cell therapy: insights from a case series** Lorenzo Fontanelli, Chiara Pizzanelli, Chiara Milano, Raffaella Cassano Cassano, Sara Galimberti, Maria Ida Rossini, Ignazio Santo, Francesco Turco, Enrica Bonanni, Gabriele Siciliano, Enrico Orciuolo, Filippo Baldacci Neurological Sciences (2023)

**Modulation of the heartbeat evoked cortical potential by hypnotizability and hypnosis** Alejandro Luis Callara, [Lorenzo Fontanelli](#), Iacopo Belcari, Gianluca Rho, Alberto Greco, Žan Zelič, Laura Sebastiani, Enrica L. Santarcangelo *Psychophysiology* (2023)

**Distinct limbic connectivity left and right benign mesial temporal lobe epilepsy: evidence from a resting state functional MRI study** C. Pizzanelli, I. Pesaresi, C. Milano, P. Cecchi, [L. Fontanelli](#), S. Giannoni, F. S. Giorgi, M. Cosottini, E. Bonanni *Frontiers in Neurology* (2022)

**Fatigue as a common signature of inflammatory myopathies: clinical aspects and care** G Ricci, [L. Fontanelli](#), F Torri, E Schirinzi, G Siciliano *Clinical and Experimental Rheumatology* (2022)

**Is hypnotic assessment relevant to neurology?** [Lorenzo Fontanelli](#), Vincenzo Spina, Carmelo Chisari, Gabriele Siciliano, Enrica L Santarcangelo, *Neurological Sciences* (2022)

**Frailties and critical issues in neuromuscular diseases highlighted by SARS-CoV-2 pandemic: how many patients are still “invisible”?** Giulia Ricci, Francesca Torri, Francesca Bianchi, [Lorenzo Fontanelli](#), Erika Schirinzi, Elisa Gualdani, Paolo Francesconi, Delia Gagliardi, Gigliola Serra, Tiziana Mongini, Gabriele Siciliano, *Acta Myologica* (2022)

**Gemcitabine Plus Nab-Paclitaxel Induces PD-L1 mRNA Expression in Plasma-Derived Microvesicles in Pancreatic cancer** Marzia Del Re, Caterina Vivaldi, Eleonora Rofi, Francesca Salani, Stefania Crucitta, Silvia Catanese, [Lorenzo Fontanelli](#), Valentina Massa, Federico Cucchiara, Lorenzo Fornaro, Annalisa Capuano, Stefano Fogli, Enrico Vasile, Romano Danesi, *Cancers* (2021)

**A multiparametric approach to improve the prediction of response to immunotherapy in patients with metastatic NSCLC** Marzia Del Re, Federico Cucchiara, Eleonora Rofi, [Lorenzo Fontanelli](#), Iacopo Petrini, Nicole Gri, Giulia Pasquini, Mimma Rizzo, Michela Gabelloni, Lorenzo Belluomini, Stefania Crucitta, Raffaele Ciampi, Antonio Frassoldati, Emanuele Neri, Camillo Porta, Romano Danesi, *Cancer Immunology, Immunotherapy* (2021)

**Clinical pharmacology of monoclonal antibodies targeting anti PD-1 axis in urothelial cancers** Eleonora Rofi, Marzia Del Re, Elena Arrigoni, Mimma Rizzo, [Lorenzo Fontanelli](#), Stefania Crucitta, Giulia Gianfilippo, Giuliana Restante, Stefano Fogli, Camillo Porta, Romano Danesi, Manuela Schmidinger, *Critical reviews in oncology/hematology* (2020)

**CYP17A1 polymorphism c.-362T> C predicts clinical outcome in metastatic castration-resistance prostate cancer patients treated with abiraterone** Stefania Crucitta, Marzia Del Re, Federico Paolieri, Francesco Bloise, Andrea Sbrana, Enrico Sammarco, Chiara Mercinelli, Federico Cucchiara, [Lorenzo Fontanelli](#), Luca Galli, Romano Danesi, *Cancer Chemotherapy and Pharmacology* (2020)

**Endothelial nitric oxide synthase c.-813C> T predicts for proteinuria in metastatic breast cancer patients treated with bevacizumab-based chemotherapy** Stefania Crucitta, Giuliana Restante, Marzia Del Re, Ilaria Bertolini, Eleonora Bona, Eleonora Rofi, [Lorenzo Fontanelli](#), Giulia Gianfilippo, Stefano Fogli, Irene Stasi, Matteo Ghilli, Andrea Fontana, Romano Danesi, *Cancer Chemotherapy and Pharmacology* (2019)

## LETTERS

**Should we prescribe approved drugs to unproven cohorts of patients? The case of disease-modifying drugs for Spinal Muscular Atrophy** [Lorenzo Fontanelli](#), Giulia Di Rauso, Gabriele Bellini, *European Journal of Neurology* (2023)

**Intravenous immunoglobulin in the treatment of sporadic inclusion body myositis: time for new evidence?** Lorenzo Fontanelli, Gabriele Vadi, Erika Schirinzi, Giulia Di Rauso, Gabriele Bellini, Giulia Ricci, Gabriele Siciliano Clinical and Experimental Rheumatology (2024)

## PEER REVIEWER EXPERIENCE

2023 - Ongoing                      **Peer reviewer for “Frontiers in Neuroimaging”**

March 2023                      **Abstract reviewer** of the Abstract Submissions for the 9th EAN Congress 2023

## POSTER AND ORAL PRESENTATIONS

**Quantitative voice analysis in Patients with Myotonic Dystrophy type 1** L. Fontanelli, G. Ricci, G. Siciliano (2023)

**Effect of hypnotizability and hypnosis on the heartbeat evoked cortical potential and heart rate variability**, Lorenzo Fontanelli, Alejandro Callara, Zan ZelicAlberto Greco, Enrica Laura Santarcangelo, Laura Sebastiani (2023)

**Frontal lobe involvement in patients treated with CAR T-cells developing ICANS** Lorenzo Fontanelli, Chiara Pizzanelli, Chiara Milano, Raffaella Cassano Cassano, Sara Galimberti, Maria Ida Rossini, Ignazio Santo, Francesco Turco, Enrica Bonanni, Gabriele Siciliano, Enrico Orciuolo, Filippo Baldacci (2023)

**Pregnancy in women with Spinal Muscle Atrophy: what mothers and clinicians should know. A case report.** G. Ricci, L. Fontanelli, R. Chiappini, G. Siciliano (2023)

**Baseline subtle neurological signs predict future Immune effector cell-Associated Neurotoxicity Syndrome (ICANS) in patient with Diffuse Large B-Cell Lymphoma (DLBCL) treated with CAR-T therapy tisagenleclucel** L. Fontanelli, C. Milano, C. Pizzanelli, I. Trivelli, M. Cosottini, S. Galimberi, G. Siciliano, E. Orciuolo, F. Baldacci (2022)

**Retrospective study of disease natural history in a large cohort of patient with Myotonic Dystrophy Type 1.** Lorenzo Fontanelli, Martina Ciccarelli, Francesca Torri, Gabriele Vadi, Giulia Ricci, Gabriele Siciliano

**Digitalized assessing of muscle endurance in adult SMA patients with a novel supervised clinical protocol.** Giulia Ricci, Francesca Torri, Roberto Chiappini, Lorenzo Fontanelli, Alessandro Tonacci, Raffaele Conte, Andrea Vannini, Roberto Lazzarini, Stefano Roccella, Gabriele Siciliano

**A patient with Amyotrophic Lateral Sclerosis and Brugada Pattern: a case report** L. Fontanelli, F Bianchi, L. Becattini, P. Sciarrone, G. Siciliano (2022)

**Coexistence of CASQ1-related myopathy and Amyotrophic Lateral Sclerosis: a case description report** L. Fontanelli, F Biachi, L. Becattini, R. Brugnoli, G. Siciliano (2022)

**When rare gets rarer: five reports of association between and other uncommon conditions from a large cohort of patients in central Italy** Francesca Bianchi, Lucrezia Becattini, Lorenzo Fontanelli, Antonella Fogli, Lucia Chico, Gabriele Siciliano

**Clinical characterization of C9orf72-mutated ALS patients in a large center for Motor Neuron Disease in Central Italy** L. Becattini, F. Bianchi, L. Fontanelli, A. Fogli, G. Siciliano (2022)

**A case of an uncommon late onset of ALS associated with an undescribed SOD1 mutation** Lucrezia Becattini, Francesca Bianchi, Lorenzo Fontanelli, Antonella Fogli, Gabriele Siciliano (2022)

**An Italian patient with Amyotrophic Lateral Sclerosis and Myasthenia Gravis: a clue for the dying-back hypothesis** F Bianchi, L Fontanelli, A Galgani, A Bacci, S Gallerini, G Siciliano (2021)

**Two contrasting clinical manifestations of amyotrophic lateral sclerosis (ALS) related to FUS mutations** F Bianchi, L Fontanelli, A Galgani, L Becattini, A Fogli, G Siciliano (2021)

**Symptomatic C9ORF72 mutation in non-fluent aphasia without neuroimaging signs of cerebral atrophy: A case study** Lorenzo Fontanelli, Francesca Bianchi, Alessandro Galgani, Daniela Frosini, Irene Ghicopulos, Antonella Fogli, Gabriele Siciliano (2021)