CURRICULUM VITAE

REDATTO AI SENSI DEGLI ARTICOLI 46, 47 E 49 DEL D.P.R. 445/00
(DICHIARAZIONI SOSTITUTIVE DI CERTIFICAZIONI E DELL'ATTO DI NOTORIETA')

Personal information

First name / Surname	**Antonio Mauro**
Nationality	Italian

Scientific experience

Dates	July 2012-July 2017
Main activities and Responsibilities	Involved in following genetic counselling activities : reproductive, prenatal, neuromuscular, cardiogenetic (dilated, hypertrophic, arrhythmogenic cardiomyopathies), oncogenetic (in particular breast/ovarian cancer), dysmorphologic fields Clinical work in multidisciplinary team for clinical trials Sub-investigator, study coordinator and data manager (Professor Alessandra Ferlini as PI) in clinical trials on antisense oligonucleotide exon skipping in Duchenne Muscular Dystrophy; all trials were conducted according to ICHGCP:

ClinicalTrials.gov Identifier: NCT02500381

(A double-blind, placebo-controlled, multi-center study with an open-label extension to evaluate the efficacy and safety of SRP-4045 and SRP-4053 in patients with Duchenne Muscular Dystrophy)

ClinicalTrials.gov Identifier: NCT02329769

(Open Label, Extension Study of PRO044 in Duchenne Muscular Dystrophy (DMD))

ClinicalTrials.gov Identifier: NCT01037309

(Phase I/II Study of PRO044 in Duchenne Muscular Dystrophy (DMD))

ClinicalTrials.gov Identifier: NCT01480245

(Open Label Study of GSK2402968 in Subjects With Duchenne Muscular Dystrophy)

ClinicalTrials.gov Identifier: NCT01254019

(A Clinical Study to Assess the Efficacy and Safety of GSK2402968 in Subjects With Duchenne Muscular Dystrophy (DMD114044))

I have received specific ICHGCP training on 15th November 2014.

Work experience

Dates	September 2017- June 2018
Occupation or position held	Medical Geneticist (Scholarship)
Name and address of employer	Medical Genetics Unit – Ferrara Hospital
Main activities and Responsibilities	Research on "Application of new clinical outcome measures in patients affected by Duchenne Muscular Dystrophy during clinical trials on drugs" Clinical follow-up for patients affected by Duchenne Muscular Dystrophy

Dates	March 2012-June 2012
Occupation or position held	Frequent Medical Doctor
Name and address of employer	Paediatric Clinic – University of Padova
Type of business or sector	Hospital Establishment

Education and training

Name and type of organisation providing education and training	6 months (February 2016-September 2016) of practical experience in Cytogenetic Laboratory at Medical Genetics School (DNA extraction, QF-PCR, acquisition of metaphases and reconstruction of karyogram with dedicated software (Cytovision), analysis of CGH-array data using bioinformatic programs (UCSC, Ensemble, Decipher))

Dates	July 11th, 2017
Title of qualification	Specialisation in Medical Genetics
Final degree mark	110/110
Thesis title	Defining the genetic bases of dystrophinopathies in a therapeutic perspective
Thesis supervisor	Professor Alessandra FERLINI
Name and type of organisation providing education and training	Reproduction and Growth Department - Medical Genetics Unit – Ferrara University/Hospital and Woman and Child Health Department – Medical Genetics Unit Padova University/Hospital

Dates	7th March 2012
Name and type of organisation providing education and training	Registration with Association of Physicians-Surgeons and Dentist in Naples N° 33581

Dates	16th February 2012
Title of qualification awarded	Qualification to the Medical-Surgeon Profession
Name and type of organisation providing education and training	Second University of Naples

Dates	11th October 2011
Title of qualification awarded	Degree in Medicine and Surgery
Final degree mark	105/110
Thesis type	Experimental
Thesis title	The stones in Idiopathic Hypercalciuria: long-term evolution in the Paediatric age group
Thesis supervisor	Professor Giuliana LAMA
Name and type of organisation providing education and training	Second University of Naples

Dates	2008-2011
Type of training awarded	Undergraduated experience at the Third Division of Paediatrics (Paediatric Nephrology) headed by Professor G. LAMA – Second University of Naples. During this period I have attended several clinical cases; I also conducted a retrospective study of a group of paediatric patients with kidney stones: it lead me to draft my thesis of degree

Dates	July 2001
Title of qualification awarded	Scientific Certificate (Italian Secondary School)
Final examination mark	100/100
Name and type of organisation providing education and training	Scientific Lyceum "E.Medi"- Cicciano (Naples)

Meeting Participations

Dates	10th March 2018
Title	Giornata Malattie Neuromuscolari
Name and type of organisation	Ospedale Bellaria Bologna - Istituto delle Scienze Neurologiche di Bologna
Dates	16th – 18th February 2018
Title	XVI Parent Project International Conference: Duchenne and Becker Muscular Dystrophy
Name and type of organisation	Parent Project Onlus
Dates	24th November 2017
Title	Attualità e controversie in neuroscienze.
Name and type of organisation	Auditorium Regione Emilia-Romagna – Bologna – Professor Valeria Tugnoli
Dates	15th – 17th November 2017
Title	SIGU (XX National Meeting)
Name and type of organisation	SIGU
Dates	Ferrara, 4th June 2017
Title	Focus on Cardiogenetics
Name and type of organisation	Ferrara Hospital – Medical Genetics Unit and Cardiology Unit – Professor Alessandra Ferlini and Professor Roberto Ferrari
Dates	Firenze, 24th March 2017
Title	GdL Genetica Clinica: II meeting annuale 2017, GdL Genetica Molecolare: I meeting annuale 2017
Name and type of organisation	Meyer Hospital – Professor Rita Giglio
Dates	Roma, 17th-19th February 2017
Title	XV Parent Project International Conference: Duchenne and Becker Muscular Dystrophy
Name and type of organisation	Parent Project Onlus
Dates	Torino, 23th-25th November 2016
Title	SIGU (XIX National Meeting)
Name and type of organisation	SIGU
Dates	Ferrara, 11th October 2016
Title	I Course of Updating on Molecular Diagnosis of Duchenne Muscular Dystrophy and Dystrophinopathies
Name and type of organisation	Medical Genetics Unit Professor Alessandra Ferlini
Dates	Roma, 12th-14th February 2016
Title	XIV Parent Project International Conference: Duchenne and Becker Muscular Dystrophy
Name and type of organisation	Parent Project Onlus

Dates	Bologna, 26th-28th November 2015
Title	SINP (XLI National Meeting): The clinical competency in Paediatric Neurology: from diagnosis to therapy (like speaker)
Name and type of organisation	SINP
Dates	Abano Terme, 24th October 2015
Title	New borders in Neuromuscular Diseases, Coordinator Dr. D. De Grandis
Name and type of organisation	Dr. De Grandis Congress studio Venezia international
Dates	Rimini, 21th-23th October 2015
Title	SIGU (XVIII National Meeting)
Name and type of organisation	SIGU
Dates	Barcelona, 17th-18th September, 2015
Title	BioMarin 051-302 Investigator Meeting
Name and type of organisation	Biomarin
Dates	Ferrara, 4th June, 2015
Title	Focus on Cardiogenetics
Name and type of organisation	Medical Genetics Unit Professor Alessandra Ferlini
Dates	Bologna, 17th April 2015
Title	XXVIII IMER Meeting on Central Nervous System Congenital Anomalies
Name and type of organisation	IMER
Dates	Rome, 21th-22th February 2015
Title	XIII Parent Project International Conference: Duchenne and Becker Muscular Dystrophy
Name and type of organisation	Parent Project Onlus
Dates	Leiden, 27th-28th November 2014
Title	Investigators' meeting of PRO044-CLIN-02 study
Name and type of organisation	Prosensa – QED Clinical Services Limited
Dates	Bologna, 30th-31th October 2014
Title	SIGU (National Meeting): The NGS in Human and Medical Genetics
Name and type of organisation	SIGU
Dates	Ferrara, 28th October 2014
Title	Phenotypes of the mutated SWI/SNF complex
Name and type of organisation	Medical Genetics Unit – Professor Alessandra Ferlini

Dates	Ferrara, 3rd October 2014
Title	Genetic disorders of the skeleton: from patient to gene and vice versa
Name and type of organisation	Medical Genetics Unit – Professor Alessandra Ferlini
Dates	Rome, 21th-23th February 2014
Title	XII Parent Project International Conference: Duchenne and Becker Muscular Dystrophy
Name and type of organisation	Parent Project Onlus
Dates	Naples, 27th-28th June 2008
Title	Updates in Nephro-Urology Paediatric
Name and type of organisation	Pediatrics Unit (Second University of Naples)
Dates	Naples, 18th May 2007
Title	Chronic critical ischemia of the lower limbs
Dates	Naples, 31th October 2006
Title	Skin ulcers: diagnosis and therapy
Dates	Naples, 20th May 2006
Title	The protocols of anticoagulation and antiplatelet in cardiovascular diseases

Course Participations

Dates	Rome, 12th-14th March 2015
Title	XXVII Updating Course: New concepts of Neuro-muscular Diseases in paediatric age
Name and type of organisation	Fondazione Mariani - Professor E. Mercuri
Dates	Rome, Gemelli Hospital 10th-11th December 2014
Title	Clinical evaluation training for clinical trials, Physiotherapist E. Mazzone
Name and type of organisation	Professor E. Mercuri
Dates	Leiden, 27th-28th November 2014
Title	Clinical evaluation training, Prosensa & QED Clinical Services Limited
Dates	Naples, 29th – 30th May 2007
Title	Theoretical and Practical Course on "The suturing in Surgery", coordinator Professor U. Parmeggiani
Dates	Naples, 11th – 12th December 2006
Title	Theoretical and Practical Course on "Basic Life Support and Defibrillation (BLS-D)", coordinator Professor M. Verza

Abstracts

Title

NEXT GENERATION SEQUENCING IN DYSTROPHINOPATHIES: MUTATION DETECTION AND SNPs PROFILING

Antonio Mauro[1], Marcella Neri[1], Simona Neri [1], Rachele Rossi[1], Cecilia Trabanelli[1], Marina Fabris[1], Paola Rimessi[1], Sergio Fini[1], Francesca Gualandi[1], Rita Selvatici[1], Fernanda Fortunato[1], Alessandra Ferlini[1,2] and the Italian DMD Clinical Network*

**Pini A., Merlini L., Mora M., Gorni K., Sansone V., Mongini T., Pegoraro E., Tonin P., Fiorillo C., Bruno, C., Filosto M., D'Amico A., Bertini E., Pane M., Mercuri E. Messina S., Vita G., (Bologna, Milano, Torino, Padova, Verona, Genova, Brescia, Roma, Messina)*

1 University Hospital of Ferrara St. Anna, Unit of Medical Genetics, Ferrara, Italy

2-Neuromuscular Unit, UCL, London UK

Trials innovativi con exon-skipping nella Distrofia Muscolare di Duchenne: valutazione delle reazioni avverse. A. Mauro1, L. Mantovani 3, M.L. Conighi2, C. Bleve2, A. Armaroli1, M.E. Michelini 2, A. Franchella2, M.R. Virgili3, A. Ferlini1

1Dip. di Riproduzione e Accrescimento, U.O. di Genetica Medica, A.O.U. Ferrara

2Dip. di Riproduzione e Accrescimento, U.O. di Chirurgia Pediatrica, A.O.U Ferrara

3Dip. Medico Specialistico, U.O. di Dermatologia, A.O.U. Ferrara

CLINICAL NEXT GENERATION SEQUENCING GENE PANEL IDENTIFIED A NOVEL ATP7A MUTATION IN TWO BROTHERS WITH DISTAL HEREDITARY MOTOR NEUROPATHY AND AUTONOMIC DYSFUNCTION.

Francesca Gualandi[1] , Eleonora Italyankina[1], Markus Storbeck[2], Katharina Vezyroglou[2], Raoul Heller[2], Chiara Scotton[1], Francesca Di Raimo[1], Antonio Mauro[1], Valeria Tugnoli[3] , Vincent Timmerman[4] , Brunhilde Wirth[2], Domenico De Grandis[5], Alessandra Ferlini[1] .

1. Department of Medical Sciences, Section of Medical Genetics, University of Ferrara- Unit of Medical Genetics – University Hospital, Ferrara, Italy

2. Institute of Human Genetics, University Hospital of Cologne, Cologne , Germany

3. Department of Neuroscience and Rehabilitation, Division of Neurology, University Hospital of Ferrara, Ferrara, Italy

4. VIB - Department of Molecular Genetics Peripheral Neuropathy Group, University of Antwerpen, Belgium

5. UILDM- Verona, Italy

TRIALS CLINICI NELLA DISTROFIA MUSCOLARE DI DUCHENNE (DMD) CON OLIGORIBONUCLEOTIDI ANTISENSO: IL PERCORSO DI DRISAPERSEN (EXON 51 SKIPPING) *Mauro A.[1], Armaroli A.[1], Bleve C.[2], Conighi M.L. [2], Michelini M.E.[2], Franchella A.[2], Pane M.[3], Mercuri E.[3] Ferlini A.[1]*

1. Dip. di Riproduzione e Accrescimento, U.O. di Genetica Medica, A.O.U. Ferrara

2. Dip. di Riproduzione e Accrescimento, U.O. di Chirurgia Pediatrica, A.O.U Ferrara

3. Sez. di Neuropsichiatria Infantile, Università Cattolica, Roma

Impatto dell'a-CGH nell'inquadramento diagnostico genetico della disabilità intellettiva

G. Parmeggiani1, A. Mauro1, B. Buldrini 1, R. Gruppioni1, M. Neri 1, S. Bigoni1, F. Gualandi1, S. Fini1, A. Ferlini1

1 Dip. di Riproduzione e Accrescimento, Unità di Genetica Medica, A.O.U. Ferrara

Trials clinici con farmaci orfani mutazione specifici per la Distrofia Muscolare di Duchenne: la nostra esperienza con gli oligoribonucleotidi antisenso

A. Mauro1, A. Armaroli1, C. Bleve 2, M.L. Conighi2, M.E. Michelini2, F. Fortunato1, E. Cesca2, A. Cazzuffi2, A. Franchella2, M.
Pane3, E. Mercuri3, A. Ferlini1

1Dip. di Riproduzione e Accrescimento, U.O. di Genetica Medica, A.O.U. Ferrara

2Dip. di Riproduzione e Accrescimento, U.O. di Chirurgia Pediatrica, A.O.U. Ferrara

3Sez. di Neuropsichiatria Infantile, Università Cattolica, Roma

Publications

Title

Genetic counseling for women referred for advanced maternal age: a telegenetic approach. Gualandi F, Bigoni S, Melchiorri L, Buldrini B, Balboni A, Neri M, Armaroli A, Parmeggiani G, Italyankina E, Mauro A, Ravani A, Fini S, Caracciolo S, Ferlini A. Genet Med. 2014 Oct;16(10):795. doi: 10.1038/gim.2014.103.

Le ipoacusie infantili di grado moderato/severo: il ruolo del genetista. Stefania Bigoni, Antonio Mauro, Alessandra Ferlini. Audiologia&Foniatria, 2016; 1(1): 16-22.

Cochlear malformation and sensorineural hearing loss in the Silver-Russell syndrome. Bigoni S, Mauro A, Ferlini A, Corazzi V, Ciorba A, Aimoni C. Minerva Pediatr. 2017 Sep 7.

Multilevel molecular analysis identifies all dystrophin gene mutations pointing out that DMD is a genetically homogeneous disease: repercussions on diagnosis, prevention and therapy. M. Neri, R. Selvatici, M. Falzarano, C. Trabanelli, A. Ravani, P. Rimessi, M. Fabris, C. Scotton, A. Mauro, F. Fortunato, H. Osman, R. Rossi, A. Armaroli, B. Buldrini, S. Fini, F. Gualandi, x. Clinical Study Group, A. Ferlini. Neuromuscular Disorders Volume 27, Supplement 2, October 2017, Page S198

Personal skills and competences

Mother tongue

Italian

Other language(s)

Self-assessment

European level (*)

English

Understanding		Speaking		Writing
Listening	Reading	Spoken interaction	Spoken production	
A2	B1	A2	A2	B1

(*) Common European Framework of Reference (CEF) level

Computer skills and competences

Excellent ability to use PC (Word, Power-Point, Excel, Internet)

July, the 7th 2018