## Chiara La Morgia, MD, PhD

**Personal data:**

Date of birth: 3rd December 1977, Lanciano (CH), Italy

Nationality: Italian

Fiscal code: LMRCHR77T43E435I

**Affiliations:**

* IRCCS Istituto delle Scienze Neurologiche di Bologna, UOC Clinica Neurologica,

Ospedale Bellaria, Via Altura 3, Bologna (BO)

* Dipartimento di Scienze Biomediche e NeuroMotorie (DIBINEM)

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“Ordine dei Medici di Bologna”, n.1472 (since 2002)

# Education and training:

* 1996: High school degree, Liceo Classico “Vittorio Emanuele II”, 1996, final mark 60/60
* 2002: MD degree, University of Bologna, Italy. Final mark: 110/110 cum laudae. Thesis: “Neurological complications in bone marrow transplantation”
* 2003-2008: Residency in Neurology, Department of Neurological Sciences, University of Bologna, Italy. Final mark: 70/70 cum laudae. Thesis: “Circadian photoreception in hereditary optic neuropathies”
* 2009-2012: PhD in Sleep Medicine, Department of Neurological Sciences, University of Bologna, Italy. Thesis: “Melanopsin retinal ganglion cells: relevance to circadian rhythms and sleep in neurodegeneration”
* April 2014-September 2014: clinical fellowship in Neurophthalmology at the Doheny Eye Institute, University of Southern California (USC), now University of California Los Angeles (UCLA), Los Angeles, California, USA

**Research experience:**

* November 2005: granted as “Expert in Sleep Medicine” by the Italian Association of Sleep Medicine (AIMS)
* 2008-2014: Research fellowship at Doheny Eye Institute, University of Southern California, Los Angeles (California, USA) for collaborative project on “Neurodegenerative disorders and circadian photoreception” with attendance periods January 25th-February 9th 2008; August 23th-September 13th 2008; February 19th- March 4th 2009; February 21st-March 13th 2010; August 20th-September 10th 2010; August 28th-September 12th 2011; February 15th-March 2nd 2012; July 17th-August 5th 2013; May 10th-May 25th 2013; March 24th-October 3rd 2014)
* 2010-2019: member of the international team ‘Projeto LHON/Brazil’ for the study of a LHON Brazilian family with a project on “Melanopsin retinal ganglion cells and pupillary reflex in Leber’s hereditary optic neuropathy patients”
* 2006-2010: component of the research team on “Telethon project GGP06233”, titled: “Pathogenic mechanisms for degeneration of retinal ganglion cells in mitochondrial optic neuropathies”
* 2010-2013: component of the research team on “Telethon project, Program Project 2010”, titled “Therapeutic strategies to combat mitochondrial disorders”
* 2010-2014: component of the research project titled “Identification of predictive factors for conversion from mild cognitive impairment to dementia” (Fondazione Gino Galletti, Italy)
* 2012-2018: component of the research project titled “Emilia-Romagna region Program Project "Recognition, diagnosis and therapy of mitochondrial disorders in neurological services of the Emilia-Romagna region" (grant ER-MITO)
* January 2012-september 2015: research fellowship, project titled: “Function of melanopsin expressing retinal ganglion cells in neurological disorders and aging” Department of Biomedical and Neuromotor Sciences (DIBINEM), University of Bologna, Italy
* 2013-ongoing: member of the committee “Sleep and Neurodegenerative disorders”, Italian Society for Sleep Medicine (AIMS)
* July 2016-Nov 2020: PI of the research project titled “Melanopsin retinal ganglion cells and circadian rhythms: function and dysfunction in Alzheimer's disease and aging”– GR-2013-02358026 granted by the Italian Ministry of Health
* Dec 2016-ongoing: Member of the Scientific Board for the Association “International Foundation for Optic Nerve Disease” dal 6 Dicembre 2016
* Component of the reaserch team “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” 2019, PI: Prof. Valerio Carelli
* March 2017-June 2018: component of the research team for the project titled “Studio multicentrico italiano dei geni implicati nel ritmo sonnoveglia e degli aplogruppi mitocondriali nella malattia di Alzheimer lieve-moderata e nel Mild Cognitive Impairment”, Coordinator Center: Azienda Ospedaliero-Universitaria Pisana, UOC Neurologia, Dr Enrica Bonanni
* August 2017-ongoing: Reviewer Editor for Frontiers in Neurology, “Neurophthalmology” section
* Dec 2017-Feb 2022: Co-PI for the research project GR-2016-02361449 granted by the Italian Ministry of Health titled “Italian Project on Hereditary Optic Neropathies (IPHON): from genetic basis to therapy”, PI: Prof. Valerio Carelli
* July 2018-2024: member of the Neurogenetic Panel, European Academy of Neurology (EAN)
* 2019-ongoing: Co-PI of the project granted by the Italian Ministry of Health RF-2018-12366703 “REtinal ganglion cells and ORganoids from Inherited Optic Neuropathies: light on pathogenesis to fight blindness (REORION Project)” 2019, coordinator (Prof. Valerio Carelli) - Italian Mininstry for Research
* Oct 2020-ongoing: part of the team for the research project “ENLIGHTENme- Innovative policies for improving citizens’ health and wellbeing addressing artificial lighting” - Horizon 2020 - Research and Innovation Framework Programme (Call: H2020-SC1-2020-Two-Stage-RTD Project: 945238 — ENLIGHTENme
* Nov 2022-ongoing: part of the research team for the project Partenariato Esteso “MNESYS – A Multiscale integrated approach to the study of the Nervous system in health and disease”, Spoke 6
* Member of the study group in Neurophthalmology and Neurogenetics of the Italian Neurology Association
* 09/24-ongoing: co-PI for the project PNRR-MR1-2023-12377223 “THERAPY4ALL-Towards therapies for mitochondrial diseases”
* 10/24-ongoing: part of the research team in the project “EPPERMED2024-360 - SEI-MITO”, finanziato nell’ambito della call Joint Transnational Call for Proposals (JTC) 2024 on "Identification or Validation of Targets for Personalised Medicine Approaches (PMTargets)", EP PerMed
* 2024-ongoing: member of the Management Group, Neurogenetic Panel, European Academy of Neurology (EAN)
* 12/24-ongoing: Associate Editor per la rivista Neuro-Ophthalmolog
* 2024-ongoing: member of the Working Group ERN-EYE
* 2024-ongoing: member of the coordinating team for rare diseases, Italian Society of Neurology

**Professional experience:**

* 2009-2012: clinical activity at the Neurologic Clinic Unit, Department of Neurological Sciences, University of Bologna (Day-Hospital, outpatient clinic and clinical ward)
* 2013-now: clinical activity at the Unit of Neurology, IRCCS Istituto delle Scienze Neurologiche di Bologna, Bellaria Hospital, Bologna, Italy
* July 2011-May 2013: contract at the Department of Neurological Sciences, University of Bologna for the clinical trial sponsored by Sigma-Tau “Effects of l-acetylcarnitine on optic pathways neural conduction in Leber’s hereditary Optic neuropathy patients”
* October 2013-March 2014: contract at the USL Bologna on a project on “Prevention of road accidents during working hours”
* October 2015-October 2020: Research contract at the University of Bologna with integration of clinical activity at the IRCCS Istituto delle Scienze Neurologiche di Bologna, Ospedale Bellaria
* Nov 2020-21August 2022: Neurologist at IRCCS Institute of Neurological Sciences, Neurology Unit, Bologna
* 22 August 2022-ongoing: Senior Researcher at the University of Bologna, Department of Biomedical and Neuromotor Sciences and Neurologist at IRCCS Institute of Neurological Sciences, Neurology Unit, Bologna
* 2015-ongoing: Chief of the Neuro-ophthalmology Clinic, UOC Clinica Neurologica, Bellaria Hospital

**Clinical trials:**

* May 2016-now: study coordinator of the clinical trials Rescue (GS-LHON-CLIN-03A) and Reverse (GS-LHON-CLIN-03B) sponsored by Gensight on gene therapy in LHON patients (11778/ND4 mutation)
* July 2016-Sept 2021: investigator in the clinical trial “External Natural History Controlled, Open-Label Intervention Study to Assess the Efficacy and Safety of Long-Term Treatment with Raxone® in Leber’s Hereditary Optic Neuropathy (LHON)”- SNT-IV-005-LEROS, sponsored by Santhera Pharmaceuticals
* Dec 2017-July 2020: PI of the Reality-LHON Registry trial sponsored by Gensight
* Nov 2018-Jul 2022: investigator in the clinical trial GS-LHON-CLIN-06 (follow-up study) sponsored by Gensight on gene therapy in LHON patients (11778/ND4 mutation)
* Jan 2019-Jul 2021: investigator in the clinical trial “Post Authorisation Safety Study with Raxone in LHON Patients (PAROS)- SNT-IV-003, sponsored by Santhera Pharmaceuticals
* March 2019-ongoing: investigator in the clinical trial GS-LHON-CLIN-05 (Reflect Study) sponsored by Gensight on gene therapy in LHON patients (11778/ND4 mutation)
* Jul 2022-ongoing: Sub-I for REN001-201 clinical trial
* Sept 2022 PI for STK-002-OA-901 FALCON Study (site to be activated in Oct 2022)
* July 2023-ongoing: Sub-I for OMT28-C0203 clinical trial

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| **AWARDS:** |
| * “Progetto Giovani”, 2005, 2006, 2007, 2008, 2009, 2010 and 2012 granted by Italian Neurological Society * “Progetto Giovani”, 2007 granted by Italian League against epilepsy |
| * Award for the best poster, Società Italiana di Neurologia, 2007, abstract titled: “Rare mtDNA variants in Leber’s hereditary optic neuropathy families with recurrence of myoclonus” * Award for the best paper presentation, ‘Annual Meeting of the Italian Society of Neuropathology, 2009, abstract entitled “Melanopsin containing retinal ganglion cells are partially spared by neurodegeneration in Leber’s hereditary optic neuropathy” * Award for the best poster, XI Congress of the European Biological Rhythms Society, 2009, Strasbourg, France, abstract entitled “Circadian Photoreception in Mitochondrial Optic Neuropathies: relative sparing of Melanopsin-Containing Retinal Ganglion Cells” * Award “Franco Michele Puca” for the best contribution of a young scientist, XIX AIMS meeting, Bologna November 15-18th 2009, paper titled: “Circadian photoreception in hereditary optic neuropathies” * Travel grantfor the Association for Research in Vision and Ophthalmology (ARVO) meeting, 2-6th may 2010, Fort Lauderdale, Miami * Award for the best oral presentation at the XLV Congresso AINP e XXXV AIRIC, Bologna, 3-6 Giugno 2010, with the paper “Melanopsin containing retinal ganglion cells are partially spared by neurodegeneration in Leber’s hereditary optic neuropathy” * International Scholarship Award for the paper titled: “Idebenone treatment in Leber’s hereditary optic neuropathy” at the 63rd AAN annual meeting, Hawaii, 9th-16th April 2011 * “Stockgrand Travel Award Grant”, XII Congress of European Biological Rhythms Society Meeting”, Oxford, 20-26th August 2011 * Award at the young investigator symposium “New tendencies in sleep medicine”, XXI Congresso Nazionale AIMS, Pavia, October 2-5th 2011 * Selected by “Magnifico Rettore” (Prof. Ivano Dionigi) of the University of Bologna for a speech at the opening ceremony, Academic Year 2012-2013 * “Marco Polo Programme” grant winner supporting a 6-months period in the USA with a research project on “Melanopsin retinal ganglion cells in neurodegenerative disorders and aging”, march 2014-september 2014 |

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| **publications:** |

1)     **La Morgia C**, Mondini S, Guarino M, Bonifazi F, Cirignotta F. “Busulfan neurotoxicity: a case report”. *Neurol Sci* 2004; 25(2):95-97. [IF 2004: 1.059]

2)     Barboni P, Savini G, Valentino ML, **La Morgia C**, Bellusci C, De Negri AM, Sadun F, Carta A, Carbonelli M, Sadun A, and Carelli V. Leber's hereditary optic neuropathy with childhood onset. *Invest Ophthalmol Vis Sci* 2006;47(12):5303-5309. [IF 2006: 3.766]

3)     Vetrugno R, **La Morgia C**, D'Angelo R, Loi D, Provini F, Plazzi G, Montagna P. Augmentation of restless legs syndrome with long-term tramadol treatment. *Mov Disord* 2007;22(3):424-427. [IF 2007: 3.207]

4)     Carelli V, Franceschini F, Venturi S, Barboni P, Savini G, Barbieri G, Pirro E, **La Morgia C**, Valentino ML, Zanardi F, Violante FS, Mattioli S. Grand Rounds: Could Occupational Exposure to n-Hexane and Other Solvents Precipitate Visual Failure in Leber Hereditary Optic Neuropathy? *Environ Health Perspect* 2007;115(1):113-115. [IF 2007: 5.636]

5)     Carelli V, **La Morgia C**, Iommarini L, Carroccia R, Mattiazzi M, Sangiorgi S, Farnè S, Maresca A, Foscarini B, Lanzi L, Amadori M, Bellan M, Valentino ML. Mitochondrial optic neuropathies: how two genomes may kill the same cell type? *Biosci Rep* 2007;27(1-3):173-184. [IF 2007: 3.115]

6)     Hudson G, Carelli V, Spruijt L, Gerards M, Mowbray C, Achilli A, Pyle A, Elson J, Howell N, **La Morgia C**, Valentino ML, Huoponen K, Savontaus ML, Nikoskelainen E, Sadun AA, Salomao SR, Belfort R Jr, Griffiths P, Man PY, de Coo RF, Horvath R,Zeviani M, Smeets HJ, Torroni A, Chinnery PF. Clinical expression of Leber hereditary optic neuropathy is affected by the mitochondrial DNA-haplogroup background. *Am J Hum Genet* 2007;81(2):228-233. [IF 2007: 11.092]

7)     Amati-Bonneau P, Valentino ML, Reynier P, Gallardo ME, Bornstein B, Boissière  A, Campos Y, Rivera H, de la Aleja JG, Carroccia R, Iommarini L, Labauge P, Figarella-Branger D, Marcorelles P, Furby A, Beauvais K, Letournel F, Liguori R, **La Morgia C**, Montagna P, Liguori M, Zanna C, Rugolo M, Cossarizza A, Wissinger B, Verny C, Schwarzenbacher R, Martín MA, Arenas J, Ayuso C, Garesse R, Lenaers G, Bonneau D, Carelli V. OPA1 mutations induce mitochondrial DNA instability and optic atrophy 'plus' phenotypes. *Brain* 2008;131(Pt 2):338-351. [IF 2008: 9.603]

8)     **La Morgia C**, Achilli A, Iommarini L, Barboni P, Pala M, Olivieri A, Zanna C, Vidoni S, Tonon C, Lodi R, Vetrugno R, Mostacci B, Liguori R, Carroccia R, Montagna P, Rugolo M, Torroni A, Carelli V. Rare mtDNA variants in Leber hereditary optic neuropathy families with recurrence of myoclonus. *Neurology* 2008;70(10):762-770. [IF 2008: 7.043]

9)     **La Morgia C**, Parchi P, Capellari S, Lodi R, Tonon C, Rinaldi R, Mondini S, Cirignotta F 'Agrypnia excitata' in a case of sporadic Creutzfeldt-Jakob disease VV2. *J Neurol Neurosurg Psychiatry* 2009;80(2):244-6. [IF 2009: 4.869]

10)   Carelli V, **La Morgia C**, Valentino ML, Barboni P, Ross-Cisneros FN, Sadun AA. Retinal ganglion cells neurodegeneration in mitochondrial inherited disorders. *Biochim Biophys Acta* 2009;1787(5):518-28. [IF 2009: 3.688]

11)   Vetrugno R, Valentino ML, **La Morgia C**, Liguori R, Stecchi S, Mascalchi M, Fabbri M, Montagna P, Carelli V. Sleep-related periodic respiration with central sleep apnea in Leber Hereditary Optic Neuropathy (LHON). *Sleep Med* 2010;11(4):426-7. [IF 2011: 3.430]

12)   Cevoli S, Pallotti F, **La Morgia C**, Valentino ML, Pierangeli G, Cortelli P, Baruzzi A, Montagna P, Carelli V. High frequency of migraine-only patients negative for the 3243 A>G tRNALeu mtDNA mutation in two MELAS families. *Cephalalgia* 2010;30(8):919-27. [IF 2010: 4.265]

13)   **La Morgia C**, Ross-Cisneros FN, Sadun AA, Hannibal J, Munarini A, Mantovani V, Barboni P, Cantalupo G, Tozer KR, Sancisi E, Salomao SR, Moraes MN, Moraes-Filho MN, Heegaard S, Milea D, Kjer P, Montagna P, Carelli V. Melanopsin retinal ganglion cells are resistant to neurodegeneration in mitochondrial optic neuropathies. *Brain* 2010;133(Pt 8):2426-38. [IF 2010: 9.232]

14)   **La Morgia C**, Ross-Cisneros FN, Hannibal J, Montagna P, Sadun AA, Carelli V. Melanopsin-expressing retinal ganglion cells: implications for human diseases. *Vision Res* 2011 Jan 28;51(2):296-302. [IF 2011: 2.414]

15)   Lodi R, Tonon C, Valentino ML, Manners D, Testa C, Malucelli E, **La Morgia C**, Barboni P, Carbonelli M, Schimpf S, Wissinger B, Zeviani M, Baruzzi A, Liguori R, Barbiroli B, Carelli V. Defective Mitochondrial Adenosine Triphosphate Production in Skeletal Muscle From Patients With Dominant Optic Atrophy Due to OPA1 Mutations. *Arch Neurol* 2011 Jan;68(1):67-73. [IF 2011: 7.584]

16)   Sadun AA, **La Morgia C**, Carelli V. Leber's Hereditary Optic Neuropathy. *Curr Treat Options Neurol* 2011 Feb;13(1):109-17. [IF 2011: 1.292]

17)   Bonazza S, **La Morgia C**, Martinelli P, Capellari S. Strio-pallido-dentate calcinosis: a diagnostic approach in adult patients. *Neurol Sci* 2011 Aug;32(4):537-45. [IF 2011: 1.315]

18)   Barboni P, Savini G, Parisi V, Carbonelli M, **La Morgia C**, Maresca A, Sadun F, De Negri AM, Carta A, Sadun AA, Carelli V. Retinal Nerve Fiber Layer Thickness in Dominant Optic Atrophy Measurements by Optical Coherence Tomography and Correlation with Age. *Ophthalmology* 2011 Oct;118(10):2076-80 [IF 2011: 5.454]

19)   Licchetta L, Bisulli F, Di Vito L, **La Morgia C**, Naldi I, Volta U, Tinuper P. Epilepsy in coeliac disease: not just a matter of calcifications. *Neurol Sci* 2011 Dec;32(6):1069-74 [IF 2011: 1.315]

20)   Carelli V, **La Morgia C**, Valentino ML, Rizzo G, Carbonelli M, De Negri AM, Sadun F, Carta A, Guerriero S, Simonelli F, Sadun AA, Aggarwal D, Liguori R, Avoni P, Baruzzi A, Zeviani M, Montagna P, Barboni P. Idebenone Treatment In Leber's Hereditary Optic Neuropathy. *Brain* 2011 Sep;134(Pt 9):e188  [IF 2011: 9.457]

21)   Achilli A, Iommarini L, Olivieri A, Pala M, Hooshiar Kashani B, Reynier P, **La Morgia C**, Valentino ML, Liguori R, Pizza F, Barboni P, Sadun F, De Negri AM, Zeviani M, Dollfus H, Moulignier A, Ducos G, Orssaud C, Bonneau D, Procaccio V, Leo-Kottler B, Fauser S, Wissinger B, Amati-Bonneau P, Torroni A, Carelli V. Rare primary mitochondrial DNA mutations and probable synergistic variants in Leber's hereditary optic neuropathy. *PLoS One* 2012;7(8):e42242. [IF 2011: 3.730]

22)   Rizzo G, Tozer KR, Tonon C, Manners D, Testa C, Malucelli E, Valentino ML, **La Morgia C**, Barboni P, Randhawa RS, Ross-Cisneros FN, Sadun AA, Carelli V, Lodi R. Secondary Post-Geniculate Involvement in Leber's Hereditary Optic Neuropathy. *PLoS One* 2012;7(11):e50230. [IF 2012: 3.730]

23)   Sadun AA, **La Morgia C**, Carelli V. Leber’s hereditary optic neuropathy: new quinone therapies change the paradigm. *Expert Rev Ophthalmol* 2012; 7(3): 251–259

24)   **La Morgia C**, Barboni P, Rizzo G, Carbonelli M, Savini G, Scaglione C, Capellari S, Bonazza S, Giannoccaro MP, Calandra-Buonaura G, Liguori R, Cortelli P, Martinelli P, Baruzzi A, Carelli V. Loss of temporal retinal nerve fibers in Parkinson disease: a mitochondrial pattern? *Eur J Neurol* 2013, 20: 198–201 [IF 2013: 3.692]

25)   Maresca A, **La Morgia C**, Caporali L, Valentino ML, Carelli V. The optic nerve: A "mito-window" on mitochondrial neurodegeneration. *Mol Cell Neurosci* 2013 Jul;55:62-76 [IF 2013: 3.734]

26)   Caporali L, Ghelli AM, Iommarini L, Maresca A, Valentino ML, **La Morgia C**, Liguori R, Zanna C, Barboni P, De Nardo V, Martinuzzi A, Rizzo G, Tonon C, Lodi R, Calvaruso MA, Cappelletti M, Porcelli AM, Achilli A, Pala M, Torroni A, Carelli V. Cybrid studies establish the causal link between the mtDNA m.3890G>A/MT-ND1 mutation and optic atrophy with bilateral brainstem lesions. *Biochim Biophys Acta* 2013 Mar; 1832 (3):445-52. [IF 2013: 5.089]

27)   Carelli V, **La Morgia C**, Sadun AA. Mitochondrial dysfunction in optic neuropathies: animal models and therapeutic options. *Curr Opin Neurol* 2013 Feb; 26 (1):52–58 [IF 2013:5.729]

28)   Barboni P, Valentino ML, **La Morgia C**, Carbonelli M, Savini G, De Negri A, Simonelli F, Sadun F, Caporali L, Maresca A, Liguori R, Baruzzi A, Zeviani M and Carelli V. Idebenone treatment in patients with OPA1-mutant dominant optic atrophy. *Brain* 2013 Feb;136(Pt 2):e231. [IF 2013: 10.226]

29)   Barboni P, Carelli V, Savini G, Carbonelli M, **La Morgia C**, Sadun AA. Microcystic macular degeneration from optic neuropathy: not inflammatory, not trans-synaptic degeneration. *Brain 2013*Jul;136(Pt 7):e239*.* [IF 2013: 10.226]

30)   Sadun AA, **La Morgia C**, Carelli V. Mitochondrial optic neuropathies: our travels from bench to bedside and back again An expansion of the 35(th) Susan Alper Memorial lecture by Alfredo A. Sadun, MD, PhD given at the Washington Hospital Medical Center on April 28, 2012. *Clin Experiment Ophthalmol* 2013 Sep-Oct; 41(7):702-712 [IF 2013: 1.953]

31)   Moura AL, Nagy BV, **La Morgia C**, Barboni P, Oliveira AG, Salomão SR, Berezovsky A, de Moraes-Filho MN, Chicani CF, Belfort R Jr, Carelli V, Sadun AA, Hood DC, Ventura DF. The Pupil Light Reflex in Leber's Hereditary Optic Neuropathy: Evidence for Preservation of Melanopsin-Expressing Retinal Ganglion Cells. *Invest Ophthalmol Vis Sci* 2013 Jul 2;54(7):4471-7. [IF 2013: 3.661]

32)   Ziccardi L, Sadun F, De Negri AM, Barboni P,5, Savini G, Borrelli E, **La Morgia C,**Carelli V, Parisi V.Retinal function and neural conduction along the visual pathways in affected and unaffected carriers with Leber’s hereditary optic neuropathy.*Invest Ophthalmol Vis Sci* 2013, Oct 21;54(10):6893-901 [IF 2013: 3.661]

33)   Sadun AA, **La Morgia C**, Carelli V. Mitochondrial optic neuropathies: additional facts and concepts - response. *Clin Experiment Ophthalmol* 2014 Mar; 42(2):207-8 [IF 2014: 2.347]

34)   Guaraldi P, Sancisi E, **La Morgia C**, Calandra-Buonaura G, Carelli V, Cameli O, Battistini A, Cortelli P, Piperno R. Nocturnal melatonin regulation in post-traumatic vegetative state: a possible role for melatonin supplementation? *Chronobiol International* 2014 Jun;31(5):741-5 [IF 2014: 3.343]

35)   Kaveh Moghadam K, Pizza F, Tonon C, Lodi R, Carelli V, Poli F, Franceschini C, Barboni P, Seri M, Ferrari S, **La Morgia C**, Testa C, Cornelio F, Liguori R, Winkelmann J, Lin L, Mignot E, Plazzi G. Polysomnographic and neurometabolic features may mark preclinical autosomal dominant cerebellar ataxia, deafness, and narcolepsy due to a mutation in the DNA (cytosine-5-)-methyltransferase gene, DNMT1. *Sleep Medicine* 2014 May; 15(5):582-5 [IF 2014:3.154]

36)   Kaveh Moghadam K, Pizza F, **La Morgia C**, Franceschini C, Tonon C, Lodi R, Barboni P, Seri M, Ferrari S, Liguori R, Donadio V, Parchi P, Cornelio F, Inzitari D, Mignarri A, Capocchi G, Dotti MT, Winkelmann J, Lin L, Mignot E, Carelli V, Plazzi G. Narcolepsy is a common phenotype in both HSAN IE and ADCA-DN. *Brain* 2014 Jun; 137(Pt 6):1634-55 [IF 2014: 9.196]

37)   Carossa V, Ghelli A, Tropeano CV, Valentino ML, Iommarini L, Maresca A, Caporali L, **La Morgia C**, Liguori R, Barboni P, Carbonelli M, Rizzo G, Tonon C, Lodi R, Martinuzzi A, De Nardo V, Rugolo M, Ferretti L, Gandini F, Pala M, Achilli A, Olivieri A, Torroni A, Carelli V. A Novel in-Frame 18-bp Microdeletion in MT-CYB Causes a Multisystem Disorder with Prominent Exercise Intolerance. *Hum  Mutat*. 2014 Aug;35(8):954-8 [IF 2014: 5.144]

38)   **La Morgia C**, Caporali L, Gandini F, Olivieri A, Toni F, Nassetti S, Brunetto D, Stipa C, Scaduto C, Parmeggiani A, Tonon C, Lodi R, Torroni A, Carelli V. Association of the mtDNA m.4171C>A/MT-ND1 mutation with both optic neuropathy and bilateral brainstem lesions. *BMC Neurol*. 2014 May 28;14:116. [IF 2014: 2.040]

39)   Barboni P, Savini G, Cascavilla ML, Caporali L, Milesi J, Borrelli E, **La Morgia C**, Valentino ML, Triolo G, Lembo A, Carta A, De Negri A, Sadun F, Rizzo G, Parisi V, Pierro L, Bianchi Marzoli S, Zeviani M, Sadun AA, Bandello F, Carelli V. Early Macular Retinal Ganglion Cell Loss in Dominant Optic Atrophy: Genotype-Phenotype Correlation. *Am J Ophthalmol*. 2014 Sep; 158(3):628-636 [IF 2014: 3.871]

40)   **La Morgia C**, Carbonelli M, Barboni P, Sadun AA, Carelli V. Medical management of hereditary optic neuropathies. *Front Neurol*. 2014 Jul 31;5:141 [IF 2015: 3.184].

41)   Maresca A, Caporali L, Strobbe D, Zanna C, Malavolta D, **La Morgia C**, Valentino ML, Carelli V. Genetic Basis of Mitochondrial Optic Neuropathies. *Curr Mol Med*. 2014;14(8):985-992. [IF 2014: 3.621]

42)   Carelli V, Sabatelli M, Carrozzo R, Rizza T, Schimpf S, Wissinger B, Zanna C, Rugolo M, **La Morgia** **C**, Caporali L, Carbonelli M, Barboni P, Tonon C, Lodi R, Bertini E. 'Behr syndrome' with OPA1 compound heterozygote mutations. *Brain* 2015 Jan;138(Pt 1):e321. [IF 2015: 10.103]

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