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CV ENGLISH

Curriculum Vitae Europass

PERSONAL INFORMATION	
Name / Surname	**ALESSANDRA FERLINI**
Personal Address	
Work Address	Unita di Genetica Medica. Università di Ferrara
Phone	Mobile:
Fax	
E-mail	
Citizenhsip	
Date of Birth	
Gender	
Current Position	-Associate Professor, tenured, permanent position, and Director of the Unit of Medical Genetics Unit, University of Ferrara, Italy -Honorary Visiting Professor, Institute of Child Health. University College London, UK

Language Competence *(*) EU frame*	**Italian (Mother tongue)**									
Other languages	English (fluent), German (good)									
selfassessment	**Comprehension**				**Spoken**				**Written**	
Livello europeo (*)	listening		reading		Oral interaction		Oral production			
English	C2	Livello avanzato	C2	Livello avanzato	C2	Livello avanzato	C2	Livello avanzato	C2	Livello avanzato
German	C1	Livello intermedio	B2	Livello intermedio	B1	Livello intermedio	A2	Livello elementare	A2	Livello elementare

EDUCATIONAL AND PROFESSIONAL TITLES

Career titles	
2006-present	-Associate Professor in Medical Genetics MED/03, Università di Ferrara Head of the Medical Genetics Unit, University Hospital Ferrara
2015-present	-Honorary Visiting Professor, University College London, Institute of Child Health
2000 - 2006	-Dirigente Medico di I livello (UO Neurologia, UO Genetica medica) Azienda Ospedaliera Universitario S. Anna, Ferrara
1995 – 1999 (short stages in 2000 and 2001)	-Senior Research Assistant, Neuromuscular Unit, Imperial College of Medicine, Hammersmith Hospital, London (UK)

Education titles	
2002	PhD genetics, Imperial College School of Medicine, London (UK)
1993	Medical Genetics Specialization, University of Ferrara (Italy), 50/50 cum laude
1988	Neurology Specialization, University of Bologna (Italy), 70/70 cum laude
1983	Iscrizione Albo Medici
1984	Abilitazione all'esercizio di Medico Chirurgo
1983	Degree in Medicine and Surgery University of Bologna (Italy), 110/110 cum laude
Participation to Scientific Committees titles	
1996-1997	Chromosome 18 Sequencing Committee (US)
2011-2014	ENMC Scientific Committee, Member
2012-2015	SOCIETA ITALIANA DI GENETICA UMANA SIGU Coordinators of the Molecular Genetics Working Group
2011-2014	IRDIRC WG on Biomarkers for Disease Progression and Therapy Response, Chair
2015-present	TREAT-NMD Ethical Committee-Member
2017-2020	Sarepta Therapeutics European Board
2018-2020	PTC Therapeutics European Board
	Regione Emilia Romagna Institutional Working Groups (member):
2019-present	-Neuromuscular diseases
2018-present	-Movement Disorders and Huntington disease
2020-present	-Birth and Neonatal working group and NIPT
2007-present	-Genetic testing
Ethical Committees	
2007-2011	Member of the Ethical Committee of the EU project NMD-Chip
2013-2015	Member of the Ethical Committee of the Regione Emilia Romagna (Area Vasta Centrale)
2010-2013	Member of the Ethical Committee of the Azienda Ospedaliera Universitaria di Ferrara
2017-oggi	Member of the Ethical Committee of the TREAT-NMD Alliance
Prizes and Honours titles	
2015-present	-Honorary Visiting Professorship, University College London, London (UK)
2011	-Telethon prize, best research project
1995 - 1999	-British Heart Foundation Grant Contract, London (UK)
1994 - 1995	-Muscle Dystrophy Group Contract, London (UK)
1986	-Boncompagni-Ludovisi (Svezia), Prize , innovative project on genome studies
1985	-EMBO (Germany), Prize best published paper on Duchenne muscular dystrophy
1984	-Comitato Sanremo Genetica Umana, prize for the best study on Rett Syndrome
1993-1994	-CNR Researcher fellowship, Nobel Prize Renato Dulbecco Unit , Human Genome - Project, ITBA-CNR, Milano
1992-1993	-Contract Professor Neurogenetics, University of Modena

Competitive research grants titles	
EU research project (FP5, FP6, FP7, H2020)	**Titles , grant agreement numbers , and websites**
2023-ongoing	**Euro-NMD ERN WP4 (EU4H-2023-ERN-IBA) https://ern-euro-nmd.eu**
2022-onogoing	**DYNALIFE** COST Action, Partner (CA21169) https://www.dynalife.eu
2021-ongoing	**SCREEN4CARE** H2020 EU-IMI project Scientific Coordinator, grant agreement n. 101034427; www.screen4care.eu
2019-ongoing	**BIND** , Partner H2020 grant agreement n. 847826, PI www.bindproject.eu
2017-ongoing	**SOLVE-RD**, Partner grant agreement n. 779257, PI www.solve-rd.eu
2011-2014	**EXONSKIPPING** COST Action, (BM1207) Partner http://exonskipping.eu/
2011 - 2014	**NEUROMICS** FP7 (Grant Agreement 311672), PI www.rd-neuromics.eu
2014 - 2018	**SIGN** Network Genetico Sloveno-Italiano, PI www.signgenetics.eu/
2009 - 2012	**BIO-NMD** FP7 (grant Agreement 241665), Scientific Coordinator www.bio-nmd.eu
2007-2011	**NMD-CHIP** FP6 (Grant agreement 223026), PI www.nmd-chip.eu
1999 - 2002	**FINGER** FP5 (Grant agreement 00920) Scientific Coordinator http://cordis.europa.eu/result/rcn/82133_en.html
Grants Telethon Italy Foundation	
2021 2021 2009 2008 2010 2007 2010 1993 1994	1. N.3-2021, Telethon : Storia naturale della distrofia muscolare di Becker: in preparazione di futuri studi clinici (2021-2025) (456.000 €) Partner 2. N. 2-2021, Telethon: Distrofia muscolare di Duchenne da duplicazione dell'esone 2: caratterizzazione e identificazione di fattori predittivi e prognostici (2021-2024) (196000 €) Partner 3. GGP09093 (2009) Coordinator: 515.700 € 4. GGP08107 (2008) Partner: 884.431 € 5. GUP07011 (2010) Coordinator: 71.700 € 6. GGP05115 (2007) Coordinator: 204.000 € 7. GGP02311 (2010) Partner: 102.200 € 8. N. 1054 (1993) Coordinator: 51.646 € 9. N. 146 (1994) Partner: 15.494 €

Others Grants	
2012-2015	1. Regione Emilia-Romagna: 'RARER: Next generation sequencing and gene therapy to diagnose and cure rare diseases in the Emilia Romagna', full partner
2009-2011	2. PRIN MIUR ITALIA , PI "Molecular bases of CollagenVI myopathies"
2008-2011	3. Regione Emilia-Romagna: 'The contribution of novel technologies to the improvement of diagnosis/therapies in Medical Genetics of breast cancer', Coordinator
2008-2013	4. DUCHENNE PARENT PROJECT ONLUS ITALIA PI (4 grants),
2015-2022	5. DUCHENNE PARENT PROJECT ONLUS ITALIA Urinary Stem Cell Biobank
2018-2022	6. PTC Therapeutics diagnostic service Grant (2018-2022) Duchenne muscular dystrophy diagnosis for patients from Eastern Europe and North African countries.
2016-2018	7. Association Francoise pour le myopathies (AFM) France, grant Full Partner; The Popeye domain containing gene 1 and its role in muscular dystrophy
2019-present	8. Sarepta Therapeutics Grants (2) to accelerate genetic diagnosis of neuromuscular diseases
2024-present	9. PNRR-MR1-2023-12377278 "Design, setting, and validation of a multiregional, genetic newborn screening study for multiple rare diseases to piloting a nationwide approach using a dual digital and genomics strategy".

	INSTITUTIONAL ACTIVITIES
Public Assignments: synopsis	
	Since 2007, AF has been **Director of the Complex Operative** Unit of Medical Genetics, at the University Hospital of Ferrara, in the role of confirmed Associate Professor, with the role of Head Physician. In this role she directs both the clinical and laboratory activities related to genetic counseling and genetic diagnosis of inherited pathologies (Unit website https://www.ospfe.it/reparti/genetica-medica). In her role as **Associate Professor of the University of Ferrara** she carried out/carries out intense teaching and training activities for students of Medicine, Biotechnology, Motor Sciences, she directed the **School of Specialization in Medical Genetics** from 2007 to 2019, training both doctors and specialist biologists. She has participated in numerous institutional working tables and commissions for the design of therapeutic-assistance paths. Since 2015 is **Honorary Visiting Professor at the University College London,** involved in research projects and scientific publications mainly on Duchenne muscular dystrophy genetic bases and therapeutical approaches.
Teaching activity	
2006--2010	23179- DIAGNOSI GENETICA CDL BIOTECNOLOGIE, UNIFE
2008-2010	015-107 GENETICA E SPORT CDL SCIENZE MOTORIE, UNIFE
2008-2010	015440 BIOLOGIA, GENETICA, MICROBIOLOGIA CDL FISIOTERAPIA, UNIFE
2008-2013	020138 GENETICA MEDICA CDL MEDICINA E CHIRURGIA, UNIFE
2009-2013	26848 GENETICA UMANA CDL MEDICINA E CHIRURGIA, UNIFE
2011-2013	020139 GENETICA MEDICA CDL MEDICINA E CHIRURGIA, UNIFE
2014-2019	015178 GENETICA MEDICA CDL MEDICINA E CHIRURGIA, UNIFE
2015-2019	58778 GENETICA DELLE POPOLAZIONI, CDL MEDICINA E CHIRURGIA, UNIFE
2020-present	67636 GENETICA MEDICA, CDL MEDICINA E CHIRURGIA, UNIFE

2021-present	019352 GENETICA MEDICA, CDL SCIENZE MOTORIE, UNIFE
2007-2018	Docente Collegio Dottorato di Ricerca Medicina Sperimentale, UNIFE
2007-2019	Director of the School of Specialization in Medical Genetics of the University of Ferrara, with educational duties for Medical Doctors and for Biologists.
2019-present	Docente Collegio Dottorato di Ricerca Scienze omiche, oncologiche e chirurgiche, UNIFE
1992-Present	Teaching activities at more than 50 national and international courses about genetic bases of Rare Diseases
Scientific Society memberships	
2013-present 2008-present 2008-present 2006-2012 2004-present 2002-present 1999 – 2006 1999-present 1996-present	Editorial Board 'Nanoletters' Member 'American Society of Gene and Cell Theraphy Editorial Board 'Neuromuscular Disorders' journal Ethical Committee Treat-NMD Member Associazione Italiana di Miologia Member 'World Muscle Society (WMS)' Member ISS Genetic test quality control scheme (Istituto Superiore di Sanità) Member American Society of Human Genetics (AMS) Member Società Italiana di Genetica Umana (SIGU)
Patents' applications (third Mission)	
	1. 2012, EURO PCT N. 10771208 Nanoparticles to deliver antisense oligonucleotides 2. 2014 TO2008A000496 CHG array to detect HLA locus copy number variations. 3. 2022 BI566R Codon usage calculation and clustering by algorithms in rare disease genes
Reviewer activities	
Scientific Journals (articles) Active reviewing for the following scientific Journals:	Main: Amyloid, Annals of Neurology, Annals of Translational Medicine, BBA, BBRC, BMC journals, Brain, Curr. Opin. Orph. Drug., Cell series, Cells, European Journal of Neurology, European Journal of Human Genetics, EMBO Mol Med, Frontiers in Genetics, Frontiers in Neurology, Gene, Human Molecular Genetics, Human Genetics, Human Gene Therapy, Human Mutation, Journal of Cachexia, Sarcopenia and Muscle, J Cell Biochemistry, Journal of Medical Genetics, Journal of Neurodiagnostics, Journal of Neuromuscular Diseases, Journal of Gene Medicine, Journal of Neuropathology and Experimental Neurology, Molecular Genetics and Metabolism, Molecular Neurobiology, Molecular Therapy, Molecular Medicine, Muscle & Nerve, Nanomedicine and Nanobiotechnology, Nature Genetics, Neurological Sciences, Nucleic Acid Therapy, Nature Comm, Nature Reviews, Neurogenetics, Neuromuscular Disorders, Oligonucleotides, Orphanet J., Pharmacogenomics, Plos One, Rare Disease and Orphan Drugs Journal, RNA, Science, Scientific Reports, Skeletal Muscle, Trends in Genetics, World J Cardiology, sporadically others.
Research Funding Agencies (project applications)	
	European Research Council (ERC); EU FP6, FP7, H2020 evaluator, Association Francoise Myopathies (AFM), Duchenne Parent Project Netherland Foundation;

	Muscular Dystrophy UK, Princess Beatrix Foundation The Netherland, Rose Trees Trust Foundation, EU ERN evaluator.
European Reference Networks Membership (Hospital HCP representative)	
	ERN Euro-NMD HCP representative (2017-present) www.ern.euro-nmd.eu ERN ITHACA, HCP representative (2020-present) www.ern.ithaca.eu ERN EuroBloodNet, HCP affiliated (2021-present) https://eurobloodnet.eu
International Scientific Networks/Agencies official participation	
2021-present	TREAT-NMD and Alliance www.treatnmd.eu , Affiliated Partner Ethical Committee Member
2022-present	ICONS (www.icons.org) International consortium for Newborn sequencing, Executive Committee member
2021-present	External expert for rare diseases and orphan drugs at European Medicines Agency (EMA)
2013 & 2020	Organizers of 2 ENMC international workshops in the field of neuromuscular diseases (www.enmc.org)
2012-2017	Chair of the Rare Diseases Partnership within the International Scientific Technology Programme (ISTIP) with Gulf Council Countries (GCC), facilitating research and education by networking with colleagues in Saudi Arabia, United Arab Emirates, Oman, Kuwait, Qatar.
2023-today	Consultant for the Saudi Genome Program 2.0 , to support in setting and detailing the strategic direction of the Program, validation of governance structure, operating models partnerships and funding models.
2023-today	Member of the Pfizer Data Monitoring Committee for Duchenne muscular dystrophy Gene Therapy
Principal Investigator in Clinical Trials based on orphan drugs titles	Since 2010 AF acts as Principal Investigator of clinical trials based on antisense oligoribonucleotide in DMD. During this more than 10 year-experience, AF has maturated an in-depth skill in DMD exon skipping based therapies, as documented not only by trials participation but also by being member of company advisory Boards and as an invited speaker in symposium and conferences.
	ClinicalTrials.gov Identifiers: 1. NCT02329769 (Open Label, Extension Study of PRO044 in Duchenne Muscular Dystrophy (DMD)) 2. NCT01037309

	(Phase I/II Study of PRO044 in Duchenne Muscular Dystrophy (DMD) 3. NCT02958202 Extension Study of BMN 044 in Duchenne Muscular Dystrophy (DMD) 4. NCT01480245 (Open Label Study of GSK2402968 in Subjects With Duchenne Muscular Dystrophy) 5. NCT01254019 (A Clinical Study to Assess the Efficacy and Safety of GSK2402968 in Subjects With Duchenne Muscular Dystrophy (DMD114044) 6. NCT02500381 Study of SRP-4045 (Casimersen) and SRP-4053 (Golodirsen) in Participants with Duchenne Muscular Dystrophy (DMD) (Essence)

	Organizational and Managing experience
Overview	AF has **significant managerial experience** in the direction and completely autonomous management of human resources, programmes, personnel, and funding/budgets, as documented by the results achieved: documented diagnostic excellence (as per official, Regional, Ministerial, and European recognition), and research, with the acquisition of numerous public and private funding. As it is often typical of Medical Genetics, and also thanks to the clinical competence due to the Specialization in Neurology and to the Scientific competence, AF has almost always directed multidisciplinary initiatives by coordinating different professional figures belonging to different Institutions. AF has worked intensively with numerous **Rare Disease Patient Associations** both nationally and internationally (for example the World Duchenne Parent Project and Eurordis), developing a very fruitful interaction capacity based on mutual trust, transparency, and a pragmatic tendency to obtain results. She has been working with great harmony, synergy, mutual esteem with numerous **Pharmaceutical Companies** (Pfizer, Roche, Sarepta, etc) for years, also thanks to her role as PI in clinical trials, and her expertise in medical genetics and she has actively participated in the design of the first clinical trials with orphan drugs (particularly antisense molecules) for muscular dystrophies.
Registries and Biobank	AF has vast and long-lasting experience in the **management and coordination of institutional working groups**, including international ones, dedicated to scientific and diagnostic activities, the drafting of guidelines, and the design of diagnostic-therapeutic pathways. The direct experience in **rare disease registries** is mainly linked to the function of the Medical Genetics Operating Unit, which she directs, as the Molecular and Clinical Hub of the Emilia Romagna Region. He is therefore the contact person for Rare Diseases and supplies data to the regional registry. Furthermore, in all the EU projects in which he participated or which he coordinated, disease registries were developed, registries related to specific evaluation measures (genetic, phenotypic, mutation, biomarker registries), of which he then coordinated the design and Development. AF is responsible for the **urinary stem cell (USC) biobank**, within a project funded by the Duchenne Parent Project, Italy.
EU funding application and supervision experience	Managing experience in designing and submitting multidisciplinary EU applications with very large partnerships (rate of success 66%: submitted 15, funded 10),

	organizational experience with EU managing agencies (many) to monitor both operational and scientific project activities.
Managing experiences	
1999-present	**Coordinator and manager of 3 EU research projects** (FP5, FP7, H2020, coordinating large international partnerships with more than 50 partners in 20 European countries and with USA affiliated partners, including Global companies and Small Medium Enterprises (SME). Vast experience in managing and delivering administrative and scientific reports.
2006-present	**Director of the Medical Genetics Unit,** coordinating 30 people, including nurses, technicians, MD, PhD, and administrative persons, competence in designing and managing budget, operational flowcharts, and health diagnostic programmes.
2007-2019	**Director of the School of Specialization in Medical Genetics** of the University of Ferrara, with educational duties for Medical Doctors and for Biologists to become medical geneticists. In this role she has taught and specialized about 60 young colleagues.
2008-2011	**Coordinator of the Regional Research Health Policy project by Regione Emilia-Romagna**: 'The contribution of novel technologies to the improvement of diagnosis/therapies of breast cancer', in charge of setting of NGS methods to rapid diagnosis hereditary breast cancer
2010-present	**Principal Investigator in multicentric, international clinical trials** for Muscular Dystrophy, managing a complex and multidisciplinary team in charge of trial activities (administrative, nurses, researchers, clinicians) as well as networking with other trial centers worldwide
2011-2014	**Chair of IRDIRC WG on Biomarkers** for Disease Progression and Therapy Response, coordinating more than 50 research Centers from 18 countries, including EU and USA
2012-2015	**Chair of the Rare Diseases Partnership within the International Scientific Technology Programme (ISTIP-ICONeT EU action)** with Gulf Council Countries (GCC), with the role of networking with colleagues in Saudi Arabia, United Arab Emirates, Oman, Kuwait, Qatar. She is still working with Saudi Arabia and Qatar colleagues for strategic and research collaborations.
2012-2015	**Coordinator of the SIGU Working Group on Molecular Genetics**, composed of 65 members, delivered documents and guidelines related to molecular genetics diagnostics pipelines

Congress and Conference Participation	
1992-present	Invited speaker for lectures, scientific data, talks, original abstracts or communications, and session chair, at **more than 200 events worldwide**

Narrative CV

Alessandra Ferlini

She graduated in medicine and surgery at the University of Bologna in 1983 under the supervision of Professor Giovanni Romeo, at that time teacher at the Bologna University, with a thesis about the genetic and enzymatic bases of Schizophrenia. Despite of her deep love and interest in Genetics, following the suggestion of Prof. Romeo, she was admitted to the School of Specialization in Neurology at the University of Bologna. During those years she worked at the Genetic Laboratory directed by Prof. Romeo, being dedicated to the setting of the new reverse genetics approach-based techniques, as well started the medical experience as genetic counsellor for neurological diseases.
During those four years, to get an economical sustaining but also to gain MD experience, she also worked as general practitioner in small villages close to Bologna, as well as in emergency medical Services in various Bologna Hospitals where she got a very important experience as medical doctor.
Since of the good musical knowledge and being talented in music, as piano player, and contralto opera singer, she worked as musician and opera, operetta, and baroque camera orchestra singer in several events and concerts in Italy and Europe, also achieving the Diploma in Choir Singer at the Bologna Opera Theater (1989).
She specialized in Neurology (July 1988) with a thesis on molecular basis of Rett Syndrome. In 1988, October, she moved to the University of Modena to work with Prof. Antonino Forabosco, being him recently granted by the Human Genome Project, Italian task (X chromosome, Xq arm). Till 1992 she enjoyed working in that team very much, and she got the first Research Contract at the University of Modena. However, the most important experience she had was the relationship with the Modena site of UILDM (Unione Italiana Lotta alla Distrofia Muscolare). The contacts with patients, families, the intense experience of genetic counselling, the very first prenatal testing for Duchenne muscular dystrophy pioneeringly performed have deeply and transformative impacted her education, formation, and research attitude. During the Modena period, she was admitted at the Medical Genetics School of specialization at the University of Ferrara (1989) where she has completed her medical geneticist education. Not without sorrow and some distress, she was forced to leave the Modena University in July 1993.
In September 1993 she won a CNR fellowship to work in the team of Dr Paolo Vezzoni, at the CNR In Milan, and being part of the Prof. Dulbecco group working in the Human Genome project. This quasi-2-year experience was profoundly important to increase her skill in molecular lab techniques and to realize the importance of the international nature of research, fact that has shaped her entire activity in the next years. And indeed, following that experience, she decided to look for appointments abroad. Very fortunately, she met Francesco Muntoni at the International Conference of Neuromuscular Diseases in Kyoto, in 1994. Following applications for UK positions she got a Muscular Dystrophy Group contract, followed by a British Heart Foundation senior research contract, and in March 1995 she started her appointment at the Neuromuscular Unit, Hammersmith Hospital, London, under the supervision of Prof. Francesco Muntoni. She has remained there till July 1999, and then for other two shorter stages in 2000 (2 months), and 2001 (3 months), to complete her PhD in Genetics at the Imperial College School of Medicine in 2022. Her VIVA questioners were Prof. Kay Davies and Prof Terry Partridge.
The London years were outstanding and fundamental for her formation as MD and researcher, she sharpened a way of thinking and research attitudes which she then always tried to tirelessly pursue in the following years. Her relationships with her colleagues in London, with her supervisor Francesco Muntoni, have left an indelible mark on her work model and on her life view.
In 1999 she decided to return to Italy since she won a MD permanent position in Neurology at the St Anna University Hospital in Ferrara, thanks to the kind and affectionate encouraging of Dr. Domenico De Grandis. In 2002 she moved to the Medical Genetics Unit, and in 2006 she became Associate Professor and Director of the Unit of Medical Genetics, her current affiliation.
The first Italian years (1999-2006) were deeply committed to the diagnostic activity both in laboratory and in the clinics, with the aim to design and consolidate a robust diagnostic Unit; she was especially dedicated to the setting up of the emerging, powerful molecular genetics technologies for rare disease diagnosis. In those years she was therefore not able to dedicate so much time to research. Situation has improved in 2007, where she was awarded with EU, Telethon and other grants, which

have allowed a restarting of her research activities, still ongoing today, being a fundamental part of her work.

Research Activities in a few words

For many years already her special interest and major research effort is concentrated on dystrophinopathies and hereditary neuromuscular pathologies. Aim of the conducted research is to identify the processes which determine the regulation of expression of this specific gene, particularly referring to the process of splicing and its modulation. Her research team is studying in vitro and in vivo systems to modulate mutations of the gene with antisense oligonucleotides while using new systems of release (nanomaterials). Furthermore, the team is in charge with the definition of the RNA profile of various hereditary pathologies and the identification of molecular markers both in humans and in animal models. She is also studying new therapeutic approaches for dystrophinopathies and other neuromuscular pathologies as well as innovative aspects of molecular diagnostics with focus on high throughput techniques and next generation sequencing. She is provided with all ethical aspects referring to medical genetics and the therapy of genetic diseases.

Personal Interests and Hobbies

Her personal interests are characterized by a great passion for music, sports, pets, agriculture, and nature in general, [illegible]
she has excellent relationship with many colleagues worldwide.

CV ITALIANO

Curriculum Vitae Europass

INFROMAZIONI PERSONALI	
Nome/Cognome	**ALESSANDRA FERLINI**
Indirizzo privato di residenza	
Indirizzo di Lavoro	nita di Genetica Medica, Università di Ferrara
Telefono	Cellulare:
Fax	
E-mail	
Cittadinanza	Italiana
Data di nascita	
genere	
Posizione attuale	-Professore Associato confermato di ruolo e Direttore della Unità di Genetica Medica, Università di Ferrara -Professore Ospite Onorario, University College London, Institute of Child Health

Competenze linguistiche *(*) EU frame*	**Italiano madrelingua**									
Altre lingue	Inglese (Fluente), Tedesco (buono)									
autovalutazione	**Comprensione**				**Parlato**				**Scritto**	
Livello europeo (*)	listening		reading		Oral interaction		Oral production			
Inglese	C2	Livello avanzato	C2	Livello avanzato	C2	Livello avanzato	C2	Livello avanzato	C2	Livello avanzato
Tedesco	C1	Livello avanzato	B2	Livello intermedio	B1	Livello intermedio	A2	Livello elementare	A2	Livello elementare

TITOLI FORMATIVI E PROFESSIONALI

Titoli di carriera	
2006-oggi	Professore associato e Direttore della Unità Operativa Complessa di Genetica Medica Università di Ferrara
2015-oggi	Professore Ospite Onorario, University College London, Institute of Child Health
2000 - 2006	Dirigente Medico di I livello (UO Neurologia, UO Genetica medica) Azienda Ospedaliera Universitario S. Anna, Ferrara
1995 – 1999 (altri periodi in 2000 and 2001)	Senior Research Assistant, Neuromuscular Unit, Imperial College of Medicine, Hammersmith Hospital, London (UK)
Titoli di Istruzione	

2002	PhD in Genetica , Imperial College School of Medicine, London (UK)
1993	Specializzazione in Genetica Medica, Università di Ferrara (Italia), 50/50 cum laude
1988	Specializzazione in Neurologia, Università di Bologna (Italia), 70/70 cum laude
1983	Iscrizione Albo dei Medici Chrurghi
1984	Abilitazione all'esercizio di Medico Chirurgo
1983	Laurea in Medicina e Chirurgia Università di Bologna (Italia), 110/110 cum laude
Titoli di Partecipazione a Comitati Scientifici	
1996-1997	Chromosome 18 Sequencing Committee (US)
2011-2014	ENMC Scientific Committee, Member
2012-2015	SOCIETA ITALIANA DI GENETICA UMANA SIGU Coordinatore del Gruppo di Lavoro Genetica Molecolare
2011-2014	IRDIRC WG on Biomarkers for Disease Progression and Therapy Response, Chair
2017-2020	Sarepta Therapeutics European Board
2018-2020	PTC Therapeutics European Board
	Regione Emilia Romagna Gruppi di lavoro Istituzionali:
2019-oggi	-Malattie neuromuscolari
2018-oggi	-Disordini del movimento e Corea di Huntington
2020-oggi	-Tavolo Nascita e NIPT
2007-oggi	-Test Genetici
Comitati etici	
2007-2011	Membro del Comitato Etico del Progetto Europeo NMD-Chip
2013-2015	Membro del Comitato Etico Unico Area Vasta Centrale Regione Emilia Romagna
2010-2013	Membro del Comitato Etico dell'Azienda Ospedaliera Universitaria di Ferrara
2017-oggi	Membro del TREAT-NMD Alliance Ethical Committee
Titoli di Premi e Riconoscimenti	
May 2015-oggi	Honorary Visiting Professorship, University College London, London (UK)
2011	Premio Telethon per il miglior Progetto
1995 - 1999	British Heart Foundation Contratto di ricerca, London (UK)
1994 - 1995	Muscle Dystrophy Group Contratto di Ricerca, London (UK)
1986	Boncompagni-Ludovisi (Svezia), premio per il miglior progetto di studi genomici
1985	EMBO (Germany), Premio per il miglior articolo pubblicato sulla Distrofia Muscolare di Duchenne
1984	Comitato Sanremo Genetica Umana, premio per il miglior studio sulla Sindrome di Rett
1993-1994	Ricercatore CNR (contratto annuale), presso la Unita di Ricerca del Premio Nobel Renato Dulbecco, Human Genome Project, ITBA-CNR, Milano
1992-1993	Professore a Contratto in Neurogenetica, University of Modena, Italia

Titoli di Finanziamenti di ricerca con bandi competitivi	
Finanziamenti della Comunità Europea (FP5, FP6, FP7, H2020)	**Titoli, numero del contratto, e siti web**
2023-in corso	**Euro-NMD ERN WP4 (EU4H-2023-ERN-IBA)** https://ern-euro-nmd.eu
2022-in corso	**DYNALIFE** COST action, Partner (CA21169) https://www.dynalife.eu
2021-in corso	**SCREEN4CARE** H2020 EU-IMI project Scientific Coordinator, grant agreement n. 101034427; www.screen4care.eu
2019-in corso	**BIND** , Partner H2020 grant agreement n. 847826, PI www.bindproject.eu
2017–in corso	**SOLVE-RD**, Partner grant agreement n. 779257, PI www.solve-rd.eu
2011-2014	**EXONSKIPPING** COST Action, (BM1207) Partner http://exonskipping.eu/
2011 - 2014	**NEUROMICS** FP7 (Grant Agreement 311672), PI www.rd-neuromics.eu
2014 - 2018	**SIGN** Network Genetico Sloveno-Italiano, PI www.signgenetics.eu/
2009 - 2012	**BIO-NMD** FP7 (grant Agreement 241665), Scientific Coordinator www.bio-nmd.eu
2007-2011	**NMD-CHIP** FP6 (Grant agreement 223026), PI www.nmd-chip.eu
1999 - 2002	**FINGER** FP5 (Grant agreement 00920) Scientific Coordinator http://cordis.europa.eu/result/rcn/82133_en.html
Finanziamenti Telethon Italia	
2021 2021 2009 2008 2010 2007 2010	1. N.3-2021, Telethon : Storia naturale della distrofia muscolare di Becker: in preparazione di futuri studi clinici (2021-2025) (456.000 €) Partner 2. N. 2-2021, Telethon: Distrofia muscolare di Duchenne da duplicazione dell'esone 2: caratterizzazione e identificazione di fattori predittivi e prognostici (2021-2024) (196000 €) Partner 3. GGP09093 (2009) Coordinator: 515.700 € 4. GGP08107 (2008) Partner: 884.431 € 5. GUP07011 (2010) Coordinator: 71.700 € 6. GGP05115 (2007) Coordinator: 204.000 €

1993 1994	7. GGP02311 (2010) Partner: 102.200 € 8. N. 1054 (1993) Coordinator: 51.646 € 9. N. 146 (1994) Partner: 15.494 €
Altri finanziamenti	
2012-2015	10. Regione Emilia-Romagna: 'RARER: Next generation sequencing and gene therapy to diagnose and cure rare diseases in the Emilia Romagna', full partner
2009-2011	11. PRIN MIUR ITALIA , PI "Molecular bases of CollagenVI myopathies"
2008-2011	12. Regione Emilia-Romagna: 'The contribution of novel technologies to the improvement of diagnosis/therapies in Medical Genetics of breast cancer', Coordinator
2008-2013	13. DUCHENNE PARENT PROJECT ONLUS ITALIA PI (4 grants),
2015-2022	14. DUCHENNE PARENT PROJECT ONLUS ITALIA Urinary Stem Cell Biobank
2018-2022	15. PTC Therapeutics service Grant (2018-2022) Duchenne muscular dystrophy diagnosis for patients from Eastern Europe and North African countries.
2016-2018	16. Association Francoise pour le myopathies (AFM) France, grant Full Partner; The popeye domain containing gene 1 and its role in muscular dystrophy
2019-in corso	17. Sarepta Therapeutics Grants (2) to accelerate genetic diagnosis of neuromuscular diseases
2024-in corso	18. PNRR-MR1-2023-12377278 "Design, setting, and validation of a multiregional, genetic newborn screening study for multiple rare diseases to piloting a nationwide approach using a dual digital and genomics strategy".

	ATTIVITA ISTITUZIONALE
Incarichi pubblici: sinossi	
	Dal 2007 AF è **Direttore della Unita Operativa Complessa** di Genetica Medica, presso la Universitaria di Ferrara, nel ruolo di Professore Associato confermato, con ruolo di Primario. In questo ruolo dirige sia le attivita cliniche che quelle laboratoristiche relative alla consulenza genetica e diagnosi genetica delle patologie ereditare (sito web della Unita https://www.ospfe.it/reparti/genetica-medica). Nel suo ruolo di **Professore Associato della Università di Ferrara** ha svolto/svolge intensa attivita didattica e formativa per studenti di Medicina, Biocnologie, Scienze Motorie, ha diretto la **Scuola di Specializzazione in Genetica Medica** dal 2007 al 2019 formando sia medici che biologi specialisti. Ha partecipato a numerosi tavoli e commissioni Aziendali e Regionali per il disegno dei percorsi terapeutici-assistenziali. Dal 2015 è Professore Ospite Onorario presso **University College di Londra (UK),** coinvolto in progetti di ricerca e pubblicazioni scientifiche riguardanti prevalentemente la distrofia muscolare di Duchennne, le sue base genetico-molecolari e i nuovi approcci di terapia a RNA.
Attività didattica	
2006--2010	23179- DIAGNOSI GENETICA CDL BIOTECNOLOGIE, UNIFE
2008-2010	015-107 GENETICA E SPORT CDL SCIENZE MOTORIE, UNIFE
2008-2010	015440 BIOLOGIA, GENETICA, MICROBIOLOGIA CDL FISIOTERAPIA, UNIFE
2008-2013	020138 GENETICA MEDICA CDL MEDICINA E CHIRURGIA, UNIFE
2009-2013	26848 GENETICA UMANA CDL MEDICINA E CHIRURGIA, UNIFE

2011-2013	020139 GENETICA MEDICA CDL MEDICINA E CHIRURGIA, UNIFE
2014-2019	015178 GENETICA MEDICA CDL MEDICINA E CHIRURGIA, UNIFE
2015-2019	58778 GENETICA DELLE POPOLAZIONI, CDL MEDICINA E CHIRURGIA, UNIFE
2020-oggi	67636 GENETICA MEDICA, CDL MEDICINA E CHIRURGIA, UNIFE
2021-oggi	019352 GENETICA MEDICA, CDL SCIENZE MOTORIE, UNIFE
2007-2018	Docente Collegio Dottorato di Ricerca Medicina Sperimentale, UNIFE
2007-2019	Direttore della Scuola di Specializzazione in Genetica Medica, Università di Ferrara, indirizzo Medico e Sanitario (Medici, Biologi, Biotecnologi)
2019-oggi	Docente Collegio Dottorato di Ricerca Scienze omiche, oncologiche e chirurgiche, UNIFE
1992-oggi	Attività didattica in oltre 50 corsi nazionali e internazionali riguardanti le basi genetiche delle le Malattie Rare
Membro di Società Scientifiche	
2013-oggi 2008-oggi 2008-oggi 2006-2012 2004-oggi 2002-oggi 1999 – 2006 1999-oggi 1996-oggi	Editorial Board 'Nanoletters' Member 'American Society of Gene and Cell Theraphy Editorial Board 'Neuromuscular Disorders' journal Ethical Committee Treat-NMD Member Associazione Italiana di Miologia Member 'World Muscle Society (WMS)' Member ISS Genetic test quality control scheme (Istituto Superiore di Sanità) Member American Society of Human Genetics (AMS) Member Società Italiana di Genetica Umana (SIGU)
Domande di Brevetti (Terza Missione)	
	4. 2012, EURO PCT N. 10771208 PMMA Nanoparticles to deliver antisense oligonucleotides (national) 5. 2014 TO2008A000496 CGH array to detect HLA locus copy number variations. (national) 6. 2022 BI566R Codon usage calculation and clustering by algorithms in rare disease genes (international)
Attività di Revisore	
Per **Riviste Scientifiche** (articoli) E' revisore attivo per le seguenti riviste scientifiche:	Main: Amyloid, Annals of Neurology, Annals of Translational Medicine, BBA, BBRC, BMC journals, Brain, Curr. Opin. Orph. Drug., Cell series, Cells, European Journal of Neurology, European Journal of Human Genetics, EMBO Mol Med, Frontiers in Genetics, Frontiers in Neurology, Gene,Human Molecular Genetics, Human Genetics, Human Gene Therapy, Human Mutation, Journal of Cachexia, Sarcopenia and Muscle, J Cell Biochemistry, Journal of Medical Genetics, Journal of Neurodiagnostics, Journal of Neuromuscular Diseases, Journal of Gene Medicine, Journal of Neuropathology and Experimental Neurology, Molecular Genetics and Metabolism, Molecular Neurobiology, Molecular Therapy, Molecular Medicine, Muscle & Nerve, Nanomedicine and Nanobiotechnology, Nature Genetics, Neurological Sciences, Nucleic Acid Therapy, Nature Comm, Nature Reviews, Neurogenetics, Neuromuscular Disorders, Oligonucleotides, Orphanet J., Pharmacogenomics, Plos One, Rare Disease and Orphan Drugs Journal, RNA, Science, Scientific Reports, Skeletal Muscle, Trends in Genetics, World J Cardiology, sporadically others.
Per Agenzie di Ricerca	

(valutazione progetti)	
	European Research Council (ERC); EU FP6, EU FP7, EU H2020 revisore; French Association for the Myopathies (AFM) revisore; Duchenne Parent Project Foundation The Netherland revisore; Muscular Dystrophy Group UK revisore; Princess Beatrix Foundation The Netherland revisore, Rose Trees Trust Foundation revisore; EU ERN valutatore.

European Reference Networks Membership (Rappresentante)	
	ERN Euro-NMD (2017-oggi) HCP rappresentante www.ern.euro-nmd.eu ERN ITHACA, HCP rappresentante (2020-oggi) www.ern.ithaca.eu ERN EuroBloodNet, HCP affiliato (2021-oggi) https://eurobloodnet.eu
Partecipazione istituzionali a Network e Agenzie internazionali	
2021-present	TREAT-NMD and Alliance www.treatnmd.eu , Affiliated Partner Ethical Committee Member
2022-present	ICONS (www.icons.org) International consortium for Newborn sequencing, Executive Committee member
2021-present	External expert for rare diseases and orphan drugs at European Medicines Agency (EMA)
2013 & 2020	Organizers of ENMC international workshops in the field of neuromuscular diseases (www.enmc.org)
2012-2017	Chair of the Rare Diseases Partnership within the International Scientific Technology Programme (ISTIP) with Gulf Council Countries (GCC), facilitating research and education by networking with colleagues in Saudi Arabia, United Arab Emirates, Oman, Kuwait, Qatar
2011-2014	IRDIRC Gruppo di Lavoro Biomarkers for Disease Progression and Therapy Response, Coordinatore
2023-oggi	Consulente scientifico del Saudi Genome Program 2.0 Strategy, per la messa a punto dei dettagli della direzione strategica del Programma , della validazione della struttura di Governance , dei modelli operativi , delle collaborazione e dei modelli di finanziamento.
2023-oggi	Membro del Pfizer Data Monitoring Committee per la terapia genica della distrofia muscolare di Duchenne

Investigatore Principale in Trial Clinici per malattie rare con farmaci orfani	
Dal 2010 AF è stato Investigatore Principale di studi clinici basati su oligoribonucleotidi antisenso nella Distrofia Muscolare di Duchenne. Durante questa decennale esperienza, AF ha maturato un'approfondita competenza nelle terapie basate sullo skipping dell'esone, come documentato non solo dalla partecipazione a sperimentazioni, ma anche dall'essere membro di comitati consultivi aziendali e come relatore invitato a simposi e conferenze.	ClinicalTrials.gov Identifiers: 7. NCT02329769 (Open Label, Extension Study of PRO044 in Duchenne Muscular Dystrophy (DMD)) 8. NCT01037309 (Phase I/II Study of PRO044 in Duchenne Muscular Dystrophy (DMD) 9. NCT02958202 Extension Study of BMN 044 in Duchenne Muscular Dystrophy (DMD) 10. NCT01480245 (Open Label Study of GSK2402968 in Subjects With Duchenne Muscular Dystrophy) 11. NCT01254019 (A Clinical Study to Assess the Efficacy and Safety of GSK2402968 in Subjects With Duchenne Muscular Dystrophy (DMD114044) 12. NCT02500381 Study of SRP-4045 (Casimersen) and SRP-4053 (Golodirsen) in Participants With Duchenne Muscular Dystrophy (DMD) (Essence)

	ESPERIENZA ORGANIZZATIVA E MANAGERIALE
Visione generale	AF ha una **consistente esperienza manageriale** nella gestione diretta e in completa autonomia di risorse umane, programmi, personale e finanziamenti/fondi, come documentato dai risultati conseguiti: documentata eccellenza diagnostica (come da riconoscimento ufficiale, Regionale, Ministeriale, e Europeo), e di ricerca, con acquisizione di numerosi finanziamenti pubblici e privati. Come è spesso proprio della Genetica Medica, e anche grazie alla competenza clinica dovuto alla Specializzazione in Neurologia e alla competenza Scientifica, AF ha coordinato iniziative quasi sempre multidisciplinari coordinando figure professionali diverse appartenenti a diverse Istituzioni. AF ha intensamente lavorato con numerosissime **Associazioni Pazienti Malattie Rare** sia a livello nazionale che Internazionale (ad esempio World Duchenne Parent Project e Eurordis), sviluppando una capacità di interazione molto fruttuosa basata sulla reciproca fiducia, trasparenza e pragmatica tendenza ad ottenere risultati. Lavora con grande sintonia, sinergia, stima reciproca con numerose **Case Farmaceutiche** (Pfizer, Roche, Sarepta, ect) da anni, grazie in particolare al suo ruolo come PI in trial clinici, e alle sue competenze in genetica medica e ha partecipato attivamente al disegno e alle prime sperimentazioni cliniche con farmaci orfani (in particolare le molecole antisenso) per la distrofie muscolari.
Registri di malattia e Biobanche	AF ha una esperienza **vasta e di lunga durata nella gestione e coordinamento** di **gruppi di lavoro istituzionali,** anche internazionali, dedicati ad attività scientifiche,

	diagnostiche, a stesura di linee guida, e al disegno di percorsi diagnostico-terapeutici. La diretta esperienza nei **registri malattie rare** è principalmente legata alla funzione della Unita Operativa di Genetica Medica, da lei diretta, di "Hub" molecolare e Clinico della Regione Emilia Romagna. È quindi referente per le Malattie Rare e fornisce dati al Registro regionale. Inoltre, in quasi tutti i progetti EU a cui ha partecipato o che ha coordinato, sono stati sviluppati registri di malattia, correlati a specifiche misure di valutazione (registri genetici, fenotipici, di mutazione, di biomarcatori), dei quali ha quindi coordinato il disegno e lo sviluppo. AF è responsabile della **Biobanca di cellule staminali urinarie** (USC biobank), in ambito di un progetto finanziato dal Duchenne Parent Project, Italia.
Esperienza di Stesura e Supervisione di Progetti della Comunità Europea	Esperienza manageriale nel disegno ed invio di progetti scientifici multidisciplinari e con numerosi partners alla Comunità Europea (EU) (percentuale di successo 66%: inviati 15, finanziati 10), esperienza organizzativa con numerose agenzie europee di gestione progetti di ricerca , e di monitoraggio sia organizzativo che scientifico delle attività dei progetti.
Esperienze manageriali	
1999-oggi	Esperienza come **Coordinatore Scientifico di 3 progetti Europei** (FP5, FP7, H2020) con ampia esperienza di coordinamento di network internazionali con oltre 50 50 partners in 20 paesi Europei e con colleghi affiliati in USA, incluse industrie e piccole imprese. Notevole esperienza nella gestione e finalizzazione del Budget e dei report amministrativi e scientifici.
2006-oggi	**Direttore della Unità Operativa Complessa di Genetica Medica** presso la Azienda Universitaria Ospedaliera di Ferrara, con coordinamento di 30 persone, incluso personale infermieristico, del comparto, medici, biologi, tecnici, e amministrativi, ampia competenza nella gestione del budget, nella definizione della linee guida operative , e disegno dei programmi assistenziali.
2007-2019	**Direttore della Scuola di Specializzazione in Genetica Medica** della Università di Ferrara, indirizzo Medico e Sanitario, in ambito della quale ha svolto attività di gestione e formativa e ha diplomato circa 60 giovani specialisti.
2008-2011	**Coordinatore del programma di ricerca regionale (Area sanitaria) della Regione Emilia-Romagna**: 'The contribution of novel technologies to the improvement of diagnosis/therapies of breast cancer' con l'obiettivo di mettere a punto la diagnostica NGS del il tumore della mammella.
2010-oggi	**Investigatore Principale in trial clinici** multicentrici e internazionali per la Distrofia Muscolare, con incarichi di gestione di gruppi complessi e multidisciplinari per tutte le attività inerenti ai trials (amministrative, infermieristiche, di reporting, analisi dei dati, cliniche) in ambito di gruppi di lavoro internazionali e con Centri clinici trials in tutto il mondo.
2011-2014	**Coordinatore scientifico del Gruppo di lavoro IRDIRC** WG "Biomarkers for Disease Progression and Therapy Response", nel quale ambito ha coordinato oltre 50 ricercatori da 18 paesi si Europei USA, Australia, e altri
2012-2015	**Coordinatore Scientifico del network Malattie Rare all'interno della partnership International Scientific Technology Programme (ISTIP)** con i paesi del Medio Oriente e del Golfo Arabo (GCC), ha facilitato lo scambio di conoscenze e prospettive con colleghi di Arabia Saudita, Emirates Arabi, Oman, Kuwait, e Qatar, con molti di loro è ancora in contatto per varie collaborazioni scientifiche e strategiche.
2012-2015	**Coordinatore del Gruppo di Lavoro di Genetica Molecolare** della Società Italiana di Genetica Umana, (SIGU), composto da 65 membri, in tale ruolo ha contribuito a stilare documenti e pubblicazione correlate alla diagnosi molecolare genetica e alle sue linee guida

Partecipazione a Congressi e Conferenze	
1992-oggi	Partecipazione come relatore invitato per presentare letture magistrali, risultati scientifici, interventi, tavole rotonde, abstract e comunicazioni orali a oltre 200 eventi nazionali e internazionali

CV Narrativo

Alessandra Ferlini .

Si laurea in Medicina e Chirurgia presso l'Università di Bologna nel 1983 sotto la guida del Prof. Giovanni Romeo, allora docente presso l'Università di Bologna, con una tesi sulle basi genetiche ed enzimatiche della Schizofrenia. Nonostante il suo profondo interesse per la Genetica, su suggerimento del Prof. Romeo, fa domanda ed è ammessa alla Scuola di Specializzazione in Neurologia dell'Università di Bologna. In quegli anni lavora presso il Laboratorio di Genetica diretto dal Prof. Romeo, dedicandosi alla messa a punto delle nuove tecniche basate sull'approccio della genetica inversa, oltre ad avviarsi all'esperienza medica come consulente genetista per le malattie neurologiche.
Durante quei quattro anni, per sostenersi economicamente ma anche per acquisire esperienza in medicina, lavora anche come medico di base in piccoli paesi vicino a Bologna, nonché nei servizi di pronto soccorso in vari Ospedali bolognesi dove matura un'esperienza molto importante come medico. Forte delle sue buone conoscenze e del talento musicale come pianista e cantante lirica, voce di contralto, lavora come musicista e cantante d'opera e d'operetta in orchestre da camera barocca e in cori, in numerose manifestazioni e concerti in Italia e in Europa, conseguendo anche il Diploma di Cantante di Coro (Corista) al Teatro dell'Opera di Bologna (1989).
Si specializza in Neurologia (luglio 1988) con una tesi sulle basi molecolari della Sindrome di Rett. Nell'ottobre 1988 si trasferisce all'Università di Modena per lavorare con il Prof. Antonino Forabosco, che aveva appena ricevuto un importante finanziamento nell'ambito del Progetto Genoma Umano, per la parte italiana che era il braccio lungo del cromosoma X. Fino al 1992 ha lavorato felicemente nel gruppo di Modena, e ha ottenuto il primo assegno di ricerca presso l'Università di Modena. Ma l'esperienza più importante che ha avuto è stata il rapporto con la sede modenese della UILDM (Unione Italiana Lotta alla Distrofia Muscolare). I contatti con i pazienti, le famiglie, l'intensa esperienza di consulenza genetica, i primissimi test prenatali per la distrofia muscolare di Duchenne eseguiti in modo molto innovativo e pionieristico hanno influenzato profondamente la sua educazione, formazione e attitudine alla ricerca. Nel periodo modenese è stata ammessa alla Scuola di Specializzazione in Genetica Medica dell'Università di Ferrara (1989) dove ha completato la formazione di Genetista Medico. Non senza dolore e qualche stress, nel luglio del 1993 è costretta a lasciare l'Università di Modena.
Nel settembre 1993 ha vinto una borsa di studio del CNR per lavorare nell'équipe del Dott. Paolo Vezzoni, presso il CNR di Milano, dove fa parte del gruppo del premio Nobel Prof. Dulbecco impegnandosi nel progetto Genoma Umano e lavorando sempre sul braccio lungo del cromosoma X. Questa esperienza di quasi 2 anni è stata profondamente importante per accrescere la sua abilità nelle tecniche di laboratorio molecolare e per rendersi conto dell'importanza della natura internazionale della ricerca, fatto che ha plasmato tutta la sua attività negli anni successivi. E infatti, in seguito a quell'esperienza, ha deciso di cercare incarichi all'estero. Molto fortunatamente, ha incontrato Francesco Muntoni alla Conferenza Internazionale delle Malattie Neuromuscolari a Kyoto, nel 1994. A seguito di questo incontro si è candidata per alcune posizioni nel Regno Unito e ha alfine ottenuto un contratto con il Muscular Dystrophy Group, seguito da un contratto di ricerca senior della British Heart Foundation, e nel marzo 1995 ha iniziato il suo lavoro presso l'Unità Neuromuscolare, Hammersmith Hospital, Londra, diretta dal Prof Victor Dubowitz, e sotto la supervisione di Francesco Muntoni. Vi è rimasta fino al luglio 1999, e poi per altri due stage più brevi nel 2000 (2 mesi) e nel 2001 (3 mesi), per completare alcuni lavori scientifici e il suo dottorato di ricerca in Genetica presso l'Imperial College School of Medicine nel 2022. I suoi controrelatori sono stati la Prof. Key Davies e il Prof. Terry Partridge.

Gli anni londinesi sono stati unici e fondamentali per la sua formazione come medico e ricercatrice, ha infatti affinato un modo di pensare e atteggiamenti di ricerca che ha poi sempre cercato di perseguire instancabilmente negli anni successivi. I suoi rapporti con i colleghi anglosassoni e internazionali, con il suo supervisore Francesco Muntoni, hanno lasciato un segno indelebile nel suo modello di lavoro e anche nella sua visione della vita.

Nel 1999 decide di rientrare in Italia avendo vinto, grazie al gentile ed affettuoso suggerimento del Dott. Domenico De Grandis, una posizione di Dirigente Medico in Neurologia presso il Policlinico Universitario Sant'Anna di Ferrara.

Nel 2002 si sposta all'Unità di Genetica Medica, e nel 2006 diventa Professore Associato e Direttore dell'Unità di Genetica Medica, sua attuale affiliazione.

I primi anni italiani (1999-2006) sono stati profondamente impegnati nell'attività diagnostica sia in laboratorio che in clinica, con l'obiettivo di progettare e consolidare una robusta Unità di Genetica Medica con vocazione diagnostica, si è quindi dedicata in particolare alla messa a punto dell'emergente e potente metodo di genetica molecolare per la diagnosi delle malattie rare. In quegli anni non ha potuto dedicare molto tempo alla ricerca. La situazione è migliorata da 2007 in poi, quando è stata premiata con finanziamenti UE, Telethon e altri, che le hanno permesso di riprendere le sue attività di ricerca, ancora oggi in corso e parte fondamentale del suo lavoro.

Attività di ricerca in breve

Già da molti anni il suo preminente interesse e il suo maggiore sforzo di ricerca sono concentrati sulle distrofinopatie e sulle patologie neuromuscolari ereditarie. Scopo della ricerca condotta è stato identificare i processi che determinano la regolazione dell'espressione di questo gene, con particolare riferimento al processo di splicing e alla sua modulazione. Il suo gruppo di ricerca sta studiando sistemi in vitro e in vivo per modulare mutazioni del gene con oligonucleotidi antisenso utilizzando nuovi sistemi di rilascio (nanomateriali). Inoltre, il team è lavora per la definizione del profilo dell'RNA di varie patologie ereditarie e dell'identificazione di marcatori molecolari sia nell'uomo che in modelli animali. Sta inoltre studiando nuovi approcci terapeutici per le distrofinopatie e altre patologie neuromuscolari, nonché aspetti innovativi della diagnostica molecolare con particolare attenzione alle tecniche ad elevato parallelismo di sequenziamento di nuova generazione. Si occupa da anni degli aspetti etici della genetica medica e delle terapie delle malattie rare genetiche.

Interessi personali e hobby

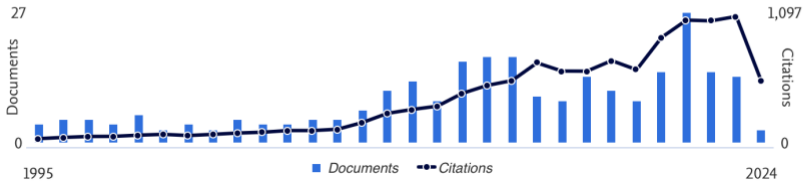
I suoi interessi personali sono caratterizzati da una grande passione per la musica, lo sport, gli animali domestici, l'agricoltura e la natura in generale. [illegible] ha ottimi rapporti con moltissimi colleghi in tutto il mondo.

	PUBLICATIONS
Peer reviewed scientific articles on indexed journals	
	298 peer reviewed publications https://pubmed.ncbi.nlm.nih.gov/?term=ferlini+a&sort=pubdate **RESEARCH METRICS AND PROFILE AUTHENTICATORS** Scopus ID 57215381030 ORCiD 0000-0001-8385-9870 RESEARCHER ID K-7532-2016 **Scopus** referenced (update 15th June 2024): **11.121 Citations by *8,321 documents*** **298 Publications** **60 *h*-index** **GoogleScholar** referenced (update 15th June 2024) **14.999 Citations** **298 Publications** **66 h-index**
Books Chapter and Books' review	
	1. A.Ferlini and P. Rimessi Methods in Molecular Biology Chapter (7) Title Exon Skipping Quantification by Real-Time PCR Copyright Year 2012 Copyright Holder Springer Science+Business Media, LLC. 2. A.Ferlini, M.S. Falzarano Nanoparticles to deliver Antisense Oligonucleotides aimed at exon skipping therapies Springer RNA Technologies Series Editors Volker A. Erdmann and Jan Barciszewski Part IV Nucleic Acids Nanotechnologies in Medicine: Diagnosis and Treatment of Diseases Editors Volker A. Erdmann and Jan Barciszewski (2015) 3. A.Ferlini, C.Scotton. Chapter metrics for "Biomarkers in Rare Genetic Diseases", published in the book: Role of Biomarkers in Medicine Edited by: Mu Wang and Frank A. Witzmann ISBN 978-953-51-2505-1 Publisher: InTech Publication date: August 2016 4. DNA and ability to reproduce: the 'Secret' of evolution, by Alessandra Ferlini European Journal of Human Genetics volume 20, pages 244–245 (2012) 5. The 'ESSENCE' of child psychiatry: lumping and splitting ADHD and associated conditions ADHD and Its Many Associated Problems by Alessandra Ferlini & Marcella Neri European Journal of Human Genetics volume 24, page 151 (2016) 6. Genetic Heterogeneity and Human Disease Reviewed by Alessandra Ferlini and Sergio Fini Eur J Hum Genet. 2015 Apr; 23(4): 559.

Publications' analyses (Scopus)

The analysis of publications types clearly shows that Rare Diseases (several types and variety of research approaches and disciplines, including genetics and genomics, pathology, molecular biology, pharmacology and novel therapies, are the master fields of research and publication areas.

Document & citation trends



Analyze author output Citation overview

Most contributed Topics 2018–2022

Dystrophin; Exon; Antisense
13 documents

Genetic Screening; Exome; Human Genomics
9 documents

Dystrophin; Duchenne Muscular Dystrophy; Animal Disease Model
5 documents

View all Topics

299 Documents | Author Metrics (New) | Cited by 8,321 documents | 4 Preprints | 2,994 Co-Authors | 39 Topics | 0 Awarded Grants (Beta)

299 documents

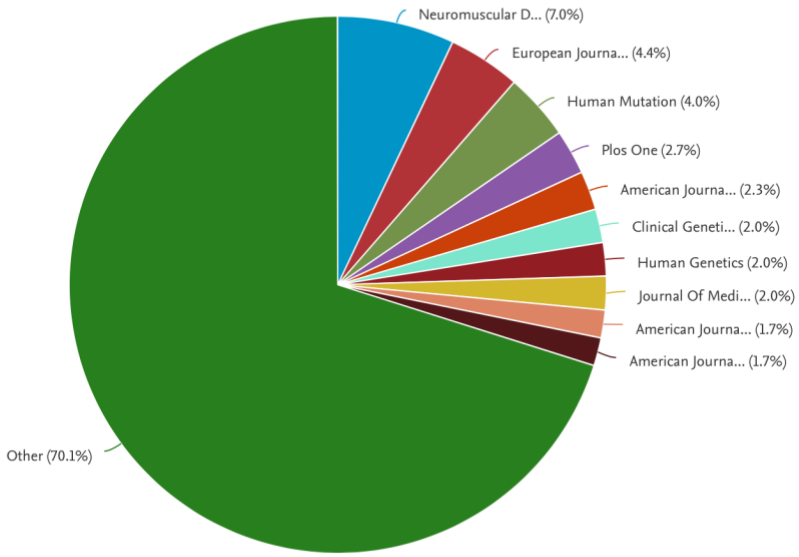
Ferlini, Alessandra

University of Ferrara, Ferrara, Italy
Author ID:57215381030

Source	Documents
Neuromuscular Disorders	21
European Journal Of Human Genetics	13
Human Mutation	12
Plos One	8
American Journal Of Medical Genetics	7
Clinical Genetics	6
Human Genetics	6
Journal Of Medical Genetics	6
American Journal Of Human Genetics	5

Documents by source

298

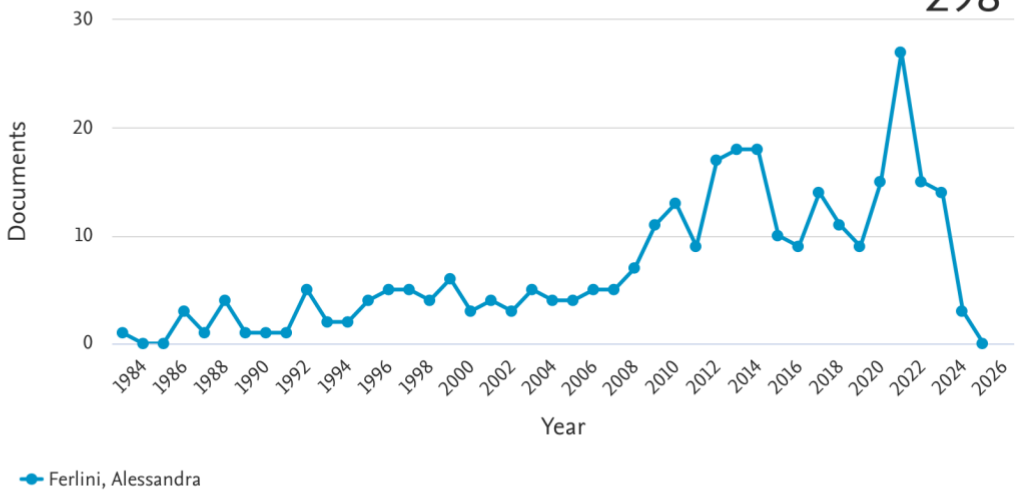


Ferlini, Alessandra
University of Ferrara, Ferrara, Italy
Author ID:57215381030

Year	Documents
2024	3
2023	14
2022	15
2021	27
2020	15
2019	9
2018	11
2017	14
2016	9
2015	10

Documents by year

298

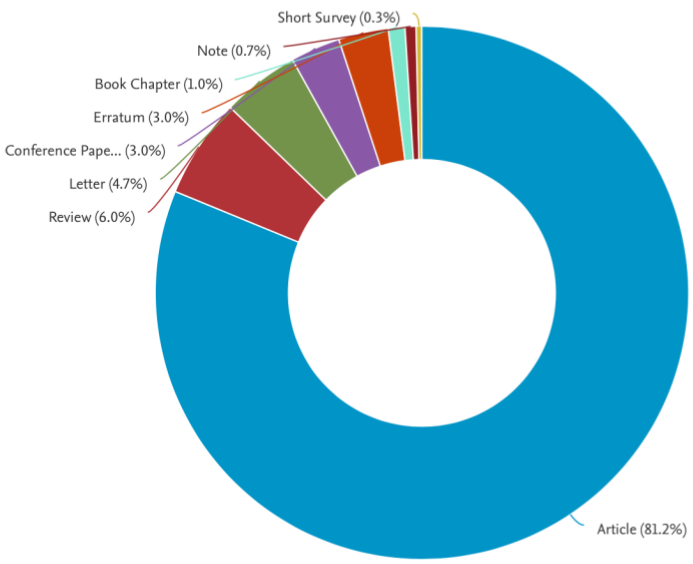


Ferlini, Alessandra
University of Ferrara, Ferrara, Italy
Author ID:57215381030

Document type	Documents
Article	242
Review	18
Letter	14
Conference Paper	9
Erratum	9
Book Chapter	3
Note	2
Short Survey	1

Documents by type

298



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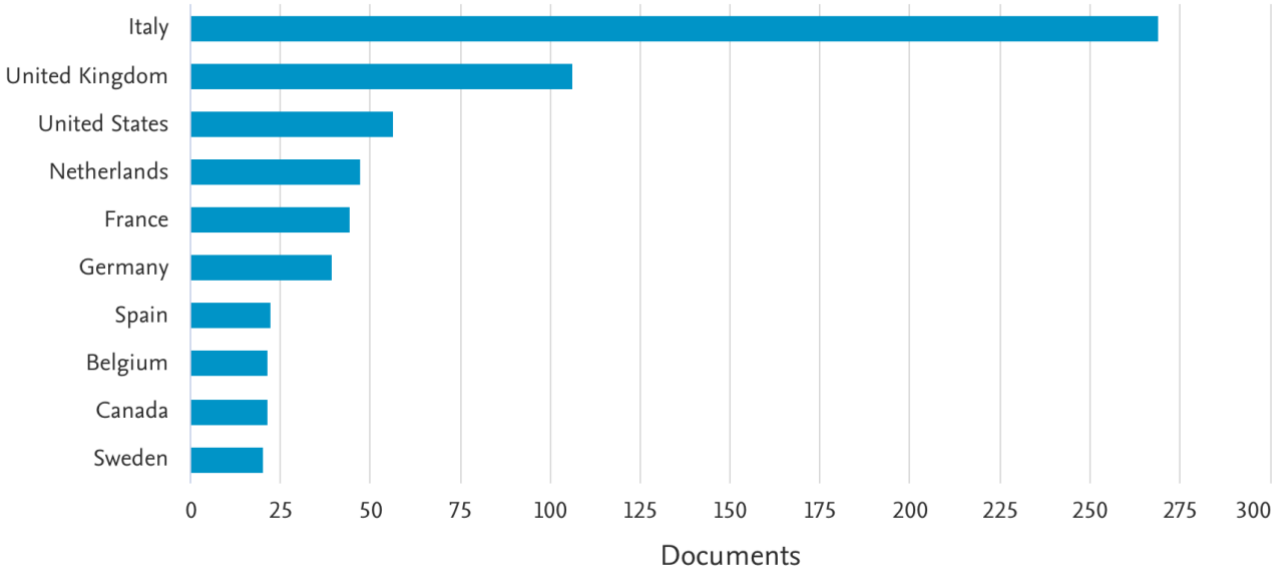

Ferlini, Alessandra

University of Ferrara, Ferrara, Italy
Author ID:57215381030

Subject Area ↓	Documents ↑
Medicine	232
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Neuroscience	46
Multidisciplinary	13
Pharmacology, Toxicology and Pharmaceutics	12
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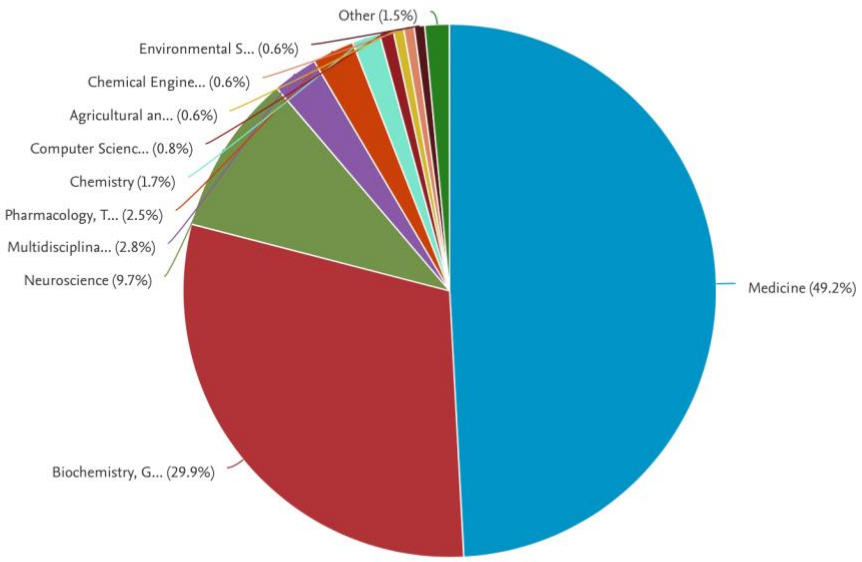
Computer Science	4
Agricultural and Biological Sciences	3
Chemical Engineering	3

Documents by subject area



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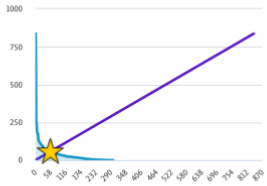

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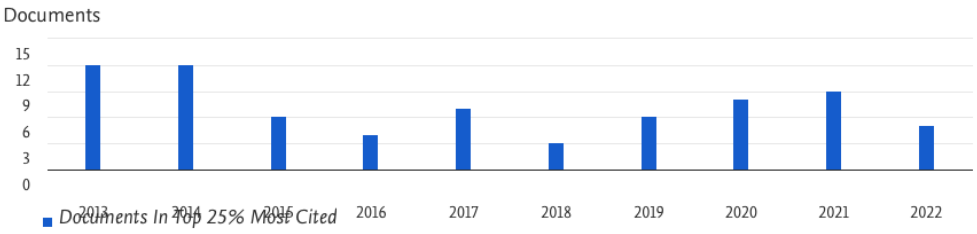

Documents in top 25% journals by CiteScore percentile

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Percent of documents in the top 25% journals by CiteScore

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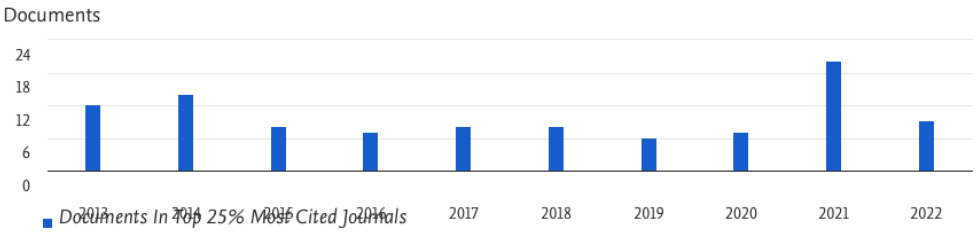

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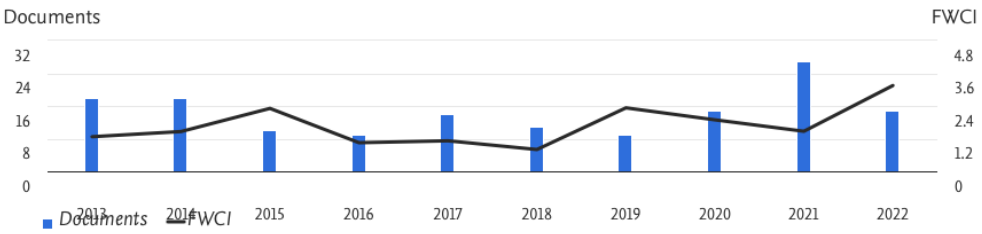
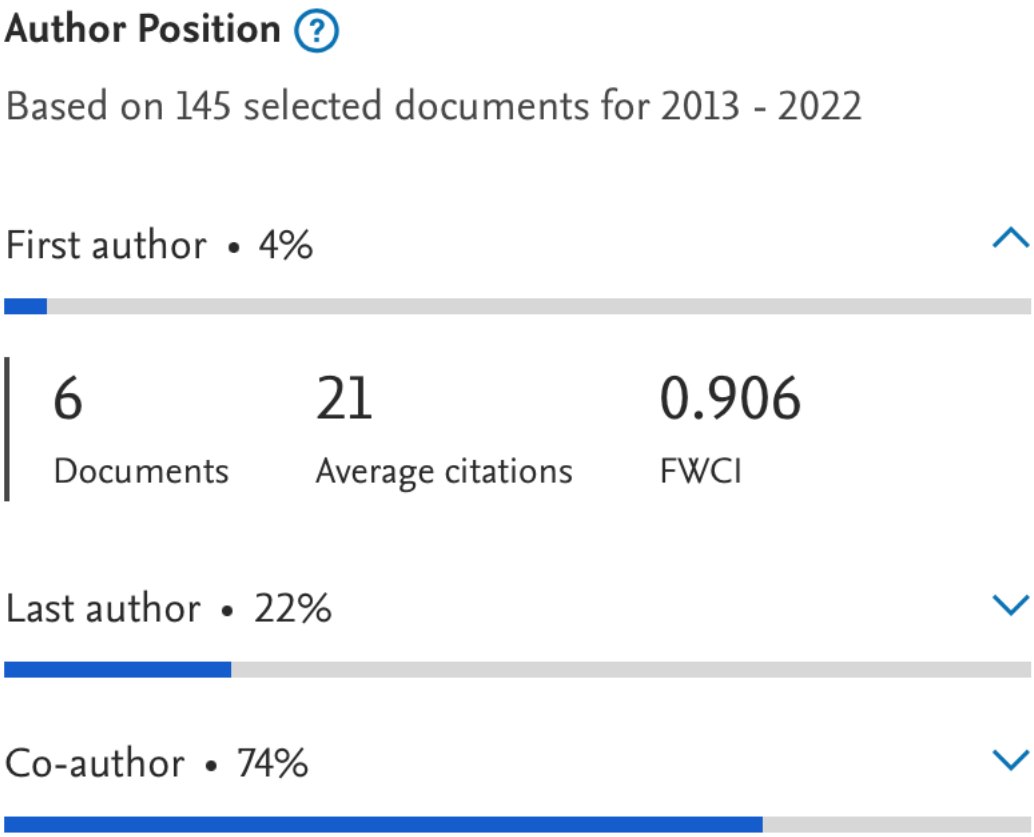
1.66
FWCI

Note: Highly cited publications for entities with a small scholarly output may skew the FWCI. This metric should be used with care when assessing performance.





Publications

LIST OF PUBLICATIONS (297)
(Scopus, PubMed)

1: Steyaert W, Sagath L, Demidov G, Yépez VA, Esteve-Codina A, Gagneur J, Ellwanger K, Derks R, Weiss M, den Ouden A, van den Heuvel S, Swinkels H, Zomer N, Steehouwer M, O'Gorman L, Astuti G, Neveling K, Schüle R, Xu J, Synofzik M, Beijer D, Hengel H, Schöls L, Claeys KG, Baets J, Van de Vondel L, Ferlini A, Selvatici R, Morsy H, Saeed Abd Elmaksoud M, Straub V, Müller J, Pini V, Perry L, Sarkozy A, Zaharieva I, Muntoni F, Bugiardini E, Polavarapu K, Horvath R, Reid E, Lochmüller H, Spinazzi M, Savarese M; Solve-RD DITF-ITHACA; Solve-RD DITF-Euro-NMD; Solve-RD DITF-RND; Solve-RD DITF-EpiCARE; Matalonga L, Laurie S, Brunner HG, Graessner H, Beltran S, Ossowski S, Vissers LELM, Gilissen C, Hoischen A. Unravelling undiagnosed rare disease cases by HiFi long-read genome sequencing. medRxiv [Preprint]. 2024 May 4:2024.05.03.24305331. doi: 10.1101/2024.05.03.24305331. PMID: 38746462; PMCID: PMC11092722.

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Titles

LIST OF TITLES

TITOLI DI FORMAZIONE (6)
EDUCATION TITLES (6)

1. -LAUREA IN MEDICINA E CHIRURGIA UNIVERSITA DI BOLOGNA 1983
2. -ESAME DI STATO ABILITAZIONE MEDICO CHIRURGO 1984-BOLOGNA
3. -ISCRIZIONE ALBO DEI MEDICI (1983-PRESENT)
4. SPECIALIZZAZIONE IN NEUROLOGIA 1988 UNIVERSITA DI BOLOGNA
5. -SPECIALIZZAZIONE IN GENETICA MEDICA 1993 UNIVERSITA DI FERRARA
6. - PhD IN GENETICS IMPERIAL COLLEGE LONDON (UK) 2002HONORARY VISITING PROFESSOR UNIVERSITY COLLEGE LONDON (UK) 2015

TITOLI DI CARRIERA (4)
CAREER TITLES (4)

1. SENIOR RESEARCH ASSISTANT HAMMERSMITH HOSPITAL LONDON UK
2. HONORARY VISITING PROFESSOR UNIVERSITY COLLEGE LONDON (UK) 2015 and 2021
3. STATO DI SERVIZIO AZIENDA OSPEDALIERA UNIVERSITARIA S. ANNA FERRARA (1997-PRESENT)
4. STATO DI SERVIZIO UNIVERSITA DI FERRARA (2006-PRESENT)

(copia dei titoli di Formazione e di Carriera in calce a questo documento)

TITOLI PROFESSIONALI E DI RICERCA : 57
PROFESSIONAL AND RESEARCH TITLES :57
(solo lista)
All listed, past, present, and pending

1. Membro del Chromosome 18 Sequencing Committee (US)
2. ENMC Scientific Committee, Membro
3. Coordinatore del Gruppo di Genetica Molecolare SOCIETA ITALIANA DI GENETICA UMANA SIGU
4. Coordinatore del gruppo Biomarkers for Disease Progression and Therapy Response IRDIRC
5. Membro del Comitato Etico del TREAT-NMD Alliance
6. Membro dell' European Board di Sarepta Therapeutics per "Exon skipping therapy"
7. Membro del PTC Therapeutics European Board
8. Membro dei gruppi di lavoro istituzionali della Regione Emilia Romagna:

-Malattie neuromuscolari
-Disordini del movimento e malattia di Huntington
-Tavolo Nascita e screening neontali
-Test Genetici

9. Membro del Comitato Etico del Progetto Europeo NMD-Chip
10. Membro del Comitato Etico Unico Area Vasta Centrale Regione Emilia Romagna
11. Membro del Comitato Etico dell'Azienda Ospedaliera Universitaria di Ferrara
12. Membro del TREAT-NMD Alliance Ethical Committee
13. Fondazione Telethon, premio per il miglior progetto di
14. British Heart Foundation Grant Research Officer, London (UK)
15. Muscle Dystrophy Group Contract, Research Assistant contract, London (UK)
16. Boncompagni-Ludovisi (Svezia), Premio di Studio Europeo per studi genomici innovativi
17. EMBO (Germania), Premio di Studio Europeo
18. Comitato Sanremo Genetica Umana, Premio Miglior studio di Sindrome di Rett
19. Professore a Contratto Universita di Modena in Neurogenetica
20. Borsa di Studio CNR, 1993-1994, MILANO Istituto Tecnologie Biomediche Avanzate, Gruppo Ricerca Premio Nobel Renato Dulbecco, Progetto Genoma Italy
21. DYNALIFE COST Action Partner
22. SCREEN4CARE H2020 EU-IMI project Scientific Coordinator
23. BIND H2020, Full Partner
24. SOLVE-RD, H2020, Full Partner
25. EXONSKIPPING COST Action, Full Partner
26. NEUROMICS, FP7 Full Partner
27. SIGN Network Genetico Sloveno-Italiano, Full Partner
28. BIO-NMD FP7 , Scientific Coordinator
29. NMD-CHIP FP6 Full Partner
30. FINGER FP5 Scientific Coordinator
31. 3-2021, Telethon Grant-Partner
32. 2-2021, Telethon Grant Partner
33. GGP09093 (2009) Telethon Grant-Coordinator
34. GGP08107 (2008) Telethon Grant-Partner
35. GUP07011 (2010) Telethon Grant -Coordinator
36. GGP05115 (2007) Telethon Grant- Coordinator
37. GGP02311 (2010) Telethon Grant - Partner
38. 1054 (1993) Telethon Grant- Coordinator
39. 146 (1994) Telethon Grant -Partner
40. Regione Emilia-Romagna: 'RARER: Next generation sequencing and gene therapy to diagnose and cure rare diseases in the Emilia Romagna', full partner
41. PRIN MIUR ITALIA , PI "Molecular bases of CollagenVI myopathies"
42. Regione Emilia-Romagna: 'The contribution of novel technologies to the improvement of diagnosis/therapies in Medical Genetics of breast cancer', Coordinator
43. DUCHENNE PARENT PROJECT ONLUS ITALIA (2007-2013) PI (4 grants),
44. DUCHENNE PARENT PROJECT ONLUS ITALIA Urinary Stem Cell Biobank
45. PTC Therapeutics diagnostic service Grant (2018-2022) Duchenne muscular dystrophy diagnosis for patients from Eastern Europe and North African countries.

46. Association Francoise pour le myopathies (AFM) France, grant Full Partner; The Popeye domain containing gene 1 and its role in muscular dystrophy
47. Sarepta Therapeutics Grants (2) to accelerate genetic diagnosis of neuromuscular diseases
48. ERN Euro-NMD , rappresentante
49. ERN ITHACA, rappresentante
50. ERN EuroBloodNet, affiliato
51. ICONS (www.icons.org) International consortium for Newborn sequencing, membro dell' Executive Committee
52. Trial NCT02329769 (Open Label, Extension Study of PRO044 in Duchenne Muscular Dystrophy (DMD))
53. Trial NCT01037309 (Phase I/II Study of PRO044 in Duchenne Muscular Dystrophy (DMD)
54. Trial NCT02958202 Extension Study of BMN 044 in Duchenne Muscular Dystrophy (DMD)
55. Trial NCT01480245 (Open Label Study of GSK2402968 in Subjects With Duchenne Muscular Dystrophy)
56. Trial NCT01254019 (A Clinical Study to Assess the Efficacy and Safety of GSK2402968 in Subjects With Duchenne Muscular Dystrophy (DMD114044)
57. Trial NCT02500381 Study of SRP-4045 (Casimersen) and SRP-4053 (Golodirsen) in Participants With Duchenne Muscular Dystrophy (DMD) (Essence)

Autorizzo il trattamento dei miei dati personali ai sensi del Decreto Legislativo 30 giugno 2003, n. 196 "Codice in materia di protezione dei dati personali (facoltativo)".

Firma

16th June 2024