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Curriculum Vitae

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Education

07/ 1974	High School Diploma Liceo Scientifico "A.Calini", Brescia, Italy
11/ 1977	BSc (cum laude) University of Pavia, Italy
10 / 1985	Doctoral Degree in Medicine (cum laude) Faculty of Medicine, University of Brescia, Italy
06 /1991	Ph.D, in Human Pathology at the University of Pavia, Italy
11/ 1993	Post Doctorate Specialization (cum laude) in Human Cytogenetics University of Pavia

Professional Activity

02/ 1991 – 06/ 1992	Postdoctoral fellow at Centre for Research on Neurodegenerative disorders, University of Toronto, Toronto, Canada
10/ 1991 – 09/ 2005	Assistant Professor of Medical Genetics, University of Pavia, Pavia, Italy.
11/ 1996 – 10/ 1998	Visiting scientist, Howard Hughes Medical Institute, Program in Molecular Medicine, University of Massachusetts, Worcester, Massachusetts, USA.
11/ 1998 – 10/2002	Instructor at the Department of Biochemistry and Molecular Biology, University of Massachusetts Medical School, Worcester, Massachusetts, USA.
06/ 2000- 05/ 2006	Director of Laboratory for Diagnosis of Neurogenetic Disorders, Pavia, Italy
11/2002 - present	Research Assistant Professor at the Program in Gene Function and Expression, University of Massachusetts Medical School, Worcester, Massachusetts, USA.
06/ 2004- 05/ 2009	Coordinator of FSH Club, Association Française contre les myopathies,
10/ 2005-present	Associate Professor of Medical Genetics at the Department of Biomedical Sciences, Faculty of Biosciences and Biotechnologies, University of Modena and Reggio Emilia, Modena, Italy
11/ 2006– 3/ 2019	Director of the Graduate School in Molecular and Regenerative Medicine University of Modena and Reggio Emilia, Modena, Italy
06/ 2006-present	Director of Miogen Laboratory for Research and Diagnosis of Neuromuscular Disorders, University of Modena and Reggio Emilia
1/ 2007- present	Italian National Registry for FSHD, Curator
4/ 2007-3/ 2010	member of the Ethics Committee of the province of Modena, Italy

2008 – 2012	Member of the Center Advisory Board (CAC) of Paul D. Wellstone Muscular Dystrophy Cooperative research Center Boston Biomedical Research Institute, Watertown, USA
2010-2012	Member of the Scientific Council of the Association Française contre les myopathies, Paris, France
2010-2012	Member of the AFM Strategic and Therapeutic Development Committee (COSET) of the Steinert Network
2012	ad hoc Member of the Study Section (Skeletal Muscle and Exercise Physiology) at Center for Scientific Review, National Institute of Health, USA
2013 - 2015	Massachusetts Wellstone Cooperative Research Center for FSHD Research, University of Massachusetts Medical School, Worcester, Member
2015	2015/10 ZRG1 GGG-Q Innovative Therapies and Tools for Screenable Disorders in Newborns Study Section, Center for Scientific Review, NIH, USA, ad hoc member
2015	2015/10 TAG (Therapeutic Approaches to Genetic Diseases) Study Section, Center for Scientific Review, National Institute of Health, USA, ad hoc member
2016-2019	Strategic Committee Unione delle Associazioni, Italy, member
2016-2020	International Consortium for FSHD, organizer
2018-present	Li Weibo Institute for Rare Diseases Research at the University of Massachusetts Medical School, member
2019-2022	TREAT-NMD : Global database oversight committee – TGDOC, Disease Subgroup Lead for FSHD

Honors and Fellowships

1991 Anna Villa Rusconi Postdoctoral Fellowship

1997 CNR (National Research Centre) Senior Scientist Fellowship

1998 FSH Society - Delta Railroad Construction Postdoctoral Fellowship

2000 Worcester Foundation Award

2000 Legato Ferrari Award.

Grants

Active

Fondo di Ateneo per la Ricerca anno 2021 - Ricerca interdisciplinare Mission Oriented (Coordinator)
Title: Artificial intelligence for the management and analysis of clinical and molecular data of the Italian National Registry of Facioscapulohumeral muscular dystrophy and the Emilia Romagna Registry of Amyotrophic Lateral Sclerosis
Duration: 06/1/22 – 05/31/24
Direct costs: € 66,000

Telethon GSP18002, Italy (D'Amico PI)
Title: A patient registry for muscular dystrophies and myopathies
Duration: 03/01/20 – 02/28/2023
Direct costs: € 240,000

Completed

Regione Emilia Romagna Progetti - Alte Competenze per la Ricerca e il Trasferimento Tecnologico -
Title: Analisi, sviluppo e implementazione della Piattaforma Software per la gestione avanzata e l'ottimizzazione della raccolta dati dell'Italian National Registry of facioscapulohumeral muscular dystrophy
Duration: 01/09/20 – 31/08/21
Direct costs: € 30,000

Regione Emilia Romagna Piano triennale alte competenze alla ricerca POR/FSE 2014/2020 (PI)
Title: *An integrated approach for the management and analysis of clinical and molecular data of the Italian National Registry for Facioscapulohumeral muscular dystrophy*
Duration: 11/01/2018 - 10/31/2021
Direct costs: € 86,743.44

Association Française contre les myopathies (Ricci PI) 21611
Title: *Characterization of the phenotypic variability in FSHD families for assisting clinical research*
Duration: 12/01/2018 – 11/30/2019
Direct costs: € 50,000

Fondo di Ateneo per la Ricerca anno 2016 - Ricerca interdisciplinare (Coordinator)
Title: In Silico Analysis and Machine Learning Techniques for the Molecular Modeling of Genomic Variants in Muscular Dystrophies
Duration: 3/1/17 – 2/28/19
Direct costs: € 80,000

Telethon GUP13012, Italy (Coordinator) 07/1/14 – 03/31/18
Title: *Phenotypic and molecular characterization of FSHD families: a systematic approach towards trial readiness.*
Direct costs: € 575,000

FSHD Global Research Foundation, Australia (PI)
Title: *Functional study of a novel candidate gene for FSHD.*
Duration: 09/1/14 – 02/28/18
Direct costs: AUD 325,000

Association Française contre les myopathies (D'Antona PI) 10/1/15 – 09/30/17
Title: *Study of the role of muscle fatigue as predictor of muscle degeneration infacioscapulohumeral muscular dystrophy*
Duration: 10/1/15 – 09/30/17
Direct costs: € 80,000

National Institutes of Health/NICHHD (Emerson PI) 2U54HD060848-07
Title: *Biomarkers for Therapy of FSHD*
Project I: Genetic Modifiers of FSHD (Wagner PI)
Duration: 09/16/13 - 05/31/15

Association Française contre les myopathies (PI) 16593
Title: *Whole exome sequencing to dissect genetic complexity in Facioscapulohumeral muscular dystrophy.*
Duration: 10/1/14 – 09/30/16
Direct costs: € 80,000

Muscular Dystrophy Association (Coordinator)
Title: *Dissecting the complexity of FSHD molecular pathogenesis*
Duration: 09/1/12 – 08/31/14
Direct cost: $ 208,335

Telethon, GUP11009, Italy (Coordinator)
Title: *Establishment of the Italian National Registry for Facioscapulohumeral muscular dystrophy*
Duration: 06/01/12 – 05/31/13
Direct costs: euros 200,000

Association Française contre les myopathies 14339 (PI)
Title: *Improvement of molecular tools for FSHD diagnosis*
Duration: 05/1/10 – 04/30/12

Direct costs: euros 130,000

European Union - Programme "Marie Curie Initial Training Networks" (Co-PI)
Title: *Chromatin diseases: from basic mechanisms to therapy*
Duration: 09/1/09 – 08/31/13
Direct costs: euros 120,000

Regione Emilia Romagna, Italy (RU-PI)
Title: *Next-generation sequencing and gene therapy to diagnose and cure rare diseases in Regione Emilia Romagna (RER)*
Duration: 06/01/2013 – 31/05/2016
Direct costs: euros 75,000

National Institutes of Health; NIAMS (PI)
Title: *An animal model to develop therapeutic strategies for FSHD*
Duration: 07/01/07 – 06/30/12
Direct cost: $ 1,250,000

Association Française contre les myopathies (PI)
Title: *Analysis of the role of FRG1 in FSHD pathogenesis*
Duration: 09/1/07 – 08/31/10
Direct costs: euros 120,000

Telethon, GUP08004, Italy (Coordinator)
Title: *Clinical and laboratory criteria for FSHD diagnosis in view of a national registry for the disease*"
Duration: 01/01/09 – 12/31/10
Direct costs: euros 250,000

FSHD Global Research Foundation (PI)
Title: *Defining the mechanism controlling muscle-specific gene expression in FSHD*
Duration: 02/01/09 – 01/31/10
Direct cost: $ 120,000

Association Française contre les myopathies (Coordinator)
Title: *Defining the pathogenesis of facioscapulohumeral muscular dystrophy (FSHD)*
Duration: 09/1/08 – 08/31/10
Direct costs: euros 444,000

Association Française contre les myopathies (Coordinator)
Title: *Defining the pathogenesis of facioscapulohumeral muscular dystrophy (FSHD)*
Duration: 03/1/06 – 02/28/08
Direct costs: euros 644,000

Ministry of Education, University and Research – PRIN 2006 (Co-investigator)
Title: *Therapeutic strategies for FSHD*
Duration: 11/1/06– 10/31/08
Direct cost: euros 30,000

Telethon, GUP07001, Italy (Coordinator)
Title: *Clinical and laboratory criteria for FSHD diagnosis in view of a national registry for the disease*"
Duration: 12/01/07 – 12/30/08
Direct costs: euros 250,000

National Institutes of Health; NIND; R01 NS047584-01 (PI)
Title: *Investigating the Molecular Basis of FSHD*
Duration: 1/10/03 - 6/30/08

Direct Costs: $1,156,250

Telethon, GGP050501, Italy (PI)
Title: *Dissecting the molecular mechanism responsible for facioscapulohumeral muscular dystrophy (FSHD)*
Duration: 07/1/05 – 06/30/08
Direct cost: euros 437,000

Ministry of Education, University and Research – PRIN 2004 (Co-investigator)
Title: *Molecular basis of FSHD*
Duration: 11/1/04 – 10/31/06
Direct cost: euros 80,000

Association Française contre les myopathies (PI)
Title: *The molecular basis of facioscapulohumeral muscular dystrophy*
Duration: 10/1/03 – 9/30/05
Direct costs: euros 80,000

Association Française contre les myopathies (PI)
Title: *Defining the pathogenesis of facioscapulohumeral muscular dystrophy (FSHD)*
Duration: 10/1/04 – 30/9/05
Direct costs: euros 322,000

National Research Council, CNR; Functional Genomics: Progetto Unità Operativa n° 2.1.6 (Co-investigator)
Title: *Dissection of Facioscapulohumeral muscular dystrophy pathogenic mechanism through the analysis of affected muscle transcription profile*
Duration: 1/1/03 – 12/31/04
Direct costs: euros 114,428.62

Ministry of Education, University, and Research, FIRB RBAU01B7TR (PI)
Title: *Identification of the pathogenic mechanism responsible for facioscapulohumeral muscular dystrophy and development of diagnostic, prognostic and therapeutic strategies.*
Duration: 11/1/02 – 31/10/05
Direct costs: euros 150,000

Muscular Dystrophy Association (PI)
Title: *Dissection of the Molecular Mechanism causing FSHD*
Duration: 1/1/02 – 12/31/04
Direct cost: $ 208,335

CARIPLO (PI)
Title: *Identification and characterization of genes involved in facioscapulohumeral muscular dystrophy*
Duration: 7/1/03 – 30/6/05
Direct costs: euros 50,000

Telethon GP0284/01 (PI)
Title: *Dissecting the molecular mechanism responsible for facioscapulohumeral muscular dystrophy (FSHD)*
Duration: 11/1/01 – 10/31/04
Direct cost: euros 185,924.52

Ministry of Instruction, University, and Research – PRIN 2003 (Co-investigator)
Title: *Identification of candidate genes in facioscapulohumeral muscular dystrophy*
Duration: 10/1/03 – 9/30/04
Direct costs: euros 39,000

National Institutes of Health; NIND; R21 NS043973 (PI)
Title: *Analysis of the Molecular and Functional Role of D4Z4 in FSHD Pathogenesis*
Duration: 9/30/01 – 8/31/04

Ministry of University, Research, and Scientific Technology– COFIN (Co-investigator)
Title: "Analysis of gene expression in FSHD affected muscles"
Duration: 10/1/01 – 9/30/03

Muscular Dystrophy Association (PI)
Title: *Analysis of gene expression in FSHD affected muscle*
Duration: 7/1/99 – 6/30/01

Telethon 1041 (PI)
Title: *Study of differentially expressed genes in facioscapulohumeral muscular dystrophy (FSHD) affected muscles: an alternative approach towards the identification of the FSHD molecular defect*
Duration: 1/1/98-12/31/00

Telethon 729 (PI)
Title: *Genetic and physiological approach to identify the defect responsible for facioscapulohumeral muscular dystrophy (FSHD)*
Duration: 10/1/95 – 9/30/96

Telethon A432 (PI)
Title: *Linkage analysis and physical mapping of the facioscapulohumeral dystrophy locus*
Duration: 10/1/93 – 9/30/94

Presentation at Meetings

July 1996	Workshop on Facioscapulohumeral muscular dystrophy, Naarden, The Netherlands
October 1997	American Society of Human Genetics, Satellite Meeting on Facioscapulohumeral muscular dystrophy, Baltimore, USA
July 1998	Workshop on Facioscapulohumeral muscular dystrophy, Pavia, Italy
October 1998	American Society of Human Genetics, Satellite Meeting on Facioscapulohumeral muscular dystrophy, Denver, USA
November 1998	International meeting on Facioscapulohumeral muscular dystrophy, Rome, Italy
May 2000	The Third International Symposium on the Cause and Treatment of Facioscapulohumeral muscular dystrophy, Bethesda, USA
July 2000	FSH Society Annual Meeting, Natick, USA,
May 2000	National Congress of the Italian Society of Clinical Neurophysiology, Bergamo, Italy
October 2000	The Eighth Congress of the Italian Society of Environmental Mutagenesis, Mondello, Italy
November 2000	National Congress of the Italian Society of Human Genetics, Orvieto, Italy,
March 2001	Association Francaise contre les Myopathies, Meeting on Facioscapulohumeral Muscular Dystrophy, Paris, France
October 2001	American Society of Human Genetics, Satellite Meeting on Facioscapulohumeral muscular dystrophy, San Diego, USA
November 2001	Italian Society of Human Genetics, Annual Meeting, Orvieto,

October 2002	7th International Congress of the World Muscle Society , Rotterdam, The Nederlands
October 2002	FSH Society Annual Meeting, Rockville, Maryland, USA
October 2002	American Society of Human Genetics Annual Meeting, Satellite Workshop on FSHD, Baltimore, Maryland, USA
November 2002	Convention Telethon, Riva del Garda, Italy
December 2002	Association Francaise contre les Myopathies - Workshop on FSHD, Paris, France,
May 2003	European Society of Human Genetics annual meeting, Birmingham, UK
October 2003	Italian Federation of Life Sciences, FISV annual meeting , Rimini, Italy,
November 2003	American Society of Human Genetics annual meeting, Los Angeles, USA
November 2003	American Society of Human Genetics Annual Meeting, Satellite Workshop on FSHD, Los Angeles, USA
January 2004	"New directions in Biology and Disease of Skeletal Muscle", San Diego, USA
October 2004	American Society of Human Genetics Annual Meeting, Toronto, Canada
October 2004	American Society of Human Genetics, Satellite Meeting on Facioscapulohumeral muscular dystrophy, Toronto, Canada.
May 2005	Myologie 2005, Nantes, France
October 2005	18th IGB Meeting - Workshop on Epigenetic Bases of Genome Reprogramming, Capri, Italy.
October 2005:	American Society of Human Genetics, Satellite Meeting on Facioscapulohumeral muscular dystrophy, Salt Lake City, USA.
February 2006	3rd Mediterrenean Congress of Neurology, Sharm El Sheikh, Egypt (cancelled).
November 2006	"Facioscapulohumeral Muscular Dystrophy: How Deletion of Repetitive Elements Leads to Muscular Dystrophy", 5th International *Conference* on *Unstable Microsatellites and Human Disease*, Granada,
January 2007	"Analysis of the role of *FRG1* in the pathogenesis of Facioscapulohumeral Muscular Dystrophy" EMBO Conference Series 1st Meeting on "pre-mRNA processing and disease" Cortina D'Ampezzo, Italia
May 2007	"Gene dysregulation in FSH", FSH Workshop, Institut de Myologie, Paris, Francia
May 2007	"Size and number of D4Z4 alleles play a role in FSHD phenotype." VII Congresso Nazionale dell'Associazione Italiana di Miologia , Ferrara
October 2007	"Selective muscle involvement in facioscapulohumeral muscular dystrophy: the role of 4q35 gene expression", FSHD International Research Consortium 2007, San Diego, USA
June 2008	"Dissecting the role of FRG1 and alternative splicing alterations of specific mRNAs in FSHD pathogenesis", EMBO Conference on RNA and Disease, Roma, Italia
October 2008	"Pathogenic hypothesis of facio-scapulo-humeral muscular dystrophy'', 13th Congress of the World Muscle Society, Newcastle, UK
November 2008	"Italian FSHD National Registry: a tool for genotype-phenotype correlation" American Society of Human Genetics, Satellite Meeting on Facioscapulohumeral muscular dystrophy, Philadelphia, USA.
May 2009	"Italian FSHD National Registry: Clinical and laboratory criteria for FSHD diagnosis" , Italian Association to fight Muscular Dystrophy, Naples, Italy
June 2009	"Unexpected high percentage of asymptomatic subjects carrying the FSHD molecular defect." IX Congresso Nazionale dell'Associazione Italiana di Miologia , Verona, Italy
June 2009	"Past, present and future of FSHD" Sen. Paul D. Wellstone Muscular Dystrophy Cooperative Research Center for FSHD Research Annual Retreat, Watertown, USA

January 2010	:The FSHD Italian Registry, FSHD clinical score and genotype-phenotype correlation" 171st ENMC International Workshop, Naarden, The Netherlands.
April 2010	I Giornata per la FSHD: Presentazione del Registro nazionale per la malattia, Modena Italy
26 April 2010	1st DISCHROM Conference "Epigenetics, Epigenomics in Health and Diseases, Naples, Italy. Relazione su "DNA damaging and chromatin modification at the FSHD locus".
May 2010	"Il Registro Nazionale per la distrofia muscolare facio-scapolo-omerale (FSHD): uno strumento per ottenere nuovi parametri prognostici." Corso di Aggiornamento. La patologia neuromuscolare in eta' evolutiva, Calambrone, Pisa, Italy
June 2010	Studio di correlazione genotipo-fenotipo per l'FSHD: quali fattori influenzano l'insorgenza e la progressione della malattia? X Congresso nazionale dell'Associazione Italiana di Miologia, Milan, Italy
June 2010	Homogenization of genetic testing, the Italian experience, Best Practice Meeting, Leiden, The netherlands
October 2010	II Giornata per la FSHD: *Presentazione del Registro nazionale per la malattia,* Messina, Italy
October 2010	Unexpected large number of compound heterozygotes revealed by cumulative effects of D4Z4 mutation, FSH Society FSHD Workshop, Watertown, MA, USA
October 2010	"Distrofia muscolare facio-scapolo-omerale: stato dell'arte e registro nazionale", XLI Italian Society of Neurology, Catania, Italy
February 2011	La distrofia muscolare facio-scapolo-omerale", "Updates nelle Malattie Neuromuscolari", Brescia, Italy
October 2011	Presentazione Registro nazionale per la malattia: risultati e prospettive, Seconda Giornata per la FSHD, Modena, Italy
November 2011	"Altered Troponin T Splicing in Fast Fibers Drives Muscle Weakness in Facioscapulohumeral Muscular Dystrophy", FSH Society FSHD Workshop, Watertown, MA, USA
November 2011	"Facioscapulohumeral muscular dystrophy: new insights from compound heterozygotes and implication for prenatal genetic counseling" FSH Society FSHD Workshop, Watertown, MA, USA
May 2012	"Facioscapulohumeral Muscular Dystrophy: genetic complexity and novel diagnostic perspective XII Congresso Nazionale dell'Associazione Italiana di Miologia , Scicli, Italy
October 2012	"Distrofia muscolare facio-scapolo-omerale: nuovi aspetti diagnostici e genetici", XLIII Italian Society of Neurology, Rimini, Italy
7 November 2012	"Prognostic tools for familial facioscapulohumeral muscular dystrophy: results of a large-scale genotype-phenotype analysis from the Italian National Registry for FSHD", ASHG satellite meeting – FSH society, San Francisco, USA
March 2013	*From the bench to the clinic: what we have learnt from the Italian National Registry for Facioscapulohumeral muscular dystrophy (FSHD).* XVII Convention Telethon, Riva del Garda (TN), Italy
April 2013	"*Il Registro Nazionale Italiano per la FSHD: Risultati e Obiettivi*." Terza gionata FSHD, Modena, Italy
July 2013	*"FSHD revisited: what we have learn from a large a genotype/phenotype study"* DisChrom Workshop "From clinic to benchwork and return: is this possible?" Modena, Italy
16-20 September 2013	International Summer School Rare Disease And Orphan Drug Registries, Roma, Italia. Relazione su The role of registries in epidemiological, clinical and genetic research on rare diseases. A case example: the Italian National Registry for facioscapulohumeral muscular dystrophy".

May 2014	Next Generation Sequencing in facioscapulohumeral muscular dystrophy patients supports the idea that FSHD is a complex genetic disease, 14° Congresso Nazionale AIM Sirmione, Italy
8-9 March 2015	Un'iniziativa per lo studio integrato delle malattie neuromuscolari, NMD Convention, XVIII Convention Telethon, Riva del Garda, Italy.
18 settembre 2015	Organizzazione Corso ECM "La variabilità fenotipica nella distrofia muscolare facio-scapolo-merale: training all'utilizzo di una nuova classificazione clinica", Modena, Italia.
19 Settembre 2015	Organizzazione Scientifica della Quarta Giornata per la FSHD. "Il Registro Nazionale Italiano per la Distrofia Muscolare Facio-scapolo-omerale:un modello da esportare", Modena, Italia.
1-4 October 2015	Facioscapulohumeral muscular dystrophy: more complex than it appears, XII IIM-Myology meeting, Reggio Emilia, Italy
March 2016	Phenotypic and molecular characterization of FSHD families: a systematic approach towards trial readiness, Myology 2016, Lyon, France.
November 2016	Disease progression and clinical history in 246 patients from the FSHD Italian Registry, FSH Society IRC Meeting, Boston.
November 2016	The adult FSHD phenotype, 225th ENMC International Workshop: A global FSHD Registry framework
18-20 November 2016	The Italian FSHD registry experiences of a professional reported registry - Clinician reported, 225th ENMC International Workshop: A global FSHD Registry framework
10 February 2017	La distrofia facio-scapolo-omerale, Malattie Neuromuscolari: Aspetti Diagnostici e Terapeutici, Brescia, Italy
2 March 2017	Distrofia Facioscapolomerale: una malattia da riscrivere, Sport, nutrizione e terapia farmacologica: un approccio integrato per il miglioramento della salute e della qualità di vita delle persone, Chiavari, Italy
4 March 2017	Giornata Nazionale per le malattie Neuromuscolari Parma, Italia. Relazione su "Il registro nazionale per la distrofia muscolare facio-scapolo-omerale".
13-15 March 2017	"Il registro della Distrofia Muscolare Facioscapolomerale", NMD Convention, XIX Convention Telethon, Riva del Garda, Italy
31 May – 3 June 2017	"La distrofia muscolare Facio-scapolo-omerale", 17° Congresso Nazionale AIM Siracusa, Italy
23 September 2017	"FSHD 2.0: dalla raccolta dati alla pratica clinica, 5ª Giornata per la FSHD Modena", Italy.
28 October 2017	Cinquantenario dell'Unione Italiana per la Lotta alla Distrofia Muscolare, sezione di Torino, Il miglioramento dell'approccio clinico e terapeutico per le malattie neuromuscolari: la svolta del nuovo millennio. La FSHD. Torino, Italy.
6-9 June 2018	Fenotipo clinico come guida allo studio molecolare, 18° Congresso Nazionale AIM Genova, Italy
15-16 November, 2018	"La diagnosi genetica nella distrofia muscolare facio-scapolo-omerale : poche certezze e molti punti oscuri, Imaging delle malattie Neuromuscolari e del nervo periferico: stato dell'arte", Milan, Italy
9 March, 2019	"Nuovi apetti terapeutici: focus sulle distrofie muscolari". Giornata Nazionale per le malattie Neuromuscolari Parma, Italia.
17 May, 2019	"La funzione dei Registri per i pazienti adulti" Convegno Nazionale UILDM, Lignano, Italy
	Imaging delle malattie Neuromuscolari e del nervo periferico: stato dell'arte
5-8 June 2019	New insights in FSHD pathogenesis and possible therapies, 19° Congresso Nazionale AIM Bergamo, Italy
25 October 2019	Il valore dei Registri per le malattie neuromuscolari, Convegno UILDM Modena, Italy

Seminars and lectures

December 1997	Informative Course on Molecular Genetics, Treviso, Italy
June 1999	Department of Human and Hereditary Pathology, University of Pavia, Pavia, Italy
July 1999	Department of Human Genetics, Memorial Sloan Kettering Institute, New York, USA
April 2000	Department of Genetics, University of Bologna, Bologna, Italy
January 2001	University of Pisa, Pisa, Italy
June 2001	Institute of Molecular Genetics CNR, Pavia, Italy
December 2001	Institute de Myologie, Paris, France
May 2002	Proficiency course in muscle diseases, Plenary lecture, University of Milan, Milan, Italy
May 2002	Department of Physiopathology, University of Florence, Florence, Italy
May 2002	School of Specialization in Human Physiology, Department of Human Anatomy and Physiology, University of Padova, Padova, Italy
June 2002	School of Specialization in Medical Genetics, University of Pavia, Italy
September 2002	Proficiency course in Neurophisiopathology, Plenary Lecture, Lido degli Estensi, Italy
October 2002	University of Minnesota, Department of Genetics, Cell Biology and Development, Minneapolis, USA.
October 2002	Columbia University, Department of Physiology & Cellular Biophysics, New York, USA,.
October 2002	Weill Medical College of Cornell University, Department of Pediatric Hematology-Oncology, New York, USA
November 2002	GBMC - Institut de Génétique et de Biologie Moléculaire et Cellulaire, Strasbourg, France,
December 2002	Telethon Institute of Genetics and Medicine, Naples,.
December 2002	Department of Genetics, University of Bologna, Bologna Italy.
December 2002	Department of Genetic and Microbiology, University of Pavia, Pavia.
February 2003	Department of Medicine, University of Massachusetts Medical School, Worcester, USA
April 2004	Department of Physiology and Biophysiscs, University of Iowa Carver College of Medicine, Iowa City, Iowa,.
June 2004	ECM course on Muscle Pathophysiology, Pavia, Italy.
May 2004	Laboratoire de Biologie Moléculaire de la Cellule CNRS UMR 5161-Ecole Normale Supérieure de Lyon, Lyon, France.
February 2005	National Institutes of Neurological Disorders and Stroke, Bethesda, Maryland, USA.
March 2005	Institute de Genetique Humaine, Montpellier, France
March 2005	Novartis Pharmaceutical, Basel, Switzerland
April 2005	The Broad Institute of Harvard and MIT, Cambridge, Massachusetts, USA
November 2005	Novartis Pharmaceutical, Basel, Switzerland.
February 2006	"Le basi molecolari della distrofia muscolare facio-scapolo-omerale", Fondazione Telethon, Milano
March 2006	"La distrofia muscolare facio-scapolo-omerale: una malattia epigenetica", Università di Ferrara, Facoltà di Medicina, Ferrara, Italia.
March 2006	"Facioscapulohumeral muscular dystrophy: a mendelian disorder with epigenetic phenotype", Institute of Human Genetics, Cardiff, Wales, UK
Aprile2006	"Facioscapulohumeral muscular dystrophy: a mendelian disorder with epigenetic phenotype", Weatherall Institute of Molecular Medicine, John Radcliffe Hospital, Oxford, UK
December 2006	"Distrofia muscolare facio-scapolo-omerale: una malattia da delezione di DNA ripetitivo". Scuole di Dottorato in Biologia e Medicina, Pavia, Italia.
March 2007	"Il nuovo mondo: prospettive per la genetica medica dopo il sequenziamento del genoma umano" Accademia delle Scienze e delle Lettere, Modena
May 2007	"Epigenetica e silenziamento Genico: novi livelli di complessita' nel controllo dell'espressione genica", Scuola di Specializzazione in Genetica Medica, Università di Verona, Verona

September 2007	"Introduzione ai concetti fondamentali della genetica", Corso di Genetica e Bioetica, Università di Modena e Reggio Emilia, Modena
February 2008	"Distrofia muscolare facio-scapolo-omerale: una malattia genetica e alcune domande a livello molecolare e oltre", Istituto Nazionale di Fisica della Materia (INFM)
March 2008	"Distrofia muscolare facio-scapolo-omerale: dalle sequenze ripetute alla miopatia" Dipartimento di Scienze Biomediche, Università di Modena e Reggio Emilia, Modena
April 2008	"Miogen Lab: un laboratorio trasparente" Azienda Policlinico di Modena, Modena
October 2008	"FRG1: a key gene to understand the pathogenesis of facioscapulohumeral muscular dystrophy", Institut de Pharmacologie Moléculaire et Cellulaire (IPMC), Sophia Antipolis, France
October 2008	"Facioscapulohumeral muscular dystrophy: transition from a mendelian trait to a complex genetic disease?" Acceleron Pharma, Cambridge, MA, USA
May 2009	"Facioscapulohumeral muscular dystrophy: transition from a mendelian trait to a complex genetic disease?" Neurologic Insttitute "C. Besta", MIlan, Italy
November 2011	"Quali altri responsabili nella patogenesi della distrofia muscolare facio-scapolo-omerale?" Istituro Neurologico "C.Mondino", Pavia, Italy
March 2013	From the bench to the clinic: what we have learnt from the Italian National Registry for Facioscapulohumeral muscular dystrophy. XVII Convention Telethon, Riva del Garda, Italy.
May 2013	"OMIM, SNPs atlas & Beyond. Come approcciare un dato genetico." Corso residenziale in neurogenetica, Pisa, Italy.
July 2014	Facioscapolohumeral muscular dystrophy: how a hereditary disease can change the life of a medical geneticist, International PhD Course in Molecular Medicine, Universita' Vita e Salute, HSR, Milan, Italy.
March 2015	"Discordance in the clinical and genetic assessments of FSHD", Wellstone Center, UMMS, Worcester, USA.
January 2017	Investigating the clinical and genetic assessment in Facioscapulohumeral muscular dystrophy, Nationwide Children's, Ohio State University. Columbus, USA
April 2017	Facioscapulohumeral muscular dystrophy: What we know, what we do not know, Université Claude Bernard Lyon 1, Lyon, France.
June 2019	Un modello molecolare per la patogenesi delle distrofie muscolari. National Research Council, Bari, Department of Biochemistry and Molecular Biology "E. Quagliariello", University of Bari "A. Moro", Bari,

What we know, what we do not know

Honorary Lectureship

October 2000	The Twentieth Anniversary of Unione Italiana per la Lotta alla Distrofia Muscolare, sezione di Modena, Maranello, Italy,
October 2004	Italian Society of Human Genetics annual meeting, Pisa, Italy
May 2012	Italian Association of Myology, Scicli, Italy

Conference chaired, organized

July 1998	Workshop on Facioscapulohumeral muscular dystrophy, Pavia, Italy
October 2003	Italian Federation of Life Sciences, FISV annual meeting, Simposium 12 – Chromatin Silencing, Rimini, Italy

March 2004	ECM course on Neurogenetics, Pavia, Italy
October 2004	American Society of Human Genetics Annual Meeting, Invited Session – Position effect: the result of altered chromatin structure from yeast to mammals. Toronto, Canada
April 2010	I Giornata per la FSHD: Presentazione del Registro nazionale per la malattia, Modena Italy
October 2011	Registro nazionale per la malattia: risultati e prospettive, Seconda Giornata per la FSHD, Modena, Italy
April 2013	III giornata per la Distrofia Muscolare Facioscapolomerale (FSHD): FSHD Ieri e Oggi: quali prospettive concrete per il Futuro? Modena, Italy
July 2013	DisChrom Workshop "From clinic to benchwork and return: is this possible?" Modena, Italy
June 2016	16° Italian Association of Myology Meeting
September 2015	IV giornata per la Distrofia Muscolare Facioscapolomerale (FSHD): Il Registro Nazionale Italiano per la Distrofia Muscolare Facioscapolomerale: Un modello da esportare. Modena, Italy
November 2016	225th ENMC International Workshop: A global FSHD Registry framework Heemkerk, The Netherlands
September 2017	V giornata per la Distrofia Muscolare Facioscapolomerale (FSHD): FSDH 2.0: dalla raccolta dati alla pratica clinica. Modena, Italy
June 2019	26th FSHD International Research Congress, Marseille, France.
26 October 2019	VI giornata per la Distrofia Muscolare Facioscapolomerale (FSHD): Dalla clinica alle risposte pratiche per la vita di tutti i giorni, Modena, Italy
November 2019	17eme Journeés de la Société Française de Myologie, Marseille, France
April 2020	Consensus Conference on FSHD diagnosis, Bologna, Italy

Memberships

1996-2005	Reference Center Neuromuscular Disorders in Childhood.
1996-2006	American Society of Human Genetics.
2001-2005	Italian Society of Human Genetics
2002-2010	World Muscle Society
2012-present	Associazione Italiana di Miologia

Teaching

1994-2012	Human Genetics undergraduate course at the University of Pavia Medical School
1998-2001	Recombinant DNA technologies undergraduate course at the University of Pavia Medical School
2001-2005	Medical Genetics undergraduate course at the University of Pavia Medical School
1998-2006	Medical Genetics post-graduate course at the University of Pavia Medical School
2005-present	Medical Genetics undergraduate course at the University of Modena and Reggio Emilia, Faculty of Biosciences and Biotechnologies
2005-present	Medical Genetics post-graduate course at the University of Modena and Reggio Emilia, Faculty of Biosciences and Biotechnologies
2010-present	Human Genetics undergraduate course at the University of Modena and Reggio Emilia, Faculty of Biosciences and Biotechnologies
2005-2010	Medical Genetics post-graduate course at the University of Ferrara, School of Medicine.
2014-present	Molecular diagnostics in Medical Genetics, undergraduate course at the University of Modena and Reggio Emilia, Faculty of Medicine
2015-present	Medical Genetics, School of Medicine at the University of Modena and Reggio Emilia.

Scientific publications

1. Zuffardi O., Caiulo A:, Maraschio P., **Tupler R**., Bianchi E., Amisano P., Beluffi G., Moratti G., Liguri G. Regional assignment of the loci for adenilate kinase to 9q32 and for a-acid glycoprotein to 9q31-q32. A locus for Goltz syndrome in region 9q32-qter? *Hum Genet*, 82:17-19, 1989.
2. Maraschio P., **Tupler R**., Dainotti E., Piantanida M., Cazzola G., Tiepolo L. Differential expression of the ICF (immunodeficiency, centromeric heterochromatin, facial anomalies) mutation in lyumphocytes and fibroblasts. *J Med Genet*, 26:452-456, 1989.
3. St. George- Hyslop P.H., Haines J.L., Farrer L.A., Polinsky R., Van Broeckhoven C., Goatel A., Mc Lachlan C. D.R., Orr H., Bruni A. C., Sorbi S., Rainero I., Foncin J.F., Pollen D., Cantù J.M., **Tupler R**., et al. Genetic linkage studies suggest that Alzheimer‘s disease is not a single homogeneous disorder. *Nature*, 347:194-197, 1990.
4. Maraschio P., Zuffardi O., Caiulo A., Dainotti E., Piantanida M., Rivera H., **Tupler R**. Deletion of specific sequences or modification of centromeric chromatin are responsible for Y chromosome centromere inactivation. *Hum Genet*, 85:491-494, 1990.
5. **Tupler R**., Bortotto L., Buhler E.M., Alkan M., Malik N.J., Bosch-Al Jadooa N., Memo L., Maraschio P. Paternal origin of the de novo deleted chromosome 4 in Wolf-Hirschhorn syndrome. *J Med Genet*, 29:53-55, 1992.
6. **Tupler R**., Maraschio P., Gerardo A., Mainieri R., Lanzi G., Tiepolo L. A complex chromosome rearrangement with 10 breakpoints: tentative assignment of the locus for Williams syndrome to 4q33-q35.1. *J Med Genet*, 29: 253-255,1992.
7. Tanzi R.E., Vaula G., Romano M.D., Mortilla M., Huang T.L., **Tupler R**., et al. Assessment of amyloid b-protein precursor gene mutations in a large set of familiar and sporadic Alzheimer’s disease cases. *Am J Hum Genet*, 51:273-282, 1992.
8. Bacciocchi G., Gibelli N., Zibera C., Pedrazzoli P., Bergamaschi G., De Piceis Polver P., Danova M., Mazzini G., Palomba L., **Tupler R**., Robustelli Della Cuna G. Establishment and characterization of two cell lines derived from human glioblastoma multiforme. *Anticancer Research*, 12: 853-862, 1991.
9. Maraschio P., Cortinovis M., Dainotti E., **Tupler R**., Tiepolo L. Interphase cytogenetics of the ICF syndrome. *Ann Hum Genet*, 56:273-278, 1992.
10. Vaula G., Mortilla M., **Tupler R**., Lukiw W., Tanzi R., Nee L., Polinsky R., Foncin J.F., Bruni A.C., Montesi M.P., Sorbi S., St: George-Hyslop P. A novel but non-pathogenic mutation in exon 4 of the human amyloid precursor protein (APP) gene. *Neuroscience letters*, 144:46-48, 1992.
11. Karlinsky H., Vaula G., Haines J.L., Ridgley J., Bergeron C., Mortilla M., **Tupler R.,** et al. Molecular and prospective phenotypic characterization of a pedigree with familial Alzheimer’s disease and a missense mutation in codon 717 of the b-amyloid precursor protein gene. *Neurology*, 42:1445-1453, 1992.
12. **Tupler R**., Rogaeva E., Vaula G., Mortilla M., Lukiw W., Liang Y., Hancock R., Rogaev E., St: George-Hyslop P. A highly infornmative microsatellite repeat polymorphism in intron 1 of the human amyloid precursor protein (APP) gene. *Hum Molec Genet*, 2:620, 1993.
13. Maraschio P., **Tupler R**., Dainotti E., Cortinovis M., Tiepolo L. Molecular analysis of a Y;1 translocation in an azoospermic male. *Cytogenet Cell Genet*, 65:256-260, 1994.
14. **Tupler R**., Barbierato L., Larizza D., Sampaolo P., Piovella F., Maraschio P. Balanced autosomal translocation and ovarian dysgenesis. *Hum Genet*, 94:171-176, 1994.
15. **Tupler R**., Hoeller H., Pezzolo A., Maraschio P. Maternal derivation of inv dup (22) and clinical variation in cat-eye syndrome. *Ann Génét*, 37:153-155, 1994.
16. Maraschio P., **Tupler R**., Babrierato L., Dainotti E., Larizza D., Bernardi F., Hoeller H., Garau A., Tiepolo L. An analysis of Xq deletions. *Hum Genet*, 97:375-381, 1996.
17. Maraschio P., **Tupler R**., Rossi E., Barbierato B., Uccellatore F., Rocchi M., Zuffardi O., Fraccaro M. a novel mechanism for the origin of supernumerary marker chromosomes. *Hum Genet*, 97:382-386, 1996.

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19. **Tupler R**., Berardinelli A., Barbierato L., Frants R., Hewitt J.E., Lanzi G., Maraschio P., Tiepolo L. Monosomy of distal 4q does not cause facioscapulohumeral muscular dystrophy. *J Med Genet*, 33:366-370, 1996.
20. **Tupler R.**, Marseglia G.L., Stefanini M., Prosperi E., Chessa L:, Nardo T., Marchi A., Maraschio P. A variant of Nijmegen breakage syndrome with unusual cytogenetic features and intermediate cellular radiosensitivity. *J Med Genet*, 34:196-202, 1997.
21. **Tupler R**., Barbierato L., Memmi M., Sewry C.A., DeGrandis D., Maraschio P., Tiepolo L., Ferlini A. Identical "de novo" mutation at D4F104S1 locus in monozygotic male twins affected by facioscapulohumeral muscular dystrophy (FSHD) with different clinical expression. *J Med Genet*, 35:778-783, 1998.
22. Maraschio P., Maserati E., Seghezzi L., **Tupler R.,** Verri M.P., Tiepolo L. Involvement of 9q22.1-q31.3 region in pyloric stenosos. *Clin Genet*, 54:159-160, 1998.
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24. Maserati E., Verri A., Seghezzi L., **Tupler R**., Federico A., Tiepolo L., Maraschio P. Cerebellar dysgenesis and mental ritardation associated with a complex chromosomal rearrangement. *Ann Génét*, 42: 210-214, 1999.
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27. Gabellini D., Green M.R., **Tupler R**. Inappropriate gene activation in FSHD: A repressor complex binds a chromosomal repeat deleted in dystrophic muscle. *Cell*, 110,339-348, 2002.
28. Soragna D., Vettori A., Carraro G., Marchioni E., Vazza G., Bellini S., **Tupler R**., Savoldi F., Mostacciuolo M.L. A locus for migraine without aura maps on chromosome 14q21.2- q22.3. *Am J Hum Genet* 72: 161-167, 2003.
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31. **Tupler R**., Gabellini D. Molecular basis of facioscapulohumeral muscular dystrophy. *Cell Mol Life Sci,* 61: 557-566, 2004.
32. Gabellini D., Green M.R., **Tupler R**. When Enough is Enough: Genetic Diseases Associated with Transcriptional Derepression. *Curr Opin Genet Dev,* 14:301-307, 2004
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35. Gabellini D, D'Antona G, Moggio M, Prelle A, Zecca C, Adami R, Angeletti B, Ciscato P, Pellegrino MA, Bottinelli R, Green MR, **Tupler R.** Facioscapulohumeral muscular dystrophy in mice overexpressing FRG1. *Nature,* 449:973-977, 2006.
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39. Filosto M, Tonin P, Scarpelli M, Savio C, Greco F, Mancuso M, Vattemi G, Govoni V, Rizzuto N, **Tupler R**, Tomelleri G. Novel mitochondrial tRNA(Leu(CUN)) transition and D4Z4 partial deletion in a patient with a facioscapulohumeral phenotype. *Neuromuscul Disord*. 18:204-9, 2008. **IF 2,667**
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41. Lamperti C, Fabbri G, Vercelli L, D'Amico R, Frusciante R Bonifazi E, Fiorillo C, Borsato C, Cao M, Servida M, Greco F, Di Leo R, Volpi L, Manzoli[j], Cudia P, Pastorello E, Ricciardi L, Siciliano G, Galluzzi G, Rodolico C, Santoro S, Tomelleri G, Angelini C, Ricci E, Palmucci L, Moggio M, **Tupler R**. A standardized clinical evaluation of patients affected by Facioscapulohumeral Muscular Dystrophy: the FSHD clinical score. *Muscle and Nerve,* 42:213-7, 2010
42. Forlani G, Giarda E, Ala U, Di Cunto F, Salani M, **Tupler R**, Kilstrup-Nielsen C, Landsberger N. The MeCP2/YY1 interaction regulates ANT1 expression at 4q35: novel hints for Rett syndrome pathogenesis *Hum Mol Genet* 19:3114-23, 2010
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46. Scionti I. Fabbri G, Fiorillo C, Ricci G, Greco F, D'Amico R, Termanini A, Vercelli, Tomelleri G, Cao M, Santoro S, Percesepe A, **Tupler R**, Facioscapulohumeral muscular dystrophy: new insights from compound heterozygotes and implication for prenatal genetic counseling, *J Med Genet* 49:171-8, 2012
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50. Ricci G, Scionti I, **Tupler** R, Siciliano G. Rippling muscle disease and facioscapulohumeral dystrophy-like phenotype Response *Neuromuscul Disord.*22:670-1, 2012
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55. Feeney SJ, McGrath MJ, Sriratana A, Gehrig SM, Lynch GS, D'Arcy CE, Price JT, McLean CA, **Tupler R**, Mitchell CA. FHL1 Reduces Dystrophy in Transgenic Mice Overexpressing FSHD Muscular Dystrophy Region Gene 1 (FRG1). *PLoS One* 10:e0117665, 2015.
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64. Salsi V, Magdinier F, **Tupler R**. Does DNA methylation matter in FSHD? *Genes* 2020,
65. Ruggiero L, Mele F, Manganelli F, Bruzzese D, Ricci G, Vercelli L, Govi M, Vallarola A, Tripodi S, Villa L, Di Muzio A, Scarlato M, Bucci E, Antonini G, Maggi L, Rodolico C, Tomelleri G, Filosto M, Previtali S, Angelini C, Berardinelli A, Pegoraro E, Moggio M, Mongini T, Siciliano G, Santoro L, **Tupler R**. Phenotypic variability among Patients with D4Z4 reduced allele Facioscapulohumeral muscular dystrophy, *JAMA Open Network,* 2020 3:e204040.
66. Ricci G, Mele F, Govi M, Ruggiero L, Sera F, Vercelli L, Santoro L, Mongini T, Villa L, Moggio M, Filosto M, Scarlato M, Previtali S, Tripodi SM, Pegoraro E, Telese R, Di Muzio A, Rodolico C, Bucci E, Antonini G, D'Angelo MG, Berardinelli A, Maggi L, Piras R, Maioli MA, Siciliano G, Tomelleri G, Angelini C, **Tupler R**. Diagnostic implications for carriers of D4Z4 borderline alleles:

evidences from 244 cases of the Italian National Registry for facioscapulohumeral muscular dystrophy. *Sci Rep* 10, 21648,2020

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