CURRICULUM VITAE VALERIO CARELLI

Dati personali

Nato il 08/08/1959 ad ANCONA (AN)

Codice fiscale CRLVLR59M08A271J

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Indirizzo abitazione: Via Mascarella 6, CAP 40126, Bologna, Italia

Madrelingua italiano, inglese parlato fluentemente

Formazione

1990 – Laurea in Medicina e Chirurgia, Università di Bologna. **Voto finale: 110/110 con lode**. Tesi: "***Neuropatia Ottica Ereditaria di Leber: studio familiare tramite polimorfismi di restrizione del DNA***"

1990/1994 – Specializzazione in Neurologia, Università di Bologna. **Voto finale: 70/70 con lode**. Tesi: "***Neuropatia Ottica Ereditaria di Leber: analisi molecolare e biochimica delle subunita' mitocondriali della NADH deidrogenasi***".

1994/1998 – Conseguimento del Dottorato in Neuroscienze, Università di Verona. Tesi: "***Neuropatia Ottica Ereditaria di Leber: studio della patogenesi a livello biochimico, cellulare e morfologico***".

Esperienza professionale

1998 – 2001: sotto la supervisione del Prof. Alfredo A. Sadun, Research Associate presso il Laboratory of Ocular Neuropathology (Sadun's lab), Doheny Eye Institute, Keck School of Medicine - University of Southern California, Los Angeles, California, USA.

2001 – 2014: ricercatore a tempo pieno con compiti assistenziali (confermato dal 2004) presso il Dipartimento di Scienze Neurologiche (2001 – 2012) e il Dipartimento di Scienze Biomediche e NeuroMotorie (DIBINEM) (2012 – 2014), dell'Università di Bologna

2001 – presente: co-Principal Investigator con il Prof. Alfredo A. Sadun (Doheny Eye Institute) del progetto di ricerca multinazionale "LHON Brazil" in collaborazione con il Department of Ophthalmology, Federal University of São Paulo (UNIFESP), São Paulo, Brazil.

2003 – 2013: Research Assistant Professor of Neuro-Ophthalmology, Voluntary Faculty presso il Doheny Eye Institute, Keck School of Medicine of the University of Southern California, Los Angeles, California, USA

2005 – presente: dirigente medico della UOC Clinica Neurologica AUSL Bologna e responsabile del Laboratorio di Neurogenetica.

2012 – presente: alta specializzazione professionale (AUSL Città di Bologna) afferente alla UOC Clinica Neurologica e all'IRCCS Istituto delle Scienze Neurologiche di Bologna, Ospedale Bellaria.

2013 – abilitazione nazionale per professore di I e II fascia nel settore 06/D6 (Neurologia) e professore di II fascia nel settore 06/A1 (Genetica Medica).

2014 – presente: Professore Universitario di ruolo - seconda fascia – Dipartimento di Scienze Biomediche e NeuroMotorie, 06/D6 - Settore Scientifico-Disciplinare MED/26 – Neurologia come da decreto rettorale del 15/09/2014

2018 – abilitazione nazionale per professore di I fascia nel settore 06/A1 (Genetica Medica)

2020 – presente: facente funzione direzione Programma di Neurogenetica, IRCCS Istituto delle Scienze Neurologiche di Bologna, Ospedale Bellaria

ATTIVITA' DI RICERCA E ALTRE ATTIVITA' PROFESSIONALI

Progetti di ricerca internazionali e nazionali, ammessi al finanziamento sulla base di bandi competitivi che prevedano la revisione tra pari

H2020-SC1-PHE-CORONAVIRUS-2020-2: EU proposal 101016167 – ORCHESTRA – attribuzione finanziamento 11 agosto 2020: euro 30.000.000 – durata 36 mesi, partecipante come **partner** all'unità di ricerca "WP6: Biobanking, genomics and virus-host interactions" (coordinatore Prof. Evelina Tacconelli)

Ricerca Finalizzata Ministero della Salute RF-2018-12366703 "REtinal ganglion cells and ORganoids from Inherited Optic Neuropathies: light on pathogenesis to fight blindness (REORION Project)" – attribuzione finanziamento marzo 2019: euro 450.000 - durata 36 mesi, **coordinatore (principal investigator)** di 3 unità di ricerca

PRIN 2017 prot. 20172T2MHH – "Keeping mitochondrial DNA in shape in health and disease: cracking the elusive relationship between the fusion protein OPA1, mitochondrial membrane lipid composition and maintenance of membrane-anchored mtDNA nucleoids" – attribuzione finanziamento marzo 2019: euro 436.358 – durata 36 mesi, **coordinatore (principal investigator)** di 3 unità di ricerca

Telethon Program Project: "MitCare-2" (grant GGP14187) anno 2014: euro 1.000.600 – durata 36 mesi, **responsabile di unità di ricerca** (coordinatore Prof. Luca Scorrano)

Program Project della regione Emilia-Romagna: "Recognition, diagnosis and therapy of mitochondrial disorders in neurological services of the Emilia-Romagna region" (grant ER-MITO) anno 2012: euro 1.323.300 – durata 36 mesi, **coordinatore (principal investigator)** di 6 unità di ricerca

UE FP7-PEOPLE-2012-ITN: "Mitochondrial European Educational Training - MEET" (grant 317433) anno 2012: euro 3.800.000 – durata 48 mesi, partecipante alla unità di ricerca del coordinatore (Prof. Marco Seri)

United Mitochondrial Disease Foundation (UMDF): "Estrogen mediated regulation of mitochondrial biogenesis and functions: possible therapeutic implications for Leber's hereditary optic neuropathy" (grant 12-059) anno 2012: $ 108.000 – durata 24 mesi, **responsabile di unità di ricerca** (coordinatore Dott. Carla Giordano)

Telethon: "Systematic gene hunting for nuclear modifiers in Leber's hereditary optic neuropathy and their validation in model systems" (grant GGP11182) anno 2011: euro 289.800 – durata 24 mesi, **coordinatore (principal investigator)** di 4 unità di ricerca

Ricerca Finalizzata Ministero della Salute "Ita-MNGIE: An Italian network for MNGIE epidemiology, molecular mechanisms and enzyme replacement therapy by stem cell transplant" (RF-2009-1492481) anno 2011: euro 300.000 – durata 24 mesi, **responsabile di unità di ricerca** (coordinatore Prof. Roberto De Giorgio)

Telethon Program Project: "Therapeutic strategies to combat mitochondrial disorders" (grant GPP10005) anno 2010: euro 1.250.000 – durata 36 mesi, partecipante alla unità di ricerca del coordinatore (Prof. Massimo Zeviani)

Telethon: "Constructing a database for a nation-wide Italian collaborative network of mitochondrial diseases" (grant GUP09004F) anno 2009: euro 217.100 – durata 24 mesi, **responsabile di unità di ricerca** (coordinatore Prof. Gabriele Siciliano)

EMBO Lecture course grant: "Mitochondrial medicine: from genetics to biological mechanisms and beyond" (grant EFLC09-043) anno 2009: euro 30.000 – durata 12 mesi, **organizzatore del corso**

E-Rare Project "European Research project on Mendelian Inherited Optic Neuropathies - ERMION" (grant JTC2009) anno 2009: euro 143.400 – durata 36 mesi, componente della unità di ricerca italiana coordinata dal Dott. Andrea Martinuzzi

Ricerca Finalizzata Ministero della Salute: "Mitochondrial Disorders: from medical genetics to molecular mechanisms, toward the development of therapeutic strategies" (RF-INN-2007-634163-1) anno 2008: euro 400.000 – durata 24 mesi, **responsabile di unità di ricerca** (coordinatore Prof. Massimo Zeviani)

Programma ricerca Regione-Università Progetto PERNO: "A primary brain tumours registry as a framework for promoting clinical research in Neuro-Oncology: Project of Emilia-Romagna region on Neuro-Oncology"- anno 2007: € 942.000 – durata 36 mesi, componente della unità di ricerca coordinata dal Prof. Vincenzo Eusebi.

Ricerca Finalizzata Ministero della Salute: "Disabilita' visiva nella neuropatia ottica ereditaria di Leber: dalla diagnosi precoce alla riabilitazione" (RF-FGB-2006-368547) anno 2007: euro 300.000 – durata 24 mesi, **responsabile di unità di ricerca** (coordinatore Prof. Vincenzo Parisi)

Telethon : "Pathogenic mechanisms for degeneration of retinal ganglion cells in mitochondrial optic neuropathies" (grant GGP06233) anno 2006: euro 645.800 – durata 36 mesi, **coordinatore (principal investigator)** di 6 unità di ricerca

Ricerca regione Veneto: "Strategie di prevenzione e trattamento del danno ossidativo in modelli cellulari di malattia neurodegenerativa" (grant 234/05) anno 2005: euro 50.000 – durata 24 mesi, **responsabile di unità di ricerca** (coordinatore Dott. Andrea Martinuzzi)

COFIN 2003 (ex 40%): "Studi dei meccanismi genetici e immunitari nella patogenesi dell'epilessia temporale mesiale" anno 2003: euro 154.000 – durata 24 mesi, partecipante alla unità di ricerca del coordinatore (Prof. Agostino Baruzzi)

Telethon: "Leber's hereditary optic neuropathy: systematic investigation of energy depletion and oxidative stress and their relevance to cell death, using cybrid cell lines with 11778, 3460, 14484, and 14459 mutant mitochondria" (grant GGP02323) anno 2002: euro 177.000 – durata 24 mesi, **coordinatore (principal investigator)** di 4 unità di ricerca

Telethon: "Pathogenic role and biochemical dysfunctions associated with the mtDNA ATPase6 gene mutations" (grant GP0280/01) anno 2001: Lire. 100.000.000 – durata 24 mesi, partecipante alla unità di ricerca del coordinatore (Prof. Giancarlo Solaini)

Telethon: "mtDNA encoded ATPase6 subunit: pathogenetic role of the T8993G mutation" (grant 1048) anno 1997: Lire. 46.000.000 – durata 12 mesi, partecipante alla unità di ricerca del coordinatore (Prof. Giancarlo Solaini)

Telethon: "In vitro phenotypic expression of mtDNA point mutations associated with Leber's hereditary optic neuropathy (LHON)" (grant 792) anno 1996: Lire. 100.000.000 – durata 24 mesi, partecipante alla unità di ricerca del coordinatore (Prof. Elio Lugaresi)

Telethon: "Elucidation of the biochemical defects in human diseases associated with missense point mutations of mitochondrial DNA: a basis for therapeutic strategies" (grant 616) anno 1995: Lire. 40.000.000 – durata 12 mesi, partecipante alla unità di ricerca del coordinatore (Dott. Mauro degli Esposti)

Telethon: "Molecular-biochemical pathophisiology of mitochondrial DNA mutations associated with Leber's hereditary optic neuropathy (LHON)" (grant 391) anno 1993: Lire. 50.000.000 – durata 12 mesi, partecipante alla unità di ricerca del coordinatore (Prof. Elio Lugaresi)

Altri finanziamenti per la ricerca (unrestricted grants) da industrie o fondazioni, e donazioni

Finanziamento Stealth Bio Therapeutics Corp.: “Screening the effects of MTP-131 (Bendavia) on cell models of Leber’s hereditary optic neuropathy (LHON) and dominant optic atrophy (DOA)” anno 2017, euro 150.000 – durata 24 mesi, **coordinatore (principal investigator)** del progetto

Finanziamento MITOCON: “Dissecting the mechanism of Idebenone cytoprotection to understand its efficacy in mitochondrial complex I diseases”, anno 2017, euro 35.000 – durata 24 mesi, **coordinatore (principal investigator)** del progetto

Donazione Famiglia Resta: progetto “Studio del gene causativo della forma di atrofia ottica denominata OPA8”, anno 2017, euro 30.000 – durata 24 mesi, **coordinatore (principal investigator)** del progetto

Donazione Famiglia Ravaglia: progetto “Screening di farmaci attivatori della biogenesi mitocondriale su modelli cellulari di MERRF: rapida trasferibilità sul paziente”, anno 2014, euro 60.000 – durata 36 mesi, **coordinatore (principal investigator)** del progetto

Finanziamento Fondazione Galletti: "Individuazione dei fattori predittivi per la conversione da deficit cognitivo lieve a demenza" anno 2010, euro 43.750 – durata 36 mesi, **coordinatore di unità di ricerca**

Finanziamento Edison Pharmaceuticals - Columbia University, New York City, subcontract: “Testing new molecules on mitochondrial dysfunction in cybrids" anno 2008, $50.000 – durata 12 mesi, **coordinatore (principal investigator)** del progetto in Italia

Finanziamento Edison Pharmaceuticals - Columbia University, New York City, subcontract: "Testing new molecules on mitochondrial dysfunction in cybrids" anno 2007, $55.000 – durata 12 mesi, **coordinatore (principal investigator)** del progetto in Italia

Finanziamento Pfizer: "Cybrids as a Tool to Investigate the Influence of Mitochondrial DNA Haplogroups on Cellular Sensitivity to Zyvox (Linezolid-Oxazolidinone Class of Antibiotics)" anno 2005, $ 103.000 – durata 24 mesi, **coordinatore (principal investigator)** del progetto

Responsabile di Trial clinici

GenSight Pharmaceuticals, trial di terapia genica in doppio-ceco, placebo controllato, randomizzato GS-LHON-CLIN-05 REFLECT (inzio reclutamento aprile 2019): "Efficacy and Safety of Bilateral Intravitreal Injection of GS010: A Randomized, Double-Masked, Placebo-Controlled Trial in Subjects Affected with G11778A *ND4* Leber Hereditary Optic Neuropathy for Up to One Year" (approvazione del comitato etico protocollo

Santhera Pharmaceuticals, trial farmacologico con Idebenone (3 anni, inizio reclutamento luglio 2016): "Open- Label Study to assess the Efficacy and Safety of Raxone in LHON Patients (LEROS)" (approvazione del comitato etico 16025 - Parere CE-BI - Em. add.1 al protocollo e IB ed. 13 - SNT-IV-005 - LHON - Fav.)

GenSight Pharmaceuticals, trial di terapia genica in doppio-ceco, placebo controllato, randomizzato GS-LHON CLIN-03A RESCUE (inizio reclutamento maggio 2016 – chiuso aprile 2019): "A Randomized, Double-Masked, Sham-Controlled, Pivotal Clinical Trial to Evaluate the Efficacy of a Single Intravitreal Injection of GS010 (rAAV2/2-*ND4*) in Subjects Affected for 6 Months or Less by Leber Hereditary Optic Neuropathy Due to the G11778A Mutation in the Mitochondrial NADH Dehydrogenase 4 Gene" (approvazione del comitato etico protocollo N.699/CE- Comitato Etico Indipendente AUSL Bologna)

GenSight Pharmaceuticals, trial di terapia genica in doppio-ceco, placebo controllato, randomizzato GS-LHON-CLIN-03B REVERSE (1 anno, inizio reclutamento maggio 2016 – chiuso aprile 2019): "Randomized, Double-Masked, Sham-Controlled, Pivotal Clinical Trial to Evaluate the Efficacy of a Single Intravitreal Injection of GS010 (rAAV2/2-ND4) in Subjects Affected for More Than 6 Months and To 12 Months by Leber Hereditary Optic Neuropathy Due to the G11778A Mutation in the Mitochondrial NADH Dehydrogenase 4 Gene" (approvazione del comitato etico protocollo N.700/CE- Comitato Etico Indipendente AUSL Bologna)

Sigma-Tau Spa, trial in doppio-ceco, placebo controllato, randomizzato (1 anno, inizio reclutamento ottobre 2011, trial concluso gennaio 2014): "Effetti della somministrazione di L-Acetilcarnitina sulla conduzione nervosa lungo le vie ottiche nei pazienti affetti da neurotticopatia di Leber" (approvazione comitato etico 10084-Comitato Etico Indipendente AUSL Bologna)

Edison Pharmaceuticals, trial open label su 3 pazienti (2 anni, inizio reclutamento maggio 2011, trial concluso ottobre 2013) "Utilizzo del farmaco EPI-743 nel trattamento della fase acuta della Atrofia Ottica ereditaria di Leber (approvazione comitato etico 780/781/336-Comitato Etico Indipendente AUSL Bologna)

Attività come consulente

2000 – consulente per Allergan (USA): ruolo della Brimonidina come neuroprotettore

2004/2006 – consulente per Pfizer (USA): tossicità mitocondriale dell'antibiotico Linezolid

2006/2008 – consulente per Edison Pharmaceuticals (USA): sviluppo di nuovi composti chinonici come terapia antiossidante nelle malattie mitocondriali

2008 – consulente per Penwest Pharmaceuticals (USA): sviluppo di nuovi composti chinonici come terapia antiossidante nelle malattie mitocondriali

2012 – consulente per European Medicine Agency (EMA) Committee for Human Medicinal Products (CHMP), partecipante al "ad hoc expert group" per l'uso di idebenone nella Neuropatia Ottica Ereditaria di Leber (LHON)

2013-2018 – consulente per l'Agenzia Italiana del Farmaco (AIFA) come esperto per il "National Scientific Advise on Raxone (idebenone) in LHON"

2013-2018 – consulente per Stealth Peptides (USA): messa a punto del protocollo per trial in doppio-ceco, placebo controllato, randomizzato con il farmaco Bendavia, stabilizzatore della cardiolipina, in pazienti con Neuropatia Ottica Ereditaria di Leber (LHON)

2014-2018 – consulente per GenSight (Francia): messa a punto del protocollo per trial in doppio-ceco, placebo controllato, randomizzato con terapia genica (espressione nucleare allotopica di geni del DNA mitocondriale), in pazienti con Neuropatia Ottica Ereditaria di Leber (LHON)

2014-2017 – consulente per il programma del National Institute of Health (NIH) – National Institute of Neurological Disorders and Stroke (NINDS) (USA) denominato "Common Data Elements (CDE) for Mitochondrial Disease": componente del gruppo di lavoro "CDE vision/hearing"

Premi e riconoscimenti per l'attività scientifica

1993/1994: Università di Bologna, borsa di studio Telethon di 1 anno (Progetto # 391, coordinatore Prof. Elio Lugaresi)

1995/1996: Università di Bologna (8 mesi) e Monash University, Clayton, Victoria, Australia (4 mesi), borsa di studio Telethon di 1 anno (Project # 616, coordinatore Dr. Mauro Degli Esposti)

2003 - Visiting Professor alla University of Southern California, Doheny Eye Institute – supportato dal premio "Research to Prevent Blindness (RPB) International Research Scholar Award"

2003 – Premio onorifico del parlamento dello Stato di Espirito Santo, Brasile "Ordem do merito medico capixaba Dr. Alfonso Schwa", October 2003

2006 - Visiting Professor al Wenzhou Medical College, Cina, su invito e supportato dall'Universita' di Wenzhou

2008 - Visiting Professor alla University of Southern California, Doheny Eye Institute – supportato dal premio "Research to Prevent Blindness (RPB) International Research Scholar Award"

Membro di società scientifiche

1. European Biological Rhythms Society (EBRS)

2. The Association for Research in Vision and Ophthalmology (ARVO)

3. North American Neuro-Ophthalmology Society (NANOS)

4. Associazione Italiana di Miologia (AIM)

5. European Neuro-Ophthalmology Society (EUNOS)

6. European Association for Vision and Eye Research (EVER)

Membro di comitati scientifici

1. Consigliere scientifico per International Foundation for Optic Nerve Disease (IFOND) (USA) – http://www.ifond.org/about.php3#about

2. Consigliere scientifico per la Fondazione Giuseppe Tommasello onlus (Italia) – http://www.giuseppetomasello.it/asp/main.asp?dbPageType=5&sez=14&dbmID=14

3. Componente del comitato scientifico di MITOCON (Italia) – http://www.mitocon.it

4. Componente del gruppo allargato del Mitochondrial Disease Sequence Data Resource ("MSeqDR") Consortium, United Mitoochondrial Disease Foundation (UMDF) – https://mseqdr.org/index.php

5. Componente del comitato scientifico internazionale del meeting EUROMIT 2014 – http://bioinf.uta.fi/euromit2014/?page_id=101

Revisore esterno di progetti scientifici per agenzie statali e fondazioni

Medical Research Council, Regno Unito

Association Francaise contre les myopathies, Francia

Revisore esterno per le seguenti riviste

Neurologia e neuroscienze: Lancet Neurology, Brain, Annals of Neurology, Jama Neurology, Neurology, Experimental Neurology, Journal of Neuroscience Methods, European Journal of Neurology, Journal of Neurology, Journal of Neurological Sciences, Neurological Sciences, Neuroscience Letters, Neurobiology of Disease, Epilepsy Research, Muscle and Nerve, Journal of Neurochemistry, Neurogenetics

Biologia e genetica: Nature Genetics, Journal of Inherited Metabolic Disease, Experimental Cell Research, FASEB Journal, BBA Molecular Basis of Disease, BBA Molecular Cell Research, BBA Bioenergetics, Human Molecular Genetics, Trends in Genetics, Journal of Human Genetics, Human Genetics, Human Mutation, American Journal of Medical Genetics, Genetics in Medicine, Molecular Genetics and Metabolism, Mitochondrion

Scienze della visione: Progress in Retinal and Eye Research, Ophthalmology, Investigative Ophthalmology and Visual Science, Experimental Eye Research, Eye

Open access: Plos One, Plos Genetics

Membro di comitati editoriali

Eye and Brain, Dovepress (https://www.dovepress.com/honorary-editorial-board-eye-and-brain-edboard79)

Relatore su invito a congressi nazionali ed internazionali

World Federation of Neurology (WFN), Neurogenetic Research Group Meeting, 30 marzo 1996, San Francisco, California, USA – Valerio Carelli: "***Genotype analysis of Leber's hereditary optic neuropathy (LHON) families detected in Italy***"

Invited lecture at Zilkha Neurogenetic Institute – Keck School of Medicine, University of Southern California, Losa Angeles, May 6 2004 – Valerio Carelli: "***Mitochondrial Optic Neuropathies Induced by mtDNA and nDNA Mutations: Tissue-specific Neurodegeneration with Incomplete Penetrance***"

13th European Bioenergetics Conference (EBEC) 21-26 agosto, 2004, Palazzo dei Congressi, Pisa, Italia – Valerio Carelli: "***Bioenergetics shapes cellular death pathways in Leber's hereditary optic neuropathy (LHON): a model of mitochondrial neurodegeneration***"

International meeting "Mitochondrial and Metabolic Disorders: diagnostic clues and new trends in therapy", University of Padua 30 giugno-1luglio 2006, Padova, Italia – Valerio Carelli: "***It takes two genomes to kill the optic nerve***"

International Symposium Mitochondrial Medicine: Advances in Understanding Pathologies and Therapeutical Strategies. University of Pisa, 25 ottobre 2006, Pisa, Italia - Valerio Carelli: "***Mitochondrial visual disorders***"

Wenzhou Medical College, International Conference on Mitochondrial Biomedicine. Chinese MiT 2006: Mitochondria and Health, 4-6 novembre, 2006, Wenzhou, Cina – Valerio Carelli: "***It takes two genomes to kill the optic nerve: update on pathogenic mechanisms in Leber's and dominant optic neuropathies***"

Wenzhou Medical College. International symposium: Leber's Hereditary Optic Neuropathy, 21-23 marzo, 2008, Wenzhou, Cina – Valerio Carelli: "***The genetic basis of LHON: the missing parts***"

60th American Academy of Neurology (AAN) annual meeting, 12-19 aprile 2008, Chicago, USA – Valerio Carelli: "***OPA1 mutations induce mitochondrial DNA instability and optic atrophy plus phenotypes***"

Euromit 7, From basic mechanisms to disease and ageing. 11-14 giugno 2008, Stoccolma, Svezia – Valerio Carelli: "***OPA1 mutations induce mtDNA instability and optic atrophy plus phenotypes***"

International Symposium on "Mitochondrial Physiology and Pathology", Bari 22-26 giugno 2008, University of Bari, Bari, Italia – Valerio Carelli: "***Mitochondrial Optic Neuropathies: two genomes kill the same cell type***"

Symposium "In the day of the honoris causa degree: from mitochondrial biology to mitochondrial medicine - a tribute to Salvatore "Billi" DiMauro". University of Pisa 12 novembre 2009, Pisa, Italia – Valerio Carelli: "***Why does the optic nerve look at the mitochondria?***"

Mitochondrial Medicine 2010 - United Mitochondrial Disease Foundation (UMDF), 16-18 giugno 2010, Scottsdale, Arizona, USA – Valerio Carelli: "***Pathogenesis and treatment of LHON***"

Second International Symposium on Vision and Visual Dysfunction, 23-26 settembre 2010, Belem, Parà, Brasile – Valerio Carelli: "***Mitochondrial optic neuropathies: from genes to pathogenesis***"

Simposio International de Neuroftalmologia, 12 novembre 2010, Colatina, Espirito Santo, Brasile – Valerio Carelli: "***Genetics of Leber's hereditary optic neuropathy***"

Invited lecture at Medical Research Council-Mitochondrial Biology Unit, 16 marzo 2011, Cambridge, Regno Unito – Valerio Carelli: "***Mitochondrial optic neuropathies as a multifaceted model for neurodegeneration***"

Meeting UILDM "Malattie Neuromuscolari: nuove strategie terapeutiche", 19 marzo 2011, Verona, Italia – Valerio Carelli: "***Neuropatie ottiche mitocondriali, patogenesi e terapia***"

V Convegno Nazionale Associazione Italiana Malattia di Parkinson e Disordini del Movimento DISMOV-SIN, 31 marzo - 2 aprile 2011, Bologna, Italia – Valerio Carelli: "***The Mitochondrial Pathogenesis of Parkinson Disease***"

I Convegno Nazionale "Le Malattie Mitocondriali: dalla Diagnosi alla Gestione Quotidiana. Lo stato della Ricerca, le Terapie, i Diritti dei Malati", 21-22 maggio 2011, Roma, Italia – Valerio Carelli: "***Opzioni terapeutiche attuali e future***"

10th European Neuro-Ophthalmology Society (EUNOS) Meeting, 18-21 giugno 2011, Barcellona, Spagna – Valerio Carelli: "***Mitochondrial optic neuropathies: understanding the pathogenic mechanism and towards a therapy***"

IV Congresso Nazionale Societa' Italiana Oftalmologia Genetica (SIOG) - Nuove Frontiere in Oftalmologia: dalla Diagnosi alla Terapia Genica. Napoli 3 dicembre 2011, Italia – Valerio Carelli: "***Otticopatie: aspetti genetici***"

North American Neuro-Ophthalmology Society (NANOS), 11-16 febbraio 2012, San Antonio, Texas, USA – Valerio Carelli: "***Hereditary mitochondrial optic neuropathies***"

XII Congresso Nazionale Associazione Italiana di Miologia, Scicli (Ragusa), 17-19 maggio 2012, Italia – Valerio Carelli: "***Le alterazioni del SNC nelle malattie muscolari: disturbi oculari***"

48 Congresso dell'Associazione Italiana di Neuropatologia e Neurobiologia Clinica (AIN&NC), 24-26 maggio 2012, Napoli, Italia – Valerio Carelli: "***The Eye: a mito-window to the brain***"

II Convegno nazionale "Le malattie Mitocondriali: dalla diagnosi alla gestione quotidiana. Lo stato della ricerca, le terapie, i diritti dei malati", 26-27 maggio 2012, Firenze, Italia – Valerio Carelli: "***Opzioni terapeutiche nelle neuropatie ottiche mitocondriali***"

III Convegno Nazionale "Malattie mitocondriali: diagnosi, ricerca e gestione della vita quotidiana", 1-2 giugno, 2013, Grand Hotel Duca D'Este, Tivoli Terme – Valerio Carelli: "***Risultati dell'uso di EPI-743 in diverse sindromi in pazienti adulti***"

Meeting United Mitochodnrial Disease Foundation (UMDF) – Mitochondrial Medicine 2013 (June 12-15) – Special LHON Program, Newport Beach Marriott June 13, 2013 – Valerio Carelli: "***LHON clinical trials***"

Meeting European Association for Vision and Eye Research (EVER) 2013, 18-21September 2013, Nice, France – Valerio Carelli: "***The unsolved genetics of LHON: beyond mtDNA primary mutations what else?***"

Meeting “Mitochondrial disease: translating biology into new treatments”, Wednesday 2 – Friday 4 October 2013, Wellcome Trust Conference Centre, Hinxton, UK – Valerio Carelli: “***The paradigm of mitochondrial optic neuropathies: naturally occurring compensatory strategies and treatment options***”

Special Interest Group “Mitochondrial dysfunction and the eye – from monogenic to complex neurodegenerative disorders”, ARVO 2014 Annual Meeting – Orlando, Fla., May 4 – 8, 2014 - Valerio Carelli: “***Retinal ganglion cell loss in primary mitochondrial optic neuropathies – what have we learnt so far?***”

IV Convegno Nazionale sulle Malattie Mitocondriali, 23-25 maggio 2014 – Grand Hotel Duca D’Este, Tivoli Terme – Valerio Carelli: “***L’apparato visivo nei pazienti mitocondriali: le disfunzioni ed i trattamenti***”

Organizzatore di simposi

Satellite al simposio internazionale EUROMIT 5 "***Mitochondrial Dysfunction and Optic Nerve Pathology: a Model for Selective Neurodegeneration***", 24 settembre 2001, Dipartimento di Scienze Neurologiche, Università di Bologna, Bologna, Italia.

Simposio internazionale "***Fissione e fusione dei mitocondri e neurodegenerazione: neuropatia ottica autosomica dominante e mutazioni nel gene OPA1***", 4 novembre 2004, Dipartimento di Scienze Neurologiche, Università di Bologna, Bologna, Italia.

Simposio internazionale "***It takes two genomes to kill the optic nerve: last breaking news on mitochondrial optic neuropathies***", 19 marzo 2007, Dipartimento di Scienze Neurologiche, Università di Bologna, Bologna, Italia.

Corso internazionale “***FEBS-EMBO joint lecture course - Mitochondrial Medicine: from genetics to biological mechanisms and beyond***”,18-22 aprile 2009, Ronzano, Bologna, Italia.

Giornata commemorative internazionale "***Attardi's day***", 22 aprile 2009, Padova, Italia.

Simposio internazionale "***Leber hereditary optic neuropathy (LHON): following for 10 years a gigantic pedigree of Italian ancestry in Brazil***", 9 febbraio 2010, Dipartimento di Scienze Neurologiche, Università di Bologna, Bologna, Italia.

Simposio internazionale "***Eye and metabolism: New diagnostic and therapeutic approaches***", 1 ottobre 2011, Bologna, Italia.

ARVO 2012 "***Special interest group symposium: mitochondrial dysfunction and retinal ganglion cell loss in optic neuropathies - from disease mechanisms to therapeutic strategies***", 7 maggio 2012, Fort Lauderdale, Florida, USA

197th ENMC (European Neuro-Muscular Centre) International workshop: “***Neuromuscular disorders of mitochondrial fusion and fission***”, 26 – 28 April 2013, Naarden, The Netherlands

ATTIVITA' DIDATTICA

Dal 1994, il Dr. Carelli ha tenuto seminari sulla genetica mitocondriale presso le seguenti scuole di specializzazione dell'Universita' di Bologna: Neurologia, Neuropsichiatria Infantile, Genetica Applicata, Cardiologia.

Scuola di Specializzazione in Neurologia (quarto anno di corso), Universita' di Bologna, insegnamento: "Neurobiologia delle malattie metaboliche". Vecchio ordinamento: dall'anno accademico 2001-2002 all'anno accademico 2011-2012, totale di 20 ore/anno. Nuovo ordinamento: dall'anno accademico 2011-2012 a tutt'oggi, totale di 16 ore/anno. Per gli anni accademici 2011-2012 sovrapposizione dei due ordinamenti.

Scuole di specializzazione di Neuropsichiatria Infantile, Universita' di Bologna, insegnamento "Neurobiologia delle malattie metaboliche".

Corso "Basi biomolecolari delle patologie d'organo - Neurologia" (3 crediti formativi) negli anni accademici 2007/2008, 2010/2011, 2012/2013, 2014/2015, per gli studenti del corso di Laurea Magistrale in Biotecnologie mediche.

Corso elettivo in "Medicina Mitocondriale" negli anni accademici 2008-2009 (1 credito formativo), 2009-2010 (1 credito formativo), 2010-2011 (2 crediti formativi), e corso elettivo in "Bioenergetica: Fisiologia e Patologia dei Mitocondri – Patologie Mitocondriali (terzo modulo)" negli anni accademici 2012-2013 (1 credito formativo) e 2013-2014 (1 credito formativo), per gli studenti del corso di Laurea in Medicina e Chirurgia, Universita' di Bologna.

Insegnamento a corsi nazionali ed internazionali

Professore a Contratto per un ciclo di lezioni presso la Scuola Superiore di Studi Universitari e di Perfezionamento Sant'Anna di Pisa – Classe di Scienze Sperimentali, Settore di Medicina corso integrativo all'insegnamento di Chimica Biologica (Titolare dell'insegnamento Prof. Giancarlo Solaini) dal titolo:

"Aspects of Human Pathologies in Relation to Genes and Biochemical Alterations".

Il corso si e' svolto in sette giornate per un totale di 20 ore di insegnamento secondo il calendario e gli argomenti trattati che seguono:

- 02/06/1998 – Il genoma mitocondriale: peculiarita' di un sistema genetico che sfugge le leggi di Mendel.
- 03/06/1998 – Replicazione e trascrizione del DNA . Il sistema dei "cibridi' come modello per lo studio in vitro dei mitocondri.
- 04/06/1998 – Eteroplasmia e segregazione mitotica e meiotica del DNA mitocondriale: implicazioni per l'espressione delle patologie mitochondriali e ipotesi del "bottleneck" nella trasmissione del DNA mitocondriale
- 15/06/1998 – "Mutational rate" del DNA mitocondriale, genetica delle popolazioni e invecchiamento
- 16/06/1998 – "Neuropatia ottica ereditaria di Leber (LHON)" la piu' studiata delle malattie mitochondriali e' ancora un enigma
- 18/06/1998 – "MELAS" e "MERRF": mutazioni nei tRNA producono meccanismi patogenetici e fenotipi clinica diversi
- 19/06/1998 – delezioni e difetti del DNAmt ad ereditarieta' mendeliana (delezioni multiple autosomi che recessive e dominanti, deplezione del DNAmt)

European School of Genetic Medicine, 19th Course in Medical Genetics, 26 Apr - 2 May 2006, University Residential Center of Bertinoro

Valerio Carelli: “Mitochondrial disorders”

European School of Genetic Medicine, 21st Course in Medical Genetics, 4-10 May 2008, University Residential Center of Bertinoro

Valerio Carelli: “Optic nerve degeneration: a mitochondrial disease of two genomes”

Corso di Aggiornamento sulle Malattie Rare del Sistema Nervoso - 8 ottobre 2008 – 18 Marzo 2009 Lavagna (GE), Villa Grimaldi

Valerio Carelli: “Aspetti eziopatogenetici delle atrofie ottiche ereditarie”

Corso di aggiornamento “Argomenti di Neuro-Oftalmologia”, 5 dicembre 2009, Este (Padova)

Valerio Carelli: “Neuropatia ottica bilaterale: malattie mitocondriali”

RESEARCH SUMMER SCHOOL ANGERS 2010 « Intensive training for earlier exposure to research » 30th June – 14th July 2010, Medical School – UNIVERSITY OF ANGERS (www.univ-angers.fr/researchsummerschool2010)

Valerio Carelli, Bologna University (IT), “Pathogenesis and Treatment of Leber hereditary optic neuropathy”

European School of Genetic Medicine, Course in Genetics and Molecular Pathology of Age Related Neurodegenerative Diseases, 29-31 Mar 2010, at the EuroMediterranean University Centre of Ronzano, Bologna Italy

Valerio Carelli: “Genetic of Parkinson disease”

Corso di aggiornamento “Malattie neurologiche rare” – Siena 19-21 maggio 2011

Valerio Carelli: “Rare atrofie ottiche ereditarie (LHON e ADOA)”

Corso di aggiornamento “IV Update sulla genetica delle malattie neurologiche”, 15/16 settembre 2011, Cagliari

Valerio Carelli: “Atrofie ottiche ereditarie”

European School of Genetic Medicine, 2nd Course in Eye Genetics, September 28th- October 1st, 2011 at the Euro-Mediterranean University Center located in Ronzano - Bologna, Italy

Valerio Carelli: “Mitochondrial eye diseases”

Corso di aggiornamento “Eye and metabolism: New diagnostic and therapeutic approaches” - Saturday, October 1th, 2011 | Sala Farnese, Palazzo D’Accursio, Piazza Maggiore, Bologna

Valerio Carelli: “New diagnostic and therapeutic approaches in cellular degeneration: therapeutic approaches in LHON”

Corso di aggiornamento “Le malattie mitocondriali in età infantile: dalla neurobiologia al letto del malato”, 7-8 ottobre 2011, Aula Magna Università degli Studi di Modena e Reggio Emilia, Reggio Emilia

Valerio Carelli: “La diagnosi molecolare”

Corso di aggiornamento “V Update sulla genetica delle malattie neurologiche”, 27/28 settembre 2012, Cagliari

Valerio Carelli: “Medicina mitocondriale”

Corso internazionale di neuro-oftalmologia “Neuropatie ottiche: attualità e prospettive”, 17 ottobre 2012, Aula Magna Ospedale C. Forlanini, Roma

Valerio Carelli: “Neuropatie ottiche ereditarie/mitocondriali: background per l’oculista/neurologo”

Corso residenziale in Neurogenetica, 29-30 Maggio 2013 – Pisa

Valerio Carelli: “Dal fenotipo al genotipo e viceversa, approccio sul campo”

European School of Genetic Medicine, 3rd Course in Eye Genetics, October 13-15, 2013 – University Residential Center of Bertinoro

Valerio Carelli: “Treatment options for mitochondrial eye disease”

XXVI Corso di aggiornamento “Disordini della funzione visiva e patologie del neuro sviluppo: dalla diagnosi alla riabilitazione” – Grand Hotel Baglioni, Firenze, 10-12 marzo 2014

Valerio Carelli: “Mitocondri e funzione visiva in eta’ evolutiva”

Corso di aggiornamento di neuro-oftalmologia “From the optic nerve to the connectome”, Parma, 21 marzo 2014

Valerio Carelli: “Casi clinici”

Commissario per discussione di tesi di Dottorato in Italia

Dott.ssa Mara Dolmo, tesi di dottorato: "Yeast Models for the Study of Mitochondrial Genetic Defect and Other Metabolic Disorders" Universita' degli Studi di Padova, Scuola di Dottorato in Medicina dello Sviluppo e Scienze della Programmazione - Indirizzo ematooncologia, immunologia e genetica XXIV Ciclo, 2012

Dott. Ivano Di Meo, tesi di dottorato: "Altered Sulfide Metabolism in Ethylmalonic Encephalopathy" Universita’ di Milano-Bicocca Scuola di Medicina e Facolta’ di Scienze, Dottorato in Medicina Molecolare e Translazionale, XXIV Ciclo, 2012

Dott. Giovanni Tricomi, tesi di dottorato: "Alterazioni della Sostanza Bianca nelle Malattie Mitocondriali in Eta' Pediatrica" Universita' degli Studi di Messina, Dottorato di Ricerca in Neuroscienze Cliniche, XXIII Ciclo, 2011

Dott. Luigi Citrigno, PhD Thesis: "Studio Genetico di una Famiglia affetta da Neuropatia Motoria Distale (dHMN) complicata da segni piramidali" Universita' degli Studi di Messina, Dottorato di Ricerca in Neuroscienze Cliniche, XXIII Ciclo, 2011

Dott. Marco Crimi, PhD Thesis: "Eziopatogenesi molecolare delle encefalopatie mitocondriali" Universita' Statale degli Studi di Milano, Dottorato in Medicina Molecolare, XVI Ciclo, 2005

Commissario per discussione di tesi di Dottorato in Europa

Dr. Mika H. Martikainen, PhD Thesis: "Mitochondrial Diseases in Southwestern Finland: population-based molecular genetic and clinical studies" University of Turku, Turku 2012, Finland

Dr. Patrick Yu-Wai-Man, PhD Thesis: "The Clinical Manifestations and Molecular Mechanisms of Mitochondrial Neuro-Ophthalmological Disorders" Newcastle University, Newcastle 2010, United Kingdom

Dr. Joanna Stewart, PhD Thesis: "Nuclear mitochondrial interactions in human disease" Newcastle University, Newcastle 2008, United Kingdom

Tutor o co-tutor per tesi di Dottorato presso l’Universita’ di Bologna

Dr. Chiara La Morgia, PhD Thesis: "Melanopsin Retinal Ganglion Cells: relevance to circadian rhythms and sleep in neurodegeneration" Alma Mater Studiorum - Universita' di Bologna, Dottorato di Ricerca in Scienze Mediche Specialistiche PFDR in Medicina del Sonno XXIV Ciclo, 2012

Dr. Marzio Bellan, PhD Thesis: "Studio degli aplogruppi mitocondriali nei pazienti con narco-cataplessia" Alma Mater Studiorum - Universita' di Bologna, di Ricerca in Scienze Mediche Specialistiche PFDR in Medicina del Sonno XXIII Ciclo, 2011

Dr. Alessandra Maresca, PhD Thesis: "Pathogenic mechanism in mitochondrial optic neuropathies" Alma Mater Studiorum - Universita' di Bologna, Dottorato di Ricerca in Biologia e Biologia Cellulare XXIII Ciclo, 2011

Tutor o co-tutor a tesi di Laurea per le Facolta' di Medicina, Biologia e Biotecnologia

Dott.ssa Rosanna Carroccia, Thesis: "Identificazione di un raro riarrangiamento complesso (delezione/inserzione invertita) analizzando i breakpoints di delezioni del genoma mitocondriale umano" Universita' degli Studi di Ferrara, Facolta' di Scienze Matematiche, Fisiche e Naturali, Corso di Laurea in Scienze Biomolecolari e Cellulari, Anno Accademico 2008-2009

Dott.ssa Daniela Strobbe, Thesis: "Analisi di possibili modificatori della penetranza delle mutazioni del gene OPA1 nell'atrofia ottica dominante" Alma Mater Studiorum - Universita' di Bologna, Facolta' di Medicina e Chirurgia, Corso di Laurea Specialistiche in Biotecnologie Mediche, Anno Accademico 2008-2009

Dott.ssa Francesca Tagliavini, Thesis: "Penetranza incompleta nella neuropatia ottica ereditaria di Leber (LHON) con mutazione mitocondriale 11778: analisi di potenziali geni modificatori nucleari" Alma Mater Studiorum - Universita' di Bologna, Facolta' di Scienze Matematiche, Fisiche e Naturali, Corso di Laurea in Scienze Biologiche, Anno Accademico 2007-2008

Dott.ssa Alessandra Maresca, Thesis: "Studio dell'influenza della biogenesi mitocondriale sulla variabilita' della penetranza nella neuropatia ottica ereditaria di Leber" Alma Mater Studiorum - Universita' di Bologna, Facolta' di Medicina e Chirurgia, Corso di Laurea Specialistiche in Biotecnologie Mediche, Anno Accademico 2006-2007

Dott.ssa Caterina Marconi, Thesis: "Studio della correlazione tra variabilita' genetica e quantitative del DNA mitocondriale in soggetti sani e affetti da atrofie ottiche ereditarie" Alma Mater Studiorum - Universita' di Bologna, Facolta' di Scienze Matematiche, Fisiche e Naturali, Corso di Laurea triennale in Biotecnologie, Anno Accademico 2006-2007

Dott.ssa Beatrice Foscarini, Thesis: "Gli aplogruppi del DNA mitocondriale come modificatori dell'espressivita' clinica nell'atrofia ottica dominante legata al gene OPA1" Alma Mater Studiorum - Universita' di Bologna, Facolta' di Medicina e Chirurgia, Anno Accademico 2006-2007

Dott. Marcello Amadori, Thesis: "Correlazione tra percentuale di copie mutanti nel DNA mitocondriale ed espressione clinico-biochimica in una famiglia portatrice della mutazione A3243G/tRNALeu(UUR)" Alma Mater Studiorum - Universita' di Bologna, Facolta' di Medicina e Chirurgia, Anno Accademico 2003-2004

Dott. Filippo Fortuna, Thesis: "Caratterizzazione clinica della neuropatia ottica nella eredoatassia di Friedreich ed identificazione di possibili geni modificatori" Alma Mater Studiorum - Universita' di Bologna, Facolta' di Medicina e Chirurgia, Anno Accademico 2003-2004

Tutor o co-tutor a tesi di Specializzazione in Neurologia

Dott.ssa Chiara La Morgia, Thesis: "Fotorecezione circadiana nelle neuropatie ottiche ereditarie" Alma Mater Studiorum - Universita' di Bologna, Scuola di Specializzazione in Neurologia, Anno Accademico 2007-2008

Dott. Marzio Bellan, Thesis: "Oftalmoplegia cronica progressiva con disfunzione mitocondriale: definizione delle caratteristiche sindromiche e delle basi molecolari" Alma Mater Studiorum - Universita' di Bologna, Scuola di Specializzazione in Neurologia, Anno Accademico 2005-2006

Dott.ssa Maria Lucia Valentino, Thesis: " Differenti modalità di segregazione lungo la linea germinale femminile delle mutazioni del DNA mitocondriale A3243G (MELAS) e T8993G (NARP): rilevanza per il consiglio genetico" Alma Mater Studiorum - Universita' di Bologna, Scuola di Specializzazione in Neurologia, Anno Accademico 2002-2003

Attività istituzionali svolte presso UNIBO:

Responsabile del "Centro per lo studio e la diagnosi delle malattie neurogenetiche" attivato il 15 luglio 2004 con D.R. 1327/44582.

Membro eletto della Giunta del Dipartimento di Scienze Neurologiche dell'Università di Bologna dal 2010 al 2012.

Membro eletto della Giunta del Dipartimento di Scienze Biomediche e NeuroMotorie (DIBINEM) dell'Università di Bologna dal 2012 al 2014.

Membro nominato della Commissione Ricerca del Dipartimento di Scienze Biomediche e NeuroMotorie dell'Università di Bologna dal 2012 al 2014.

Membro di commissioni di sedute di laurea e dell'esame di ammissione alla Scuola di Medicina.

Membro per nomina rettorale del Gruppo Tematico di Ateneo GTA-Health dal 20/04/2017 - presente.

Membro eletto della Giunta del Dipartimento di Scienze Biomediche e NeuroMotorie (DIBINEM) dell'Università di Bologna dal 2018 - presente.

Coordinatore per nomina rettorale del Gruppo Tematico di Ateneo GTA-Health da gennaio 2020 - presente.

Attività istituzionali svolte presso l'IRCCS Istituto delle Scienze Neurologiche di Bologna:

Membro nominato dalla Direzione scientifica del Comitato Tecnico Scientifico dell'Istituto.

Coordinatore della Linea di Ricerca 2 "MALATTIE NEUROMUSCOLARI, MALATTIE RARE E NEUROIMMUNOLOGICHE"

ATTIVITA' ASSISTENZIALE

Dal 2001 – presa di servizio come medico neurologo presso la clinica neurologica, con attivita' di reparto.

Dal 2002 – inserimento nei turni di guardia.

Dal 2005 – inserimento come dirigente medico (neurologo) di primo livello, AUSL citta di Bologna, nel servizio di Day Hospital dell' UO Clinica Neurologica, e attivita' ambulatoriale.

Dal 2012 – trasferimento all'Ospedale Bellaria con inserimento come dirigente medico nello staff di neurologi in servizio presso il reparto, day hospital, e attivita' ambulatoriale (ambulatorio malattie mitocondriali) dell'UOC Clinica Neurologica e turni di guardia notturna. Inoltre, nella veste di referente del Laboratorio di Neurogenetica, refertazione diagnosi genetiche e attivita' di certificazione di malattie rare.

Dal 2020 – incarico di facente funzione direzione Programma di Neurogenetica, IRCCS Istituto delle Scienze Neurologiche di Bologna, Ospedale Bellaria

TOTALE PUBBLICAZIONI: 287 lavori + 6 capitoli di libro

TOTALE CITAZIONI: 14337 (da Scopus)

TOTALE H INDEX: 69 (da Scopus)

N.B. Il valore dell'I.F. applicato è quello del JCR Science dell'anno di pubblicazione, salvo diversamente specificato in parentesi. Ai lavori pubblicati prima del 2000 è stato attribuito il valore di I.F. più vicino all'anno di pubblicazione ancora reperibile dal sito online.

Lavori pubblicati indicizzati su PubMed (Agosto 2020)

2020

287. Maresca A, Del Dotto V, Romagnoli M, La Morgia C, Di Vito L, Capristo M, Valentino ML, **Carelli V**; ER-MITO Study Group.

Expanding and validating the biomarkers for mitochondrial diseases [published online ahead of print, 2020 Aug 26].

J Mol Med (Berl). 2020;10.1007/s00109-020-01967-y. doi:10.1007/s00109-020-01967-y

I.F. 4.427

286. Ronchi D, Caporali L, Manenti GF, Meneri M, Mohamed S, Bordoni A, Tagliavini F, Contin M, Piga D, Sciacco M, Saetti C, **Carelli V**, Comi GP.

TYMP Variants Result in Late-Onset Mitochondrial Myopathy With Altered Muscle Mitochondrial DNA Homeostasis.

Front Genet. 2020;11:860. Published 2020 Aug 5. doi:10.3389/fgene.2020.00860

I.F. 3.258

285. Romagnoli M, Stanzani Maserati M, De Matteis M, De Matteis M, Capellari S, Carbonelli M, Amore G, Cantalupo G, Zenesini C, Liguori R, Sadun AA, **Carelli V**, Park JC, La Morgia C.

Chromatic Pupillometry Findings in Alzheimer's Disease.

Front Neurosci. 2020;14:780. Published 2020 Aug 11. doi:10.3389/fnins.2020.00780

I.F. 3.707

284. Zaninello M, Palikaras K, Naon D, Iwata K, Herkenne S, Quintana-Cabrera R, Semenzato M, Grespi F, Ross-Cisneros FN, **Carelli V**, Sadun AA, Tavernarakis N, Scorrano L.

Inhibition of autophagy curtails visual loss in a model of autosomal dominant optic atrophy.

Nat Commun. 2020;11(1):4029. Published 2020 Aug 12. doi:10.1038/s41467-020-17821-1

I.F. 12.121

283. Peron C, Mauceri R, Cabassi T, Segnali A, Maresca A, Iannielli A, Rizzo A, Sciacca FL, Broccoli V, **Carelli V**, Tiranti V.

Generation of a human iPSC line, FINCBi001-A, carrying a homoplasmic m.G3460A mutation in MT-ND1 associated with Leber's Hereditary optic Neuropathy (LHON) [published online ahead of print, 2020 Aug 3].

Stem Cell Res. 2020;48:101939. doi:10.1016/j.scr.2020.101939

I.F. 4.489

282. Minardi R, Licchetta L, Baroni MC, Pippucci T, Stipa C, Mostacci B, Severi G, Toni F, Bergonzini L, **Carelli V**, Seri M, Tinuper P, Bisulli F.

Whole-exome sequencing in adult patients with developmental and epileptic encephalopathy: it is never too late [published online ahead of print, 2020 Jul 29].

Clin Genet. 2020;10.1111/cge.13823. doi:10.1111/cge.13823

I.F. 4.104

281. D'Angelo R, Boschetti E, Amore G, Costa R, Pugliese A, Caporali L, Gramegna LL, Papa V, Vizioli L, Capristo M, Contin M, Mohamed S, Cenacchi G, Lodi R, Morelli MC, Fasano L, Pisani L, Cescon M, Tonon C, Pinna AD, Dotti MT, Sicurelli F, Scarpelli M, Filosto M, Casali C, Pironi L, **Carelli V**, De Giorgio R, Rinaldi R.

Liver transplantation in mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): clinical long-term follow-up and pathogenic implications [published online ahead of print, 2020 Jul 18].

J Neurol. 2020;10.1007/s00415-020-10051-x. doi:10.1007/s00415-020-10051-x

I.F. 3.956

280. Biousse V, Newman NJ, Najjar RP, Vasseneix C, Xu X, Ting DS, Milea LB, Hwang JM, Kim DH, Yang HK, Hamann S, Chen JJ, Liu Y, Wong TY, Milea D; BONSAI (Brain and Optic Nerve Study with Artificial Intelligence) **Group**: Barnabé Rondé-Courbis, Philippe Gohier, Valérie Biousse, Nancy J Newman, Caroline Vasseneix, Neil Miller, Tanyatuth Padungkiatsagul, Anuchit Poonyathalang, Yanin Suwan, Kavin Vanikieti, Leonard B Milea, Giulia Amore, Piero Barboni, Michele Carbonelli, **Valerio Carelli**, Chiara La Morgia, Martina Romagnoli, Marie-Bénédicte Rougier, Selvakumar Ambika, Swetha Komma, Pedro Fonseca, Miguel Raimundo, Steffen Hamann, Isabelle Karlesand, Wolf Alexander Lagrèze, Nicolae Sanda, Gabriele Thumann, Florent Aptel, Christophe Chiquet, Kaiqun Liu, Hui Yang, Carmen Km Chan, Noel Cy Chan, Carol Y Cheung, Thi Ha Chau Tran, James Acheson, Maged S Habib, Neringa Jurkute, Patrick Yu-Wai-Man, Richard Kho, Jost B Jonas, John J Chen, Nouran Sabbagh, Catherine Vignal-Clermont, Rabih Hage, Raoul K Khanna, Jeong-Min Hwang, Dong Hyun Kim, Hee Kyung Yang, Tin Aung, Ching-Yu Cheng, Ecosse Lamoureux, Jing Liang Loo, Dan Milea, Raymond P Najjar, Shweta Singhal,

Daniel Ting, Sharon Tow, Caroline Vasseneix, Tien Yin Wong, Yong Liu, Xinxing Xu, Zhubo Jiang, Clare L Fraser, Luis J Mejico, Masoud Aghsaei Fard.

Optic disc classification by deep learning versus expert neuro-ophthalmologists.

Ann Neurol. 2020 Jul 3. doi: 10.1002/ana.25839. Epub ahead of print.

I.F. 9.037

279. Vianello C, Cocetta V, Caicci F, Boldrin F, Montopoli M, Martinuzzi A, **Carelli V**, Giacomello M.

Interaction between Mitochondrial DNA Variants and Mitochondria/Endoplasmic Reticulum Contact Sites: A Perspective Review.

DNA Cell Biol. 2020 Jun 26. doi: 10.1089/dna.2020.5614. Epub ahead of print.

I.F. 3.191

278. Charif M, Chevrollier A, Gueguen N, Bris C, Goudenège D, Desquiret-Dumas V, Leruez S, Colin E, Meunier A, Vignal C, Smirnov V, Defoort-Dhellemmes S, Drumare Bouvet I, Goizet C, Votruba M, Jurkute N, Yu-Wai-Man P, Tagliavini F, Caporali L, La Morgia C, **Carelli V**, Procaccio V, Zanlonghi X, Meunier I, Reynier P, Bonneau D, Amati-Bonneau P, Lenaers G.

Mutations in the m-AAA proteases AFG3L2 and SPG7 are causing isolated dominant optic atrophy.

Neurol Genet. 2020;6(3):e428. Published 2020 May 20. doi:10.1212/NXG.0000000000000428

I.F. 3.509

277. Palombo F, Graziano C, Al Wardy N, Nouri N, Marconi C, Magini P, Severi G, La Morgia C, Cantalupo G, Cordelli DM, Gangarossa S, Al Kindi MN, Al Khabouri M, Salehi M, Giorgio E, Brusco A, Pisani F, Romeo G, **Carelli V**, Pippucci T, Seri M.

Autozygosity-driven genetic diagnosis in consanguineous families from Italy and the Greater Middle East [published online ahead of print, 2020 Jun 2].

Hum Genet. 2020;10.1007/s00439-020-02187-7. doi:10.1007/s00439-020-02187-7

I.F. 5.207

276. La Morgia C, Maresca A, Caporali L, Valentino ML, **Carelli V**.

Mitochondrial diseases in adults.

J Intern Med. 2020;287(6):592-608. doi:10.1111/joim.13064

I.F. 6.051

275. Milea D, Najjar RP, Zhubo J, Ting D, Vasseneix C, Xu X, Aghsaei Fard M, Fonseca P, Vanikieti K, Lagrèze WA, La Morgia C, Cheung CY, Hamann S, Chiquet C, Sanda N, Yang H, Mejico LJ, Rougier M-B, Kho R, Thi Ha Chau T, Singhal S, Gohier P, Clermont-Vignal C, Cheng C-Y, Jonas JB, Yu-Wai-Man P, Fraser CL, Chen JJ, Ambika S, Miller NR, Liu Y, Newman NJ, Wong TY, Biousse V; BONSAI (Brain and Optic Nerve Study with Artificial Intelligence) Group: Dan Milea, Valérie Biousse, Nancy J Newman, Raymond P Najjar, Tien Y Wong, Daniel Ting, Yong Liu, Philippe Gohier, Barnabé Rondet-Courbis, Caroline Vasseneix, Neil Miller, Tanyatuth Padungkiatsagul, Anuchit Poonyathalang, Yanin Suwan, Kavin Vanikieti, Giulia Amore, Piero Barboni, Michele Carbonelli, **Valerio Carelli**, Chiara La Morgia, Martina Romagnoli, Marie-Bénédicte Rougier, Selvakumar Ambika, Swetha Komma, Pedro Fonseca, Miguel Raimundo, Steffen Hamann, Isabelle Karlesand, Lars Fuhrmann, Sebastian Küchlin, Wolf Alexander Lagrèze, Nicolae Sanda, Gabriele Thumann, Florent Aptel, Christophe Chiquet, Kaiqun Liu, Hui Yang, Carmen K M Chan, Noel C Y Chan, Carol Y Cheung, Thi Ha Chau Tran, James Acheson, Maged S Habib, Neringa Jurkute, Patrick Yu-Wai-Man, Richard Kho, Jost B Jonas, John J Chen, Nouran Sabbagh, Catherine Vignal-Clermont, Rabih Hage, Raoul Kanav Khanna, Tin Aung, Ching-Yu Cheng, Ecosse Lamoureux, Jing Liang Loo, Leopold Schmetterer, Shweta Singhal, Sharon Tow, Xinxing Xu, Zhubo Jiang, Clare L Fraser, Luis J Mejico, Andrew L Orenberg, Masoud Aghsaei Fard

Artificial Intelligence to Detect Papilledema from Ocular Fundus Photographs.

N Engl J Med. 2020;382(18):1687-1695. doi:10.1056/NEJMoa1917130

I.F. 70.670

274. Romagnoli M, La Morgia C, Carbonelli M, Di Vito L, Amore G, Zenesini C, Cascavilla ML, Barboni P, **Carelli V.**

Idebenone increases chance of stabilization/recovery of visual acuity in OPA1-dominant optic atrophy.

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