MASSIMO ZEVIANI - CURRICULUM VITÆ

Interessi professionali

- Fisiopatologia muscolare
- Biochimica e genetica del metabolismo muscolare
- Genetica dei disturbi mitocondriali e metabolismo energetico
- Genetica delle malattie neurodegenerative ereditarie
- Biologia e fisiopatologia dei Mitocondri
- Malattie mitocondriali e patologie correlate

Dati personali

Data di nascita: 5 novembre 1955

Luogo di nascita: Genova (Italia)

Cittadinanza: italiana

Stato civile: coniugato, 3 figli

Servizio militare

Scuola di Medicina Militare, Firenze: maggio-luglio 1982. Agosto 1982- agosto 1983, Treviso: Sottotenente Medico dell'Esercito Italiano

Formazione scolastica

- Diploma di maturità scientifica ("Lyceum Classicum"): Padova, 1974
- Università di Padova - Facoltà di Medicina: Laurea in Medicina e Chirurgia: 25 luglio 1980.
- Università degli Studi di Padova - Scuola di Specializzazione in Endocrinologia: Specializzazione in Endocrinologia: immatricolato il 30 luglio 1983.
- Università degli Studi di Verona - Scuola di Specializzazione in Neurologia: Specializzazione in Neurologia: iscritto il 26 luglio 1989.
- Università di Parigi - Dottorato in Genetica: registrato il 25 febbraio 1997.

Ricerca ed esperienza professionale

1976-1977 Istituto di Patologia Generale, Università di Padova.

Sotto la supervisione del Prof. Alfredo Margreth ha appreso la biochimica sperimentale delle proteine, l'enzimologia e l'immunodetezione, nonché i principi di base dell'istopatologia. Ha poi partecipato a studi

sulla fosforilazione delle vescicole del reticolo sarcoplasmatico relative all'accoppiamento eccitazione-contrazione nel muscolo scheletrico.

1978-1979 Istituto di Semeiotica Medica, Università di Padova.
Sotto la supervisione del Prof. Cesare Scandellari ha partecipato a studi sulla carenza di carnitina nell'uomo e negli animali da esperimento (ratti). Ha studiato medicina clinica come tirocinante universitario.

1979-1980 Istituto di Patologia Generale, Università di Padova.
Ha svolto attività di ricerca per la tesi finale richiesta per la laurea in Medicina e Chirurgia. Sotto la supervisione dei Proff. Alfredo Margreth e Giovanni Salviati, hanno studiato i cambiamenti delle proteine contrattili e l'assorbimento del calcio nel reticolo sarcoplasmatico indotti dall'ipotiroidismo sperimentale nei conigli.

1981-1983 Per sei mesi dopo la laurea ottiene una borsa di studio universitaria come miglior dottorando in Medicina, che gli ha permesso di continuare la sua esperienza clinica presso l'Istituto di Semeiotica Medica dell'Università di Padova. Successivamente è stato arruolato come Resident in Endocrinologia, partecipando attivamente alla gestione clinica dei pazienti affetti da condizioni endocrinologiche o patologie pertinenti alla medicina interna. Nei tre anni successivi ha svolto la Residenza presso l'Istituto di Semeiotica per la Scuola di Specializzazione in Endocrinologia; parallelamente ha ottenuto un posto di Medico di Guardia Medica Festiva e Notturna presso il Pronto Soccorso dell'Ospedale Generale di Schio (provincia di Vicenza) e ha proseguito un programma di ricerca con l'Istituto di Patologia Generale, relativo agli aspetti endocrinologici della plasticità muscolo scheletrica. Come risultato di questo lavoro, in collaborazione con il Prof. Giovanni Salviati ha esteso gli studi sugli effetti dell'ipo e dell'ipertiroidismo alle singole fibre muscolari umane "skinned". Ha inoltre partecipato a lavori di ricerca sul metabolismo intermedio in collaborazione con il Prof.Cesare Scandellari e Giovanni Federspil, e sulla rilevazione di autoanticorpi nell'ipotiroidismo idiopatico mediante l'allora nuova metodica basata su ELISA. Durante il suo servizio militare, è stato nominato Responsabile della Infermeria di una grande caserma militare (Caserma Postumia) a Treviso, Italia.

1984-1985 College of Physicians & Surgeons della Columbia University, New York: Post-doctoral Fellow della Muscular Dystrophy Association of America, presso il H. Houston Merritt Clinical Research Center for Muscular Dystrophy and Related Diseases, diretto dal Prof.Salvatore DiMauro. Ha condotto studi sugli aspetti biochimici delle encefalomiopatie mitocondriali, un gruppo di disturbi neurologici causati da difetti della catena respiratoria mitocondriale. Ha definito un nuovo metodo di purificazione della citocromo c ossidasi umana (COX). Ha identificato e caratterizzato diversi disordini umani ereditari dovuti a difetti della COX e di altri complessi respiratori, analizzati mediante tecniche enzimologiche, immunologiche ed elettroforetiche.

1986-1988 Scienziato di ricerca post-dottorato del H. Houston Merritt Center. Sotto la guida del Dr. Eric A. Schon, ha appreso i principi e le tecniche della biologia molecolare. Ha partecipato alla clonazione e caratterizzazione del gene della transtiretina, responsabile della amiloidosi neuropatica negli esseri umani. Pur continuando ad aiutare nella caratterizzazione delle caratteristiche cliniche e biochimiche di nuove malattie mitocondriali ha clonato e caratterizzato il primo gene nucleare umano che codifica per una subunità della citocromo c ossidasi, subunità COX-IV, ed ha escluso l'ipotesi che la nebulina potesse essere il gene recentemente scoperto responsabile della DMD, isolando il gene corrispondente e mappandolo sul cromosoma 2, insieme alla titina. In un programma volto a identificare e vagliare le subunità codificate dal nucleo della COX, ha poi clonato il gene umano COX-Vb e ha partecipato alla clonazione di altre subunità COX in collaborazione con altri borsisti post-dottorato. La scoperta delle prime mutazioni del mtDNA lo ha spinto a caratterizzare le delezioni del mtDNA in pazienti con miopatia mitocondriale e ha dimostrato per la prima volta che la sindrome di Kearns-Sayre e la PEO sporadica sono specificamente associati a questo tipo di lesioni molecolari. In collaborazione con Eric Schon et al, ha dimostrato che le delezioni si verificano attraverso ripetizioni dirette e ha ipotizzato un meccanismo molecolare per la generazione di tali lesioni **(Schon EA, Rizzuto R, Moraes CT, Nakase H, Zeviani M, DiMauro S. A direct repeat is a hotspot for large-scale deletion of human mitochondrial DNA. Science 1989;244(4902):346-349)**. Inoltre ha contribuito alla caratterizzazione molecolare della malattia di McArdle ed è coautore di revisioni sui disturbi mitocondriali. 1989, Ricercatore presso il Dipartimento di Biochimica e Genetica dell'Istituto Neurologico "C. Besta" di Milano, diretto dal Prof. Stefano Di Donato. Ha organizzato un piccolo laboratorio di biologia molecolare dedicato alla caratterizzazione dei disturbi mitocondriali. È stato anche coinvolto nella caratterizzazione del deficit di fumarasi e MERRF. Studiando famiglie con PEO trasmessa come tratto dominante, ha scoperto che i pazienti specificamente accumulano delezioni multiple del mtDNA, proponendo per la prima volta l'esistenza di difetti della comunicazione intergenomica nucleo-mitocondriale. Ha inoltre condotto diversi studi sul metabolismo della carnitina, e ancora sulla nebulina.

1990-1993 Assistente di Neurologia presso il Dipartimento di Biochimica e Genetica dell'Istituto Neurologico "C. Besta" di Milano, diretto dal Prof. Stefano DiDonato. Direttore del Laboratorio di Patologia Molecolare. Identificate nuove mutazioni puntiformi del mtDNA associate a diverse caratteristiche cliniche, ad es. cardiomiopatia e riarrangiamenti su larga scala del DNA mitocondriale che portano a disturbi umani. Identificati nuovi geni candidati come potenziali responsabili di disturbi mitocondriali dovuti alla compromissione del cross-talk nucleo-mitocondri, ad esempio la proteina di legame del DNA a filamento singolo mitocondriale. Completata, in collaborazione con altri, la caratterizzazione di ulteriori geni COX. Ha partecipato a numerosi studi sulla caratterizzazione molecolare e clinica delle sindromi neurologiche associate al mtDNA, in particolare MERRF e Kearns-Sayre s, e sulla valutazione molecolare e biochimica della malattia di Parkinson idiopatica.

1993-1996 Associato di Neurologia. Ha continuato la caratterizzazione di nuove mutazioni e geni associati a disturbi mitocondriali e ha messo a punto la tecnica dei cybridi transmitochondriali per studiare le mutazioni del mtDNA.

Nel 1993-1994 ha trascorso un anno sabbatico presso l'INSERM Unit 393 diretto dal Prof. Arnold Munnich a l'Hôpital Necker-Enfants Malades di Parigi, dove ha collaborato con la Dott.ssa Judith Melki all'identificazione prima di delezioni della regione SMA (**Melki J, Lefebvre S, Burglen L, Burlet P, Clermont O, Millasseau P, et al. De novo and inherited deletions of the 5q13 region in spinal muscular atrophies. Science 1994;264(5164):1474-1477**) e poi dell'SMN, il gene responsabile dell'atrofia muscolare spinale (SMA) (**S Lefebvre, L Bürglen, S Reboullet, O Clermont, P Burlet, L Viollet, B Benichou, C Cruaud, P Millasseau, M Zeviani, et al. Identification and characterization of a spinal muscular atrophy-determining gene. Cell 1995;80(1):155-165**). Tornato a Milano, ha pubblicato numerosi articoli sull'identificazione e la caratterizzazione di nuovi fenotipi, mutazioni e geni candidati, compreso l'mtRNA polimerasi umana, finora il più elusivo e il più ricercato gene tracrizionale mitocondriale, in malattie mitocondriali, ed effettuato ulteriori studi sulla caratterizzazione del gene SMN nei neuroni.

1996-1997 Ha ottenuto la nomina a Direttore di una nuova Unità di Medicina Molecolare presso l'Ospedale Pediatrico "Bambino Gesù" di Roma (dal 01/04/1996 fino al 05/12/1997), dove ha istituito un laboratorio di biochimica, biologia molecolare e morfologia muscolare dedicato ai dosaggi metabolici di neonati e bambini compresi quelli mitocondriali, e disturbi ceroido-lipofuscinosi e atassie infantili. È stato inoltre nominato Neurogenetista presso la Casa Sollievo della Sofferenza a San Giovanni Rotondo (Foggia, Italia), dove ha collaborato con il Prof. Paolo Gasparini in studi di linkage e di associazione su sclerosi multipla, malattie mitocondriali e malattie neurodegenerative ereditarie. In collaborazione con Andrea Ballabio (TIGEM) ha trovato papraplegina, un componente della via del controllo della qualità delle proteine mitocondriali, responsabile dell'SPG7, una forma recessiva di paraplegia spastica ereditaria (**Casari G, De Fusco M, Ciarmatori S, Zeviani M, Mora M, Fernandez P, et al. Spastic paraplegia and OXPHOS impairment caused by mutations in paraplegin, a nuclear-encoded mitochondrial metalloprotease. Cell 1998;93(6):973-983**). 1998-2001 Accetta l'offerta per diventare Direttore dell'Unità di Biochimica e Genetica, presso l'Istituto Neurologico "C. Besta" di Milano. Nel 1998 scoprì Surf1 come il primo fattore di assemblaggio ad essere mutato nella sindrome di Leigh associata al deficit di COX (**Tiranti V, Hoertnagel K, Carrozzo R, Calimberti C, Munaro M, Granatiero M, et al. Mutations of SURF-1 in Leigh disease associated with cytochrome C oxidase deficiency. Am J Hum Genet 1998;63(6):1609-1621**), e caratterizzò ulteriormente questa proteina in pazienti e condizioni normali, così come in lievito. Sono state scoperte e caratterizzate più mutazioni del mtDNA in quel periodo di tempo associate a un ampio spettro di presentazioni cliniche. In collaborazione con Anu Suomalainen, Helsinki Università, Finlandia, ha scoperto il primo gene mutante associato a PEO autosomica dominante, ANT1. La prima mutazione nella subunità 18kDa del complesso I è stata anche trovata e

caratterizzata in un neonato, insieme a variazioni di sequenza in NDUFA1, un'altra subunità del complesso I. Altri interessi di quel periodo comprendevano la caratterizzazione delle mutazioni SOD1 in FALS, l'epilessia nei disturbi mitocondriali, il tracciamento genealogico delle mutazioni patogene del mtDNA, e il contributo alle linee guida europee sui disturbi neurologici ereditari. Nel 2001, un secondo gene responsabile dell'AD-PEO, Twinkle, l'mtDNA helicase, è stato scoperto da Massimo Zeviani in collaborazione con Hans Spelbrink e Howy Jacobs, Università di Tampere, Finlandia (**Spelbrink JN, Li F-, Tiranti V, Nikali K, Yuan Q-, Tariq M, et al. Human mitochondrial DNA deletions associated with mutations in the gene encoding Twinkle, a phage T7 gene 4-like protein localized in mitochondria. Nat Genet 2001;28(3):223-231**).

2001-2013 Diventa Direttore di una nuova Unità di Neurogenetica Molecolare presso l'Istituto Neurologico "C. Besta" di Milano. Le indagini sui pazienti sono proseguite, permettendoci di scoprire nuove alterazioni del mtDNA o DNA nucleare, mutazioni correlate a OXPHOS che sono state caratterizzate in vitro, in vivo e attraverso la creazione di modelli animali, integrando questa attività sperimentale con la caratterizzazione dei fenotipi clinici, radiologici e biochimici nei pazienti. La scoperta di pol-A come uno dei principali geni malattia nei disturbi mitocondriali, ci ha spinto a procedere a un'indagine sistematica sulle caratteristiche fenotipiche e biochimiche delle condizioni associate alle mutazioni di pol-A, che ha prodotto una serie di numerosi articoli che illustrano lo spettro di presentazioni cliniche e biochimiche associate a mutazioni in questo gene (**Graziewicz MA, Longley MJ, Bienstock RJ, Zeviani M, Copeland WC. Structure-function defects of human mitochondrial DNA polymerase in autosomal dominant progressive external ophthalmoplegia. Nat Struct Mol Biol 2004;11(8):770-776**). Allo stesso tempo, abbiamo dedicato un grande sforzo alla creazione di modelli animali per OXPHOS geni della malattia e per la scoperta di ulteriori nuovi geni codificati nel nucleo associati alla disfunzione mitocondriale. I risultati di questo lavoro sono stati la scoperta della mutazione PUS1, EFTu ed EFG1 nei difetti di traduzione del mtDNA, ETHE1 come gene responsabile dell'encefalopatia etilmalonica, di Mpv17 come responsabile di una sindrome da deplezione del mtDNA epatocerbrale, la caratterizzazione di MR-1 nella discinesia non chinesigenica, FASTKD2 come responsabile di un'encefalopatia con deficit di COX, SDHAF1 come primo fattore di assemblaggio del complesso II, ancora responsabile di una leucoencefalopatia infantile, AIF, fattore induttore di apoptosi, come responsabile di un'encefalomiopatia infantile mitocondriale legata all'X. Lo sforzo di creare modelli in vivo di disturbi OXPHOS ci ha portato ad ottenere un modello murino Surf1 KO, un modello murino Mpv17 KO un modello murino SCO2KO / KI (in collaborazione con EA Schon), e diversi modelli aggiuntivi in topi e mosche che sono stati in parte pubblicati e in parte sono ancora oggetto di indagine attiva.

Un topo ricombinante ETHE1 KO ci ha permesso di sezionare il difetto molecolare dell'encefalopatia etilmalonica e di comprendere il ruolo del prodotto del gene mutante. Le intense collaborazioni con Ileana Ferrero e il suo gruppo sulla modellazione in lievito dei disordini mitocondriali umani, e con Rodolfo Costa per i modelli di mosca, ci hanno entrambe portato ad ampliare lo spettro dell'indagine sperimentale in vivo sulla patogenesi di una serie di difetti OXPHOS dovuti a mutazioni geniche nucleari.

Allo stesso tempo, abbiamo aperto un nuovo campo di indagine sulla caratterizzazione molecolare dei disturbi del movimento ereditari nell'infanzia e dei pazienti adulti, compreso il morbo di Parkinson.
Collaborazioni consolidate hanno portato ad una serie di interessanti pubblicazioni congiunte sulla caratterizzazione ed epidemiologia della Neurodegenerazione Ottica Ereditaria di Leber (con Valerio Carelli e Patrick Chinnery), ulteriore definizione fenotipica di mutazioni pol-A (con Laurence Bindoff), assemblaggio di supercomplessi respiratori e complesso III in particolare (con Tonio Enriquez) o l'uso di ossidasi alternative in modelli di mosca di deficit di COX (con Howy Jacobs). Il sequenziamento dell'esoma è stato utilizzato in collaborazione con Holger Prokisch e Thomas Meitinger a Monaco (Germania) per identificare nuovi geni, tra cui ACAD9, un fattore di assemblaggio del complesso I (**Haack TB, Danhauser K, Haberberger B, Hoser J, Strecker V, Boehm D, et al. Exome sequencing identifies ACAD9 mutations as a cause of complex i deficiency. Nat Genet 2010;42(12):1131-1134**) e, indipendentemente, TTC19, un fattore di assemblaggio del complesso III. Lo screening genetico di un singolo iniziale paziente nel nostro laboratorio ha portato alla identificazione di mutazioni in FBXL4, una E3 ligasi della membrana esterna dei mitocondri la cui funzione ne regola la biogenesi (**Gai X, Ghezzi D, Johnson MA, Biagosch CA, Shamseldin HE, Haack TB, et al. Mutations in FBXL4, encoding a mitochondrial protein, cause early-onset mitochondrial encephalomyopathy. Am J Hum Genet 2013;93(3):482-495**). Mutazioni in una singola famiglia norvegese di PTRM1, una proteasi della matrice mitocondriale, ci ha portato ad evidenziare l'accumulo del recursore della beta amiloide e di peptidi di beta amiloide stessa nelle cellule mutanti sia umane che di un modello murino, suggerendo un'interessante connessione tra disturbi della proteostasi mitocondriale e neurodegenerazione (**Brunetti D, Torsvik J, Dallabona C, Teixeira P, Sztromwasser P, Fernandez-Vizarra E, et al. Defective PITRM1 mitochondrial peptidase is associated with Aβ amyloidotic neurodegeneration. EMBO Mol Med 2016;8(3):176-190**) (**Pérez MJ, Ivanyuk D, Panagiotakopoulou V, Di Napoli G, Kalb S, Brunetti D, et al. Loss of function of the mitochondrial peptidase PITRM1 induces proteotoxic stress and Alzheimer's disease-like pathology in human cerebral organoids. Mol Psychiatry 2020**). Analogamente, lo studio NGS di un singolo paziente ci ha portato alla scoperta di mutazioni di RNAseH1, che sono poi state trovate in numerosi altri casi (**Reyes A, Melchionda L, Nasca A, Carrara F, Lamantea E, Zanolini A, et al. RNASEH1 Mutations Impair mtDNA Replication and Cause Adult-Onset Mitochondrial Encephalomyopathy. Am J Hum Genet 2015;97(1):186-193**). Abbiamo recentemente aperto una nuova attività finalizzata allo sviluppo della terapia per la malattia mitocondriale, inclusa la correzione farmacologica dell'encefalopatia etilmalonica e l'induzione della biogenesi mitocondriale e OXPHOS in modelli animali di carenza di COX. Lo scereening genetico profondo ad alta efficienza è stato allestito in situ presso l'Istituto Neurologico Carlo Besta.

Oltre alle attività di ricerca, ci siamo impegnati anche ad offrire un ampio e complesso pannello di saggi biochimici e molecolari per la diagnosi dei disturbi mitocondriali e del movimento, nonché un'assistenza clinica per pazienti ambulatoriali e ricoverati con sospetta disfunzione metabolica e mitocondriale. L'Unità diretta da Massimo Zeviani è stata composta da 3 membri dello Staff del Dottorato, 2 membri dello Staff MD

(incluso MZ), 3 membri dello Staff Tecnici, 5 Tecnici esterni al personale, 4 dottorandi, 4 dottorandi e 2 studenti universitari. Il laboratorio diagnostico offre diagnosi biochimiche e genetiche molecolari di disturbi mitocondriali, disturbi del movimento (ad es. Analisi HPLC di neurotrasmettitori nel liquido cerebrospinale per distonia genetica; geni associati alla malattia di Parkinson, ecc.). Abbiamo organizzato una clinica (una volta alla settimana) per pazienti con malattie mitocondriali e consulenza genetica e abbiamo accesso a letti nel servizio di day-hospital e nelle Divisioni di Neurologia Clinica (adulti e bambini) per indagare su pazienti con sospetta disfunzione mitocondriale. La sezione di ricerca del laboratorio comprendeva due dottorandi membri del personale, due tecnici membri del personale, dottorandi e studenti laureati e universitari. Le principali linee di indagine riguardano: (a) la ricerca genica e la caratterizzazione delle proteine malattia nei disturbi mitocondriali; (b) studi di fisiopatologia e terapia sperimentale sui disturbi mitocondriali nei sistemi cellulari (l'Unità controlla una biobanca di fibroblasti cutanei, una banca del DNA e ha accesso alla biopsia muscolare e alle biobanche di cellule linfoblastoidi dell'Istituto); (c) creazione e studio della fisiopatologia e terapia sperimentale sui disturbi mitocondriali in modelli murini; (d) proteine in vitro e analisi trascrizionale delle malattie mitocondriali; (e) enzimologia e bioenergetica dei disturbi mitocomdriali. Inoltre, l'Unità è stata coinvolta in studi di associazione sulla base genetica del morbo di Parkinson e ha avviato un progetto (Group Leader Valeria Tiranti, PhD) sulla patogenesi molecolare e cellulare delle malattie degenerative da accumulo di ferro cerebrale (es. Malattia di Hallervorden-Spatz). Una struttura per la cura degli animali è stata gestita da uno dei membri dello Staff del Dottorato (Carlo Viscomi, PhD).
Per una descrizione più completa delle attività e delle iniziative di ricerca del nostro centro si rimanda all'elenco delle pubblicazioni o la visita del nostro sito www.mitopedia.org.

2013-2019. Il 14 gennaio 2013 Massimo Zeviani ha iniziato un nuovo lavoro come Direttore dell'Unità di Biologia Mitocondriale (Mitochondrial Biology Unit, MBU) presso l'Università di Cambridge, nella quale è stato nominato Professore Ordinario di Medicina Mitocondriale. È succeduto nella carica di direttore della MBU al Prof. Sir John Walker, Premio Nobel. La sua missione principale, accettata dopo un bando internazionale competitivo lanciato per identificare il nuovo direttore della MBU, da parte di una commissione formata da un consiglio internazionale di scienziati insieme a diversi membri dello staff dirigenziale dell'MRC, tra cui Sir John Savill, presidente dell'MRC, era di rimodulare l'Unità verso una missione più traslazionale, che potesse comprendere, insieme allo studio dei fondamenti della biologia mitocondriale, anche la sua connessione con l'area in espansione crescente delle malattie mitocondriali e dell'impatto che le disfunzioni mitocondriali hanno in processi cronici degenerativi quali il morbo di Parkinson, la Sclerosi Laterale Amiotrofica, la malattia di Alzheimer, e lo stesso invecchiamento. A tal fine, ha reclutato su base competitiva il dottor Alex Whitworth, PhD, esperto in neurodegenerazione associata a disfunzione mitocondriale in *Drosophila melanogaster*, e il dottor Julien Prudent, PhD, esperto in mitodinamica e caratterizzazione strutturale del compartimento interno dei mitocondri. Ha anche fondato il proprio laboratorio, Mitochondrial Medicine, dedicato allo studio dei disturbi mitocondriali, alla modellazione in vivo

e allo sviluppo di strategie terapeutiche (quest'ultimo progetto è stato sostenuto da una sovvenzione ERC avanzata vinta dal dott. Zeviani di 2,5 m € in 5 anni iniziata a metà 2013). Allo stesso tempo, ha deciso di ospitare nell'Unità il Prof. Patrick Chinnery in un gruppo chiamato Genomica Mitocondriale. Patrick Chinnery si è trasferito dalla Newcastle University come nuovo Direttore delle Neuroscienze Cliniche dell'Università di Cambridge. Ha anche rimosso il dottor Ian Holt, la dott.ssa Antonella Spinazzola e il dottor Leonid Sazanov come capogruppi dell'MBU per un irrisolvibile problema di conflitto di interessi e, con l'aiuto dell'MRC, ha attribuito una posizione indipendente con una borsa di studio separata dal budget dell'MBU al Prof. Sir John Walker. Una volta che l'MRC-MBU è entrato a far parte dell'Università di Cambridge nel 2016, ha proposto una cattedra da assegnare al Dr. Mike Murphy, al Dr. Judy Hirst e al Dr. Edmund Kunji. Tutte e tre le proposte sono state accettate. Durante i quasi sette anni del suo mandato, in cui è stata raggiunta con successo la valutazione e il relativo finanziamento di 28 milioni di sterline associato alla Quinquennial Review, l'Unità ha ottenuto risultati scientifici molto significativi tra cui la completa risoluzione atomica mediante cryo-EM del Complesso Bovino I (Prof. Judy Hirst), la struttura e la funzione del traslocatore nucleotidico dell'ADP/ATP dei mitocondri (Prof. Edmund Kunji), l'identificazione di diversi nuovi geni nelle malattie mitocondriali (tra cui COA7, COA8, varie subunità della citocromo ossidasi e del complesso I, FBXL4, TTC19, etc.) e lo sviluppo di terapie farmacologiche e geniche contro queste malattie (Massimo Zeviani), la caratterizzazione fisica e funzionale del collo di bottiglia genetico selettivo mitocondriale nei gameti femminili e dell'impatto delle mutazioni somatiche del mtDNA sulla salute umana (Patrick Chinnery), lo sviluppo di nuove modificazioni antiossidanti e post-traduzionali delle proteine associate allo stress ossidativo (Mike Murphy), l'editing mediante sofisticati strumenti di biologia molecolare e cellulare di mutazioni patogene nel mtDNA in vivo e la scoperta di diversi nuovi componenti del meccanismo di espressione mitocondriale (Michal Minczuk), il legame tra deterioramento neurodegenerativo e diverse anomalie della bioenergetica mitocondriale (Alex Whitworth), la scoperta di nuovi fattori associati alla mitodinamica e ai processi adattivi bioenergetici (Julien Prudent). Pubblicazioni di questi risultati sono apparsi su Nature, Science, PNAS, EMBO Journal, Cell, e altre riviste di alto impatto scientifico. La MBU è attualmente considerata la struttura di maggior prestigio, produttività ed eccellenza scientifica del mondo per la fisiopatologia mitocondriale. La proteomica, la metabolomica e la bioinformatica sono state ulteriormente sviluppate e organizzate come strutture comuni per lo sviluppo dei progetti scientifici dell'Unità. La MBU, sotto la direzione di Massimo Zeviani, comprendeva 8 gruppi di ricerca coordinati da altrettanti direttori di ricerca, un centinaio di scienziati in staff, una trentina di studenti di PhD e circa 40 post doc, e inoltre un piccolo ma efficiente gruppo di personale amministrativo.

Nel 2019 Massimo Zeviani ha accettato la proposta dell'Università degli Studi di Padova, Italia, di essere nominato professore di Neurologia presso l'Università locale per meriti scientifici, ed è stato arruolato stabilmente come docente a tempo pieno dell'Università di Padova da Ottobre 2019 ad oggi. Nell'anno accademico 2019-2020, facendo parte del Dipartimento di Neuroscienze, ha tenuto corsi di Neurologia,

Fisiologia umana e Patologia generale a studenti universitari. È membro di facoltà del dottorato in medicina traslazionale dell'Università degli studi di Padova.

Ulteriori esperienze di insegnamento

1981-1982 Istituto di Semeiotica Medica, Università di Padova. Ha insegnato esame fisico a studenti di medicina del 3 ° anno. 1983 Istituto di Patologia Generale, Università di Padova. Ha insegnato istopatologia a studenti di medicina del 3 ° anno. 1990-95 Scuola Europea di Genetica Medica, III Corso. Conferenza sulla genetica del mtDNA e la relativa patologia umana. 1993-1995 Professore presso la Scuola di Specializzazione in Genetica Medica dell'Università degli Studi di Milano (Professore a contratto) 1996 Scuola Europea di Genetica Medica, III Corso. Conferenza sulla genetica del mtDNA e la relativa patologia umana. 1998 FEBS corso avanzato in Fosforilazione Ossidativa. 2002 Corso di Neurogenetica a studenti della Facoltà di Medicina e Chirurgia dell'Università di Padova 2007-2008 Corso di Medicina Mitocondriale a studenti universitari dell'Istituto Univesitario Studi Superiori, Facoltà di Medicina dell'Università di Pavia Ha tenuto numerosi seminari e conferenze (> 500) in congressi nazionali e internazionali e in diverse istituzioni universitarie in Italia, Europa, Giappone, Canada, Brasile, Argentina, Arabia Saudita, Israele e Stati Uniti. Opponente per tesi di dottorato in Genetica molecolare presso l'Università di Helsinki (1993, Anu Suomalainen), l'Università di Parigi (1994, Thomas Bourgeron), l'Università di Glasgow (1994, Fiona Reid), il Karolinska Institutet (2000, Jang-Minh), l'Università di Tampere (2001, Olli Kajander); Karolinska Institutet (2005, Matt Ekstrand); Università di Maastricht (2005; Bianca van der Bosch), etc, etc. oltre a numerosi dottorandi in Cambrdige, Oxford, Newcastle, UCL, Harwell, ed altre prestigiose Università dell'UK. Sotto la sua supervisione numerosi studenti si sono laureati o dottorati in Biologia, Biotecnologie e Medicina svolgendo lavori sperimentali per la tesi finale.

Onori

- Laurea in Medicina e Chirurgia "magna cum laude", Università di Padova, luglio 1980.
- Specializzazione in Endocrinologia "magna cum laude", Università di Padova, luglio 1983
- Specializzazione in Neurologia "magna cum laude", Università degli Studi di Verona, luglio 1989
- Premio di dottorato "magna cum laude", Università di Parigi, febbraio 1997
- Premio Giovane Ricercatore al 1 ° Congresso Internazionale su "Genetica molecolare delle malattie neurologiche e neuromuscolari", Saint Vincent, settembre 1986
- Premio "Valigia dell'Intelletto" di A.R.I.N. (Associazione Italiana per la Promozione della Ricerca neurologica) for Clinical Research in Neurology, gennaio 1986

- Premio Giovane Ricercatore al 4 ° Congresso Nazionale della Federazione Italiana per lo Studio dei Disordini Ereditari (FISME), Milano, Settembre 1990.
- Premio Associazione Italiana Ricerche sull'Handicap (AIRH), 1991
- Premio INSERM “Post Vert” per Visiting Researchers, 1994.
- Premio "Best Presentation" al 4 ° Meeting della European Neurological Society, giugno 1995.
- Lezione commemorativa "Anita Harding", IX ° Meeting of the European Neurological Society, giugno 1998.
- Lezione sulle ultime notizie all'incontro annuale dell'American Society of Human Genetics, Denver, USA, ottobre 1998
- Premio "Brain" per la ricerca sulla neurogenetica, maggio 2000
- Premio UE “René Descartes” per la ricerca transnazionale europea, Praga, dicembre 2004
- Premio “Gaetano Conte” della Società Mediterranea di Miologia
- Riunione annuale SSIEM "The Annual George Komrower Honorary Lecture", Roma, settembre 2016
- "The George Karpati Honorary Lecture" Institute of Neurology, McGill University, Montreal, CA, maggio 2016
- Prix de la Fondation NRJ 2013 (Institut de France) Génétique des maladies dégénératives - giugno 2013
- "Adam Barski Honorary Lecture in Mitochondrial Disease" The Sick kids Hospital, University of Toronto, ON, CA, aprile 2009
- Memorial Lecture in onore di Lewis P. Rowland, Columbia University, New York, NY, 2017
- Lezione in onore della messa a riposo di Salvatore Di Mauro, New York, NY, 2019
- Responsabile del gruppo di studio della Società Italiana di Neurologia per la Neurogenetica Clinica, 2014-2018

Attività Editoriale

Co-editor di Neurological Sciences, Biochimica Biophysica Acta, Mitochondrion, Journal of Medical Genetics, Embo Molecular Medicine, Human Molecular Genetics.

Reviewer frequente di Nature, Science, Cell, Nature Genetics, Nature Medcine, Embo Molecular Medicine, EMBO Journal, PLOS Genetics, Human Molecular Genetics, Cell Report, Nature Communications, Neurology, Annals of Neurology, European Journal of Human Genetics, American Journal of Human Genetics, Gene, Nucleic Acids Research, etc. etc.

Sovvenzioni

(Mlit: milioni di lire; MEuro: milioni di euro)

- 1991-1993 Borsa Telethon n. 116, Mlit 130
- 1993-1994 Borsa Telethon n. 456, Mlit 140

- 1991-1994 Borsa di studio CEE per la stimolazione e l'azione sulla "Caratterizzazione molecolare di titina e nebulina"
- 1995-1998 Sovvenzione UE per il capitale umano e la mobilità "Biogenesi mitocondriale nello sviluppo e nella malattia"
- 1995-1998 Borsa Telethon n. 767, Mlit 330
- 1999-2001 Ricerca Finalizzata Min. San. ICS 030.3 / RF98.37 Mlit 530
- 1999-2001 Borsa Telethon n. 1180, Mlit 349
- 2000-2003 Grant Ricerca 2000 della Fondazione Pierfranco e Luisa Mariani, Mlit 1730
- 2002-2004 EU MITEURO progetto EU n. QLG1 - CT - 2001-00966, Euro 54.000
- 2002-2004 EU Gendeaf Thematic Network - Progetto Amplifon n. QLG1 - CT - 2001-01429, Euro 2.400
- 2002-2004 Min. SAL. RF 2002.128 ("S.Matteo - Pavia" - Zeviani) Euro 17.000
- 2003-2005 Telethon GGP030039, Euro 193.500
- 2003-2005 Ricerca FINALIZZATA CARIPLO - Caratterizzazione delle basi molecolari dei disordini mitocondriali Euro 90.000,00
    - 2003-2005 min. SAL. RF 2002.158 - Malattie mitocondriali: screening diagnostico ad alto rendimento di malattie e geni modulatori, Euro 115.000
- 2004-2005 Borsa di studio “Centro Malattie Mitocondriali” della Fondazione Pierfranco e Luisa Mariani, Euro 106.520
- 2004-2008 EUMITOCOMBAT CONSORTIUM - AN FP6 INTEGRATED PROJECT, MEuro 8.1, for the Unit of Molecular Neurogenetics Euro 786.800).
- 2004-2006 CARIPLO e IST. SPERIM. IT. LAZZARO SPALLANZANI: "Generazione bovina knockout per il gene PrPc (prion) per la produzione di biomateriale sicuro e prevenzione BSE" Euro 77.780
- 2004-2006 "Genetica clinica e molecolare del morbo di Parkinson e traduzione clinico-diagnostica" in collaborazione con Inst. Mendel, Roma - Casa Sollievo Sofferenza, S.G.Rotondo Euro 45.000
- 2004-2006 Ministero della Salute / Regione Puglie (MinSal 25/2003): "Disturbi metabolici e mitocondriali dovuti a geni nucleari: epidemiologia, diagnostica molecolare e fisiopatologia" Euro 20.000
- 2005-2006 Borsa di studio “Centro Malattie Mitocondriali” della Fondazione Pierfranco e Luisa Mariani, Euro 120.000
- 2006-2007 Borsa di studio “Centro Malattie Mitocondriali” della Fondazione Pierfranco e Luisa Mariani, Euro 150.000
    - 2007-2008 Borsa di studio “Centro Malattie Mitocondriali” della Fondazione Pierfranco e Luisa Mariani, Euro 150.000
- 2010-2011 AFM Grant “Strategia terapeutica per la cura dell'encefalopatia etilmalonica, una grave malattia mitocondriale infantile”. 80.000 €.
- 2008-2011 Telethon Grant “Identificazione e caratterizzazione di geni nucleari per disordini mitocondriali umani. 227.400 €.
- 2011-2013 Fondazione Telethon – Program Project - “Therapeutic strategies to combat mitochondrial disorders”" 1.235 M€.

- GGP11139A Mitochondrial Aspartate/Glutamate Carrier 1 Deficiency: Pathogenetic Mechanism and mutatio 31.900,00
- 2012-2015 - RF2010 - 2312766 - A multicenter collaborative research network for the identification and study of rare undiagnosed patients: the impact on the rare disease National Health Service network 29.975,00 €
- 2012-2014 – CARIPLO - Definition and characterization of disease genes in mitochondrial disorders 200.400,00 €
- 2012-2015 – E-Rare Mitochondrial disorders – Connecting Biobanks, Empowering Diagnostics and Exploring disease models 128.000,00 €
- 2013-2015 MEET – Marie Curie - Mitochondrial European Educational Training. 225.135,00 €.
- MITCARE: Mitochondrial Medicine: developing treatments of OxPhos-defects in recombinant mammalian models. Advanced grant dell'ERC (European Research Council) (FP7-322424) 2013-2018, 2.5 M€
- 2015-2020 Sovvenzione di base del Medical Research Council del Regno Unito (MC_UU_00015 / 5) 28 M£
- 2016-2018 COEN (EU) Mito-ND, Neurodegenerazione mitocondriale, 350.000 €
- 2020-2022 Telethon-Italia GGP19007: Terapia genica sperimentale nei disturbi mitocondriali 300.000 €
- 2019-2024 Mitofight - Associazione Renato Comini Onlus, 500.000 €

PUBBLICAZIONI SCIENTIFICHE (ottobre 2020)

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1. Salviati G, Betto R, Danieli Betto D, Zeviani M. Myofibrillar-protein isoforms and sarcoplasmic-reticulum Ca2+-transport activity of single human muscle fibres. Biochem J 1984;224(1):215-225.
2. Bresolin N, Zeviani M, Bonilla E, Miller RH, Leech RW, Shanske S, et al. Fatal infantile cytochrome c oxidase deficiency: Decrease of immunologically detectable enzyme in muscle. Neurology 1985;35(6):802-812.
3. DiMauro S, Zeviani M, Bonilla E, Bresolin N, Nakagawa M, Miranda AF, et al. Cytochrome c oxidase deficiency. Biochem Soc Trans 1985;13(4):651-653.
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5. DiMauro S, Bonilla E, Zeviani M, Nakagawa M, DeVivo DC. Mitochondrial myopathies. Ann Neurol 1985;17(6):521-538.
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7. Zaccaria M, Giordano G, Pasquali C, Ragazzi E, Zeviani M, Valentini P, et al. Effects of pirenzepine on plasma insulin, glucagon and pancreatic polypeptide levels in normal man. Eur J Clin Pharmacol 1985;27(6):701-705.

8. Betterle C, Caretto A, Zeviani M, Pedini B, Salviati C. Demonstration and characterization of anti-human mitochondria autoantibodies in idiopathic hypoparathyroidism and in other conditions. Clin Exp Immunol 1985;62(2):353-360.
9. Herbert J, Wilcox JN, Pham K-C, Fremeau Jr. RT, Zeviani M, Dwork A, et al. Transthyretin: A choroid plexus-specific transport protein in human brain. The 1986 S. Weir Mitchell Award. Neurology 1986;36(7):900-911.
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