BIOGRAPHICAL SKETCH

NAME: **Carlo Viscomi**

Researcher unique identifier (ORCID): 0000-0001-6050-0566

POSITION TITLE: Associate Professor of Genetics

EDUCATION/TRAINING

INSTITUTION AND LOCATION	DEGREE *(if applicable)*	Start Date MM/YYYY	Completion Date MM/YYYY	FIELD OF STUDY
University of Milan, Italy	B.Sc.+M.Sc.	1993	1999	Biological Sciences
University of Milan, Italy	PhD	1999	2002	Physiology
Neurological Institute "C. Besta", Milan, Italy	Postdoctoral fellow	2002	2004	Mitochondrial Medicine

A. Personal Statement

I entered the mitochondrial medicine field in 2004 as a postdoc in dr Massimo Zeviani's lab at the Neurological Institute "C. Besta" in Milan, Italy, where in 2009 I was appointed as a junior group leader under the supervision of Dr. Massimo Zeviani. My main research interest since then has been focused on translational aspects with the ultimate goals of clarifying the biological basis of human diseases and developing innovative and effective therapies. To this end, I developed a panel of animal models of mitochondrial disease and characterized them by using several techniques ranging from in vivo tests to investigate the neurometabolic basis of the diseases to in vitro approaches based on metabolomics and proteomics to clarify the metabolic consequences of the deletion of genes responsible for human mitochondrial diseases and investigating the interactome to understand their physiological role. Building on the knowledge of the mechanisms leading to disease, I developed new therapeutic approaches using both pharmacological and gene therapy strategies. The main achievements of these researches have been (i) the discovery of the pathogenetic mechanism of ethylamalonic encephalopathy (EE), i.e. the accumulation of the powerful cytochrome c oxidase inhibitor hydrogen sulfide (H2S) (Tiranti et al, Nat Med, 2009) (ii) the development of a therapy based on N-acetylcysteine and metronidazole highy effective in the treatment of EE in mice and, most importantly, patients (Viscomi et al, Nat Med, 2010) (iii) the discovery that the stimulation of the PGC1alpha-dependent mitochondriogenic pathway by using either the AMPK agonist AICAR or the NAD+ precursor Nicotinamide Riboside (NR) was effective in ameliorating the phenotype in mouse models of cytochrome c oxidase deficiency (Viscomi et al, Cell Metab, 2011; Cerutti et al, Cell Metab, 2013) (iv) the development of AAV-mediated gene therapy approaches to treat mitochondrial diseases due to accumulation of toxic compounds, such as EE and mitochondrial gastro-intestinal-encephalomyopathy (MNGIE) (Di Meo et al, EMBO Mol Med, 2012; Torres-Torronteras et al, Mol Ther, 2014). More recently, my laboratory has demonstrated the potential of AAV-based gene therapy in other mitochondrial diseases due to defects in nuclear genes (Bottani et al, Mol Ther, 2014, Di Meo et al, Gene Therapy, 2017, Pinheiro et al, Mol Ther, 2020, Corrà et al, Brain, 2022). Finally, in collaboration with Michal Minczuk, MBU, Cambridge, UK, he helped develop an AAV-based approach to correct specific mtDNA mutations through the use of Zinc-finger Nucleases (Gammage et al Nat Med, 2018). These studies constitute proof of fundamental principle for the transfer of these therapies to humans in the years to come.

Other studies that we carried out in my laboratory were aimed at studying the possibility of by-passing respiratory chain defects through the use of alternative oxidases (Dogan et al, Cell Metab, 2018), defining the mechanism by which rapamycin improves the phenotype of various mouse models of mitochondrial

disease (Civiletto et al, EMBO Mol Med 2018), and the possibility to shape mitochondrial cristae to correct mitochondrial defects (Civiletto et al, Cell Metab, 2015; Luna-Sanchez et al, Mol Ther, 2020)
Finally, I have collaborated on numerous studies to define the functional role of various disease genes (Signes et al, EMBO Mol Med, 2019, Bottani et al, Mol Ther, 2017, Brunetti et al, EMBO Mol Med 2016).

These studies took advantage of a number of collaborations in Italy (Prof Andrea Ballabio, Alberto Auricchio at TIGEM, Naples; Dr Massimo Lasorsa, University of Bari; Prof Luca Scorrano, University of Padova) and abroad (Prof Anthony Moore, Sussex University, UK, Prof Johan Auwerx, EPFL, Lausanne, Switzerland; Dr Michio Hirano and Eric Schon, Columbia University, NY; Dr Ramon Marti', Vall D'Hebron Research Institute, Barcelona, Spain; Prof Howy Jacobs, Institute of Biotechnology, University of Helsinki, Finland, Prof Anu Suomalainen, Research Program for Molecular Neurology, University of Helsinki, Finland), Professors: Judy Hirst, Michal Minczuk and Mike Murphy, MBU, Cambridge, Patrick Chinnery, University of Cambridge, UK).

B. Positions, Scientific Appointments and Honors

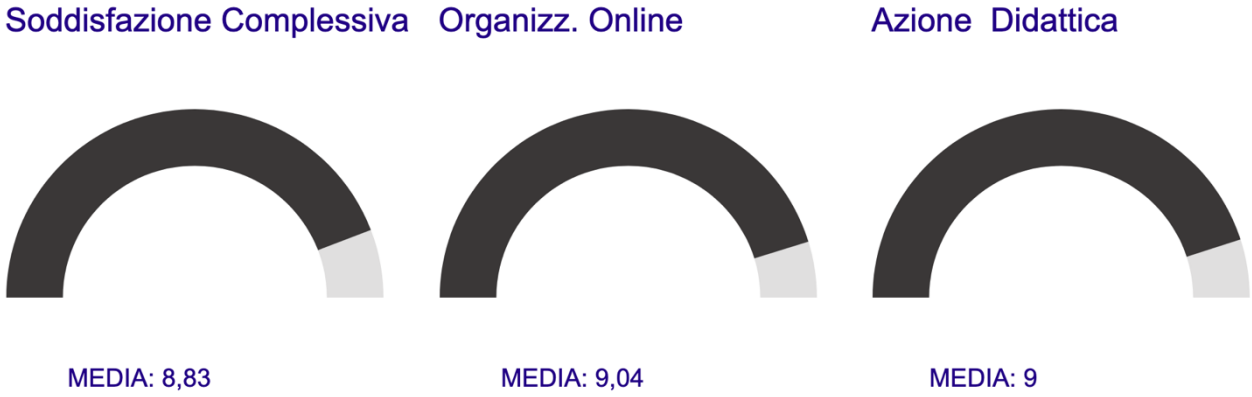
2004-2009: Postdoctoral Fellow IRCCS Foundation Neurological Institute “C. Besta”.
2009-2013: Staff Scientist, IRCCS Foundation Neurological Institute “C. Besta”.
2013-2019: Senior Investigating Scientist, MRC - Mitochondrial Biology Unit, Cambridge, UK.
2020-present: Associate Professor of Genetics, University of Padova, IT

Honors
Kelsey Wright Award for Excellence in Mitochondrial Medicine given by the United Mitochondrial Disease Foundation (Scottsdale, AZ, 2010).

Memberships of scientific societies
2020-present: member of the Italian Genetics Society (AGI)
2022-present: founder and President of the European Society for Mitochondrial Medicine and Research (E-Mit)

Teaching activities

2020-present: Epigenetics and Epigenomics, Degree in Molecular Biology University of Padova, Italy
This course started in 2020/21. Since then, it has been highly appreciated by the students. The following graphs refer to AA21/22 (currently not available for the 22/23 due to maintenance of the system):



In the same year the average scores for the Master in Molecular Biology were:

Corso di Studio MOLECULAR BIOLOGY - Padova

Questionari *Entrambe le tipologie di frequenza*

Attività didattiche-Docente (AD) valutate	% su valutabili	Questionari compilati (inclusi i non frequentanti*)	Soddisfazione complessiva: Nr risposte valide	Media	Mediana	Azione didattica: Nr risposte valide	Media	Mediana
28	96,6%	848	766	7,8	7,76	758	7,79	7,82

**I questionari dei non frequentanti non presentano le domande che contribuiscono al calcolo degli Indicatori Soddisfazione complessiva e Azione didattica.*

2020-present: Animal Models in Genetic Research, University of Padova, Italy. This is specialized course in collaboration with Université Paris Cité, lasting one week in the form of a series of seminars.
2020-present: Physiology, University of Padova, Italy. This course is for the students in Nursing Sciences.
From 2024:
Human Genetics in the course of Biology of Human and Environmental Health, University of Padova
Metabolic Disorders in the course of Molecular Biology, curriculum in Bioenergetics, University of Padova.

Supervision of graduate students and postdoctoral fellows
Past PhD students:
Emanuela Bottani, now Assistant Professor at Univeristy of Verona);
Gabriele Civiletto, now Group Leader at Nestlè Institute, Lausanne, Switzerland,
Alba Signes-Marrahi, now in the private sector
Anabel Martinez-Lyons, now postdoc at University of Glasgow, UK) and
Pedro Silva-Pinheiro, now postdoctoral fellow at University of Cambridge/MRC-Mitochondrial Biology Unit, UK.
Raffaele Cerutti, now postdoc at University of Padova
Current PhD students: Giacomo Giacchin, Alessandro Zuppardo and Enea Vogrig, Department of Biomedical Sciences, University of Padova, Italy
Past Postdocs:
Ivano di Meo, now Biologist at Fondazione IRCCS Neurological Instiute “C. Besta”, Milan, Italy
Tatiana Gorletta, now in the private sector
Serena Barbaro, now in the private sector
Sukru Anil Dogan, now Professor of Molecular Biology, Bosphorus University, Istanbul, Turkey
Dario Brunetti, now RTDB at University of Milano
Marta Luna Sanchez, now postdoc at Universidad Autonoma di Barcelona, Spain

Editorial activity
Reviewer for: Nature, Cell Metabolism, Nature Communications, Nature Neuroscience, BBA, Human Molecular Genetics, EMBO Molecular Medicine, Current Biology.

Organisation of scientific meetings
2022-present: Co-organizer of the “Mitomeetings”, series of seminars for at University of Padova
2024: Winter School in Mitochondrial Biomedicine, Bressanone, Italy (January 28-February 02)

Commissions of trust
2021-present: member of the Scientific Board of the Italian Association of Mitochondrial Patients (Mitocon)
2022-present member of the Scientific Board of AFM-Telethon, France

National Scientific habilitation (ASN)
Habilitated as full Professor (Professore Ordinario) in Genetics (SSD: BIO/18: valid from 18/11/2020 to 18/11/2031).

INVITED TALKS
EMBO WORKSHOP: MOLECULAR BIOLOGY OF MITOCHONDRIAL GENE MAINTENANCE AND EXPRESSION, 19-23 May 2024, Poland
Lectio Magistralis, University of Bologna, March 2024
Lectio Magistralis, IRCCS Burlo Garofolo, Trieste, February 2024

UCLA Mitochondrial Symposium, UCLA, CA, USA, December 7-9 2023
Genomit, Pisa, November 17-18 2023
Euromit, Bologna, Italy, 2023
Neuromuscular Translational Research Conference, London, 2023
Mitocon: annual symposium of the Italian association of the mitochondrial patients, Rome, 2022
MitoNice, Nice, France, AFM-Telethon meeting, 2022
Mitocon: annual symposium of the Italian association of the mitochondrial patients, Rome, 2021
Bosphorus University, Instanbul, 2021 (host: Prf. Anil Dogan)
University of Sussex, Brighton, UK, 2019 (host: Prof Tony Moore)
Ebec 2018, Budapest 25-30 August 2018
Emerging concepts in Mitochondrial Biology, Weizman Institute, Reohovot, Israel 4-8 February 2018
Mitochondrial Medicine, Cambridge, UK 9-11 May 2018
VII National Meeting on Mitochondrial Diseases, Milan 22-24 September 2017
Mitox, Oxford, UK 2 December 2015
Institute of Pharmacological Sciences “Mario Negri”, Milan, Italy October 2015
Nuffield Dept Obstetrics and Gynaecology, University of Oxford 2014
Yearly seminars at the IRCCS Foundation Neurological Institute “C. Besta” in 2014-2018.

Current Research Support

Title: AAV-based gene therapy for mitochondrial diseases (30K€)
Duration: 2022-2024
Granting Agency: Department of Biomedical Sciences, University of Padova,
Role: PI
Title: MitoFight2 (120K€),
Duration: 2022-2024,
Granting Agency: Associazione Luigi Comini Onlus,
Role: PI
Title: MitMed: identification and characterization of new disease genes for mitochondrial disorders. (236.5K€), *Duration*: 2021-2024,
Granting Agency: Telethon (Project 20013),
Role: PI
Title: MITOPHAGYTREAT: harnessing mitophagy to treat mitochondrial myopathies. (300€)
Duration: 2021-2023,
Granting Agency: AFM-Telethon (Project 23706),
Role: PI
Title: PNRR - PE - AGE-IT - SP. 2 (160K€),
Duration: 2023-2026,
Granting agency: EU (Grant: PE_00000015 - SP. 2),
Role: Co-PI
Title: PNRR - CN - G.T.RNA SP. 1 (120K€),
Duration: 2023-2026,
Granting agency: EU (Grant: PE_00000015 - SP. 2),
Role: Co-PI

Past Research Support

Title: MTPHAGYTREAT: harnessing mitophagy to treat mitochondrial myopathies. (186€)
Duration: 2021-2023,
Granting Agency: Marie Curie Actions (Project 101023390),
Role: PI
Title: Mito-ND: Mitochondrial Neurodegeneration
Duration: 2016-2018
Granting agency: CoEN (Centres of Excellence in Neurodegeneration); £300K
Role: CO-PI

Title: MitCare. Mitochondrial medicine: developing treatments of OXPHOS-defects in recombinant mammalian models.
Duration: 2013-2018
Granting Agency: ERC (FP7-322424); 2500 k€.
Role: Co-investigator
Title: MitoCure: developing treatments for mitochondrial diseases.
Duration: 2013-2016
Granting Agency: Italian Ministry of Health (GR-2010-2306756); 450K€.
Role: PI
Title: Therapeutic strategies for the cure of Ethylmalonic Encephalopathy, a severe infantile mitochondrial disease
Duration: 2012-2013
Granting Agency: AFM (160 K€)
Role: Co-investigator

C. Contributions to Science

H index Scopus: 42
H index Google Scholar: 46

Full list of publications (highlights in bold)

1. Peruzzotti-Jametti L, Willis CM, Krzak G, Hamel R, Pirvan L, Ionescu RB, Reisz JA, Prag HA, Garcia-Segura ME, Wu V, Xiang Y, Barlas B, Casey AM, van den Bosch AMR, Nicaise AM, Roth L, Bates GR, Huang H, Prasad P, Vincent AE, Frezza C, **Viscomi C**, Balmus G, Takats Z, Marioni JC, D'Alessandro A, Murphy MP, Mohorianu I, Pluchino S. Mitochondrial complex I activity in microglia sustains neuroinflammation. Nature. 2024 Apr;628(8006):195-203

2. Balmaceda V, Komlódi T, Szibor M, Gnaiger E, Moore AL, Fernandez-Vizarra E, **Viscomi C**. The striking differences in the bioenergetics of brain and liver mitochondria are enhanced in mitochondrial disease. Biochim Biophys Acta Mol Basis Dis. 2024 Mar;1870(3):167033.

3. Nicol T, Falcone S, Blease A, Vikhe P, Civiletto G, Omairi SS, **Viscomi C**, Patel K, Potter PK. Tissue-specific differences in the assembly of mitochondrial Complex I are revealed by a novel ENU mutation in ECSIT. Cardiovasc Res. 2023 Oct 16;119(12):2213-2229

4. Keshavan N, Minczuk M, Viscomi C, Rahman S. Gene therapy for mitochondrial disorders. J Inherit Metab Dis. 2024 Jan;47(1):145-175. doi: 10.1002/jimd.12699. Epub 2024 Jan 3. PMID: 38171948.

5. Brischigliaro M, Cabrera-Orefice A, Arnold S, Viscomi C, Zeviani M, Fernández-Vizarra E. Structural rather than catalytic role for mitochondrial respiratory chain supercomplexes. Elife. 2023 Oct 12;12:RP88084. doi: 10.7554/eLife.88084. PMID: 37823874; PMCID: PMC10569793.

6. Corrà S, Checchetto V, Brischigliaro M, Rampazzo C, Bottani E, Gagliani C, Cortese K, De Pittà C, Roverso M, De Stefani D, Bogialli S, Zeviani M, Viscomi C, Szabò I, Costa R. Drosophila Mpv17 forms an ion channel and regulates energy metabolism. iScience. 2023 Sep 16;26(10):107955. doi: 10.1016/j.isci.2023.107955. PMID: 37810222; PMCID: PMC10558772.

7. Di Donfrancesco A, Berlingieri C, Giacomello M, Frascarelli C, Magalhaes Rebelo AP, Bindoff LA, Reeval S, Renbaum P, Santorelli FM, Massaro G, Viscomi C, Zeviani M, Ghezzi D, Bottani E, Brunetti D. PPAR-gamma agonist pioglitazone recovers mitochondrial quality control in fibroblasts from PITRM1deficient patients. Front Pharmacol. 2023 Jul 26;14: 1220620. doi: 10.3389/fphar.2023.1220620. PMID: 37576821; PMCID: PMC10415619.

8. Nicol T, Falcone S, Blease A, Vikhe P, Civiletto G, Omairi SS, Viscomi C, Patel K, Potter PK. Tissue-specific differences in the assembly of mitochondrial Complex I are revealed by a novel

ENU mutation in ECSIT. Cardiovasc Res. 2023 Oct 16;119(12):2213-2229. doi: 10.1093/cvr/cvad101. PMID: 37395010; PMCID: PMC10578914.

9. **Viscomi C, van den Ameele J, Meyer KC, Chinnery PF. Opportunities for mitochondrial disease gene therapy. Nat Rev Drug Discov. 2023 Jun;22(6):429-430. doi: 10.1038/d41573-023-00067-z. PMID: 37106085**.

   Beltrà M, Pöllänen N, Fornelli C, Tonttila K, Hsu MY, Zampieri S, Moletta L, Corrà S, Porporato PE, Kivelä R, Viscomi C, Sandri M, Hulmi JJ, Sartori R, Pirinen E, Penna F. NAD⁺ repletion with niacin counteracts cancer cachexia. Nat Commun. 2023 Apr 3;14(1):1849. doi: 10.1038/s41467-023-37595-6. PMID: 37012289; PMCID: PMC10070388.

10. Brischigliaro M, Fernandez-Vizarra E, Viscomi C. Mitochondrial Neurodegeneration: Lessons from Drosophila melanogaster Models. Biomolecules. 2023 Feb 16;13(2):378. doi: 10.3390/biom13020378. PMID: 36830747; PMCID: PMC9953451.

11. Viscomi C, Zeviani M. Experimental therapy for mitochondrial diseases. Handb Clin Neurol. 2023;194:259-277. doi: 10.1016/B978-0-12-821751-1.00013-0. PMID: 36813318.

12. Legati A, Ghezzi D, Viscomi C. Mitochondrial DNA Sequencing and Heteroplasmy Quantification by Next Generation Sequencing. Methods Mol Biol. 2023;2615:381-395. doi: 10.1007/978-1-0716-2922-2_26. PMID: 36807805.

13. Viscomi C, Moore AL, Zeviani M, Szibor M. Xenotopic expression of alternative oxidase (AOX) to study mechanisms of mitochondrial disease. Biochim Biophys Acta Bioenerg. 2023 Apr 1;1864(2):148947. doi: 10.1016/j.bbabio.2022.148947. Epub 2022 Dec 6. PMID: 36481273.

14. **Corrà S, Cerutti R, Balmaceda V, Viscomi C*, Zeviani M. Double administration of self-complementary AAV9NDUFS4 prevents Leigh disease in Ndufs4-/- mice. Brain. 2022 Oct 21;145(10):3405-3414. doi: 10.1093/brain/awac182. PMID: 36270002; PMCID: PMC9586549.**

15. **Falabella M, Minczuk M, Hanna MG, Viscomi C, Pitceathly RDS. Gene therapy for primary mitochondrial diseases: experimental advances and clinical challenges. Nat Rev Neurol. 2022 Nov;18(11):689-698. doi: 10.1038/s41582-022-00715-9. Epub 2022 Oct 18. PMID: 36257993**.

16. Allen FM, Costa ASH, Gruszczyk AV, Bates GR, Prag HA, Nikitopoulou E, Viscomi C, Frezza C, James AM, Murphy MP. Rapid fractionation of mitochondria from mouse liver and heart reveals in vivo metabolite compartmentation. FEBS Lett. 2023 Jan;597(2):246-261. doi: 10.1002/1873-3468.14511. Epub 2022 Oct 27. PMID: 36217875; PMCID: PMC7614208.

17. Szibor M, Heyne E, Viscomi C, Moore AL. Measuring the Mitochondrial Ubiquinone (Q) Pool Redox State in Isolated Respiring Mitochondria. Methods Mol Biol. 2022;2497:291-299. doi: 10.1007/978-1-0716-2309-1_19. PMID: 35771450.

18. Viscomi C, Soriano ME. Molecular Research on Mitochondrial Dysfunction. Int J Mol Sci. 2022 Jun 20;23(12):6845. doi: 10.3390/ijms23126845. PMID: 35743286; PMCID: PMC9224555.

19. Brischigliaro M, Cabrera-Orefice A, Sturlese M, Elurbe DM, Frigo E, Fernandez-Vizarra E, Moro S, Huynen MA, Arnold S, Viscomi C, Zeviani M. CG7630 is the Drosophila melanogaster homolog of the cytochrome c oxidase subunit COX7B. EMBO Rep. 2022 Aug 3;23(8):e54825. doi: 10.15252/embr.202254825. Epub
20. 2022 Jun 14. PMID: 35699132; PMCID: PMC9346487.

21. Brischigliaro M, Badocco D, Costa R, Viscomi C, Zeviani M, Pastore P, Fernández-Vizarra E. Mitochondrial Cytochrome c Oxidase Defects Alter Cellular Homeostasis of Transition

Metals. Front Cell Dev Biol. 2022 May19;10:892069. doi: 10.3389/fcell.2022.892069. PMID: 35663391; PMCID: PMC9160823.

22. Brischigliaro M, Frigo E, Fernandez-Vizarra E, Bernardi P, Viscomi C. Measurement of mitochondrial respiratory chain enzymatic activities in Drosophila melanogaster samples. STAR Protoc. 2022 Apr 15;3(2):101322. doi: 10.1016/j.xpro.2022.101322. PMID: 35479112; PMCID: PMC9036317.

23. **Zeviani M, Viscomi C. Mitochondrial Neurodegeneration. Cells. 2022 Feb 11;11(4):637. doi: 10.3390/cells11040637. PMID: 35203288; PMCID: PMC8870525**.

24. Dogan SA, Giacchin G, Zito E, Viscomi C. Redox Signaling and Stress in Inherited Myopathies. Antioxid Redox Signal. 2022 Aug;37(4-6):301-323. doi: 10.1089/ars.2021.0266. Epub 2022 Apr 18. PMID: 35081731.

25. Zhang H, Esposito M, Pezet MG, Aryaman J, Wei W, Klimm F, Calabrese C, Burr SP, Macabelli CH, Viscomi C, Saitou M, Chiaratti MR, Stewart JB, Jones N, Chinnery PF. Mitochondrial DNA heteroplasmy is modulated during oocyte development propagating mutation transmission. Sci Adv. 2021 Dec 10;7(50):eabi5657. doi: 10.1126/sciadv.abi5657. Epub 2021 Dec 8. PMID: 34878831; PMCID: PMC8654302.

26. Silva-Pinheiro P, Pardo-Hernández C, Reyes A, Tilokani L, Mishra A, Cerutti R, Li S, Rozsivalova DH, Valenzuela S, Dogan SA, Peter B, Fernández-Silva P, Trifunovic A, Prudent J, Minczuk M, Bindoff L, Macao B, Zeviani M, Falkenberg M, Viscomi C. Correction to 'DNA polymerase gamma mutations that impair holoenzyme stability cause catalytic subunit depletion'. Nucleic Acids Res. 2021 Oct 11;49(18):10803. doi: 10.1093/nar/gkab837. Erratum for: Nucleic Acids Res. 2021 May 21;49(9):5230-5248. PMID: 34520541; PMCID: PMC8501975.

27. Brunetti D, Catania A, Viscomi C, Deleidi M, Bindoff LA, Ghezzi D, Zeviani M. Role of PITRM1 in Mitochondrial Dysfunction and Neurodegeneration. Biomedicines. 2021 Jul 17;9(7):833. doi: 10.3390/biomedicines9070833. PMID: 34356897; PMCID: PMC8301332.

28. **Silva-Pinheiro P, Pardo-Hernández C, Reyes A, Tilokani L, Mishra A, Cerutti R, Li S, Rozsivalova DH, Valenzuela S, Dogan SA, Peter B, Fernández-Silva P, Trifunovic A, Prudent J, Minczuk M, Bindoff L, Macao B, Zeviani M, Falkenberg M, Viscomi C. DNA polymerase gamma mutations that impair holoenzyme stability cause catalytic subunit depletion. Nucleic Acids Res. 2021 May 21;49(9):5230-5248. doi: 10.1093/nar/gkab282. Erratum in: Nucleic Acids Res. 2021 Oct 11;49(18):10803. PMID: 33956154; PMCID: PMC8136776.**

29. Peruzzo R, Corrà S, Costa R, Brischigliaro M, Varanita T, Biasutto L, Rampazzo C, Ghezzi D, Leanza L, Zoratti M, Zeviani M, De Pittà C, Viscomi C, Costa R, Szabò I. Exploiting pyocyanin to treat mitochondrial disease due to respiratory complex III dysfunction. Nat Commun. 2021 Apr 8;12(1):2103. doi: 10.1038/s41467-021-22062-x. PMID: 33833234; PMCID: PMC8032734.

30. Peruzzotti-Jametti L, Bernstock JD, Willis CM, Manferrari G, Rogall R, Fernandez-Vizarra E, Williamson JC, Braga A, van den Bosch A, Leonardi T, Krzak G, Kittel Á, Benincá C, Vicario N, Tan S, Bastos C, Bicci I, Iraci N, Smith JA, Peacock B, Muller KH, Lehner PJ, Buzas EI, Faria N, Zeviani M, Frezza C, Brisson A, Matheson NJ, Viscomi C, Pluchino S. Neural stem cells traffic functional mitochondria via extracellular vesicles. PLoS Biol. 2021 Apr 7;19(4):e3001166. doi: 10.1371/journal.pbio.3001166. PMID: 33826607; PMCID: PMC8055036.

31. Yin Z, Burger N, Kula-Alwar D, Aksentijević D, Bridges HR, Prag HA, Grba DN, Viscomi C, James AM, Mottahedin A, Krieg T, Murphy MP, Hirst J. Structural basis for a complex I mutation that blocks pathological ROS production. Nat Commun. 2021 Jan 29;12(1):707. doi: 10.1038/s41467-021-20942-w. PMID: 33514727; PMCID: PMC7846746.

32. Filipe A, Chernorudskiy A, Arbogast S, Varone E, Villar-Quiles RN, Pozzer D, Moulin M, Fumagalli S, Cabet E, Dudhal S, De Simoni MG, Denis R, Vadrot N, Dill C, Giovarelli M, Szweda L, De Palma C, Pinton P, Giorgi C, Viscomi C, Clementi E, Missiroli S, Boncompagni S, Zito E, Ferreiro A. Defective endoplasmic reticulum-mitochondria contacts and bioenergetics in SEPN1-related myopathy. Cell Death Differ. 2021 Jan;28(1):123-138. doi: 10.1038/s41418-020-0587-z. Epub2020 Jul 13. PMID: 32661288; PMCID: PMC7853070.

33. Pérez MJ, Ivanyuk D, Panagiotakopoulou V, Di Napoli G, Kalb S, Brunetti D,Al-Shaana R, Kaeser SA, Fraschka SA, Jucker M, Zeviani M, Viscomi C, Deleidi M. Loss of function of the mitochondrial peptidase PITRM1 induces proteotoxic stress and Alzheimer's disease-like pathology in human cerebral organoids. Mol Psychiatry. 2021 Oct;26(10):5733-5750. doi: 10.1038/s41380-020-0807-4. Epub 2020 Jul 7. PMID: 32632204; PMCID: PMC8758476.

34. Luna-Sanchez M, Benincá C, Cerutti R, Brea-Calvo G, Yeates A, Scorrano L, Zeviani M, Viscomi C. Opa1 Overexpression Protects from Early-Onset Mpv17^{-/-}-Related Mouse Kidney Disease. Mol Ther. 2020 Aug 5;28(8):1918-1930. doi: 10.1016/j.ymthe.2020.06.010. Epub 2020 Jun 12. PMID 32562616; PMCID: PMC7403474.

35. Silva-Pinheiro P, Cerutti R, Luna-Sanchez M, Zeviani M, Viscomi C. A Single Intravenous Injection of AAV-PHP.B-hNDUFS4 Ameliorates the Phenotype of Ndufs4-/- Mice. Mol Ther Methods Clin Dev. 2020 May 4;17:1071-1078. doi: 10.1016/j.omtm.2020.04.026. PMID: 32478122; PMCID: PMC7248291.

36. Steele H, Gomez-Duran A, Pyle A, Hopton S, Newman J, Stefanetti RJ, Charman SJ, Parikh JD, He L, Viscomi C, Jakovljevic DG, Hollingsworth KG, Robinson AJ, Taylor RW, Bottolo L, Horvath R, Chinnery PF. Metabolic effects of bezafibrate in mitochondrial disease. EMBO Mol Med. 2020 Mar 6;12(3):e11589. doi: 10.15252/emmm.201911589. Epub 2020 Feb 28. PMID: 32107855; PMCID: PMC7059007.

37. Szibor M, Schreckenberg R, Gizatullina Z, Dufour E, Wiesnet M, Dhandapani PK, Debska-Vielhaber G, Heidler J, Wittig I, Nyman TA, Gärtner U, Hall AR, Pell V, Viscomi C, Krieg T, Murphy MP, Braun T, Gellerich FN, Schlüter KD, Jacobs HT. Respiratory chain signalling is essential for adaptive remodelling following cardiac ischaemia. J Cell Mol Med. 2020 Mar;24(6):3534-3548. doi: 10.1111/jcmm.15043. Epub 2020 Feb 10. PMID: 32040259; PMCID: PMC7131948.

38. Szibor M, Gainutdinov T, Fernandez-Vizarra E, Dufour E, Gizatullina Z, Debska-Vielhaber G, Heidler J, Wittig I, Viscomi C, Gellerich F, Moore AL. Bioenergetic consequences from xenotopic expression of a tunicate AOX in mouse mitochondria: Switch from RET and ROS to FET. Biochim Biophys Acta Bioenerg. 2020 Feb 1;1861(2):148137. doi: 10.1016/j.bbabio.2019.148137. Epub 2019 Dec 9. PMID: 31825809.

39. Boncompagni S, Pozzer D, Viscomi C, Ferreiro A, Zito E. Physical and Functional Cross Talk Between Endo-Sarcoplasmic Reticulum and Mitochondria in Skeletal Muscle. Antioxid Redox Signal. 2020 Apr 20;32(12):873-883. doi: 10.1089/ars.2019.7934. Epub 2019 Dec 11. PMID: 31825235.

40. McGrath TM, Spreckley E, Rodriguez AF, Viscomi C, Alamshah A, Akalestou E, Murphy KG, Jones NS. The homeostatic dynamics of feeding behaviour identify novel mechanisms of anorectic agents. PLoS Biol. 2019 Dec 5;17(12):e3000482. doi: 10.1371/journal.pbio.3000482. PMID: 31805040; PMCID: PMC6894749.

41. Viscomi C, Zeviani M. Breathe: Your Mitochondria Will Do the Rest… If They Are Healthy! Cell Metab. 2019 Oct 1;30(4):628-629. doi: 10.1016/j.cmet.2019.09.004. PMID: 31577932.

42. Segel M, Neumann B, Hill MFE, Weber IP, Viscomi C, Zhao C, Young A, Agley CC, Thompson AJ, Gonzalez GA, Sharma A, Holmqvist S, Rowitch DH, Franze K, Franklin RJM, Chalut KJ. Author

Correction: Niche stiffness underlies the ageing of central nervous system progenitor cells. Nature. 2019 Sep;573(7773):E3. doi: 10.1038/s41586-019-1552-1. Erratum for: Nature. 2019 Sep;573(7772):130-134. PMID: 31462780.

43. Segel M, Neumann B, Hill MFE, Weber IP, Viscomi C, Zhao C, Young A, Agley CC, Thompson AJ, Gonzalez GA, Sharma A, Holmqvist S, Rowitch DH, Franze K, Franklin RJM, Chalut KJ. Niche stiffness underlies the ageing of central nervous system progenitor cells. Nature. 2019 Sep;573(7772):130-134. doi: 10.1038/s41586-019-1484-9. Epub 2019 Aug 15. Erratum in: Nature. 2019 Aug 29;: PMID: 31413369; PMCID: PMC7025879.

44. Robb EL, Hall AR, Prime TA, Eaton S, Szibor M, Viscomi C, James AM, Murphy MP. Correction: Control of mitochondrial superoxide production by reverse electron transport at complex I. J Biol Chem. 2019 May 10;294(19):7966. doi: 10.1074/jbc.AAC119.008929. Erratum for: J Biol Chem. 2018 Jun 22;293(25):9869-9879. PMID: 31076523; PMCID: PMC6514609.

45. Signes A, Cerutti R, Dickson AS, Benincá C, Hinchy EC, Ghezzi D, Carrozzo R, Bertini E, Murphy MP, Nathan JA, Viscomi C, Fernandez-Vizarra E, Zeviani M. APOPT1/COA8 assists COX assembly and is oppositely regulated by UPS and ROS. EMBO Mol Med. 2019 Jan;11(1):e9582. doi: 10.15252/emmm.201809582. PMID: 30552096; PMCID: PMC6328941.

46. Civiletto G, Dogan SA, Cerutti R, Fagiolari G, Moggio M, Lamperti C, Benincá C, Viscomi C, Zeviani M. Rapamycin rescues mitochondrial myopathy via coordinated activation of autophagy and lysosomal biogenesis. EMBO Mol Med. 2018 Nov;10(11):e8799. doi: 10.15252/emmm.201708799. PMID: 30309855; PMCID: PMC6220341.

47. Garone C, Viscomi C. Towards a therapy for mitochondrial disease: an update. Biochem Soc Trans. 2018 Oct 19;46(5):1247-1261. doi: 10.1042/BST20180134. Epub 2018 Oct 8. PMID: 30301846; PMCID: PMC6195631.

48. Gammage PA, Viscomi C, Simard ML, Costa ASH, Gaude E, Powell CA, Van Haute L, McCann BJ, Rebelo-Guiomar P, Cerutti R, Zhang L, Rebar EJ, Zeviani M, Frezza C, Stewart JB, Minczuk M. Genome editing in mitochondria corrects a pathogenic mtDNA mutation in vivo. Nat Med. 2018 Nov;24(11):1691-1695. doi: 10.1038/s41591-018-0165-9. Epub 2018 Sep 24. PMID: 30250142; PMCID: PMC6225988.

49. Dogan SA, Cerutti R, Benincá C, Brea-Calvo G, Jacobs HT, Zeviani M, Szibor M, Viscomi C. Perturbed Redox Signaling Exacerbates a Mitochondrial Myopathy. Cell Metab. 2018 Nov 6;28(5):764-775.e5. doi: 10.1016/j.cmet.2018.07.012. Epub 2018 Aug 16. PMID: 30122554; PMCID: PMC6224544.

50. Agip AA, Blaza JN, Bridges HR, Viscomi C, Rawson S, Muench SP, Hirst J. Cryo-EM structures of complex I from mouse heart mitochondria in two biochemically defined states. Nat Struct Mol Biol. 2018 Jul;25(7):548-556. doi: 10.1038/s41594-018-0073-1. Epub 2018 Jun 18. PMID: 29915388; PMCID: PMC6054875.

51. Robb EL, Hall AR, Prime TA, Eaton S, Szibor M, Viscomi C, James AM, Murphy MP. Control of mitochondrial superoxide production by reverse electron transport at complex I. J Biol Chem. 2018 Jun 22;293(25):9869-9879. doi: 10.1074/jbc.RA118.003647. Epub 2018 May 9. Erratum in: J Biol Chem. 2019 May 10;294(19):7966. PMID: 29743240; PMCID: PMC6016480.

52. Deepa SS, Pharaoh G, Kinter M, Diaz V, Fok WC, Riddle K, Pulliam D, Hill S, Fischer KE, Soto V, Georgescu C, Wren JD, Viscomi C, Richardson A, Van Remmen H. Lifelong reduction in complex IV induces tissue-specific metabolic effects but does not reduce lifespan or healthspan in mice. Aging Cell. 2018 Aug;17(4):e12769. doi: 10.1111/acel.12769. Epub 2018 Apr 25. PMID: 29696791; PMCID: PMC6052393.

53. Torres-Torronteras J, Cabrera-Pérez R, Vila-Julià F, Viscomi C, Cámara Y, Hirano M, Zeviani M, Martí R. Long-Term Sustained Effect of Liver-Targeted Adeno-Associated Virus Gene Therapy for Mitochondrial Neurogastrointestinal Encephalomyopathy. Hum Gene Ther. 2018 Jun;29(6):708-718. doi: 10.1089/hum.2017.133. Epub 2018 Feb 26. PMID: 29284302; PMCID: PMC7647931.

54. Bottani E, Cerutti R, Harbour ME, Ravaglia S, Dogan SA, Giordano C, Fearnley IM, D'Amati G, Viscomi C, Fernandez-Vizarra E, Zeviani M. TTC19 Plays a Husbandry Role on UQCRFS1 Turnover in the Biogenesis of Mitochondrial Respiratory Complex III. Mol Cell. 2017 Jul 6;67(1):96-105.e4. doi: 10.1016/j.molcel.2017.06.001. Epub 2017 Jun 29. PMID: 28673544.

55. Viscomi C, Zeviani M. MtDNA-maintenance defects: syndromes and genes. J Inherit Metab Dis. 2017 Jul;40(4):587-599. doi: 10.1007/s10545-017-0027-5. Epub 2017 Mar 21. PMID: 28324239; PMCID: PMC5500664.

56. Profilo E, Peña-Altamira LE, Corricelli M, Castegna A, Danese A, Agrimi G, Petralla S, Giannuzzi G, Porcelli V, Sbano L, Viscomi C, Massenzio F, Palmieri EM, Giorgi C, Fiermonte G, Virgili M, Palmieri L, Zeviani M, Pinton P, Monti B, Palmieri F, Lasorsa FM. Down-regulation of the mitochondrial aspartate-glutamate carrier isoform 1 AGC1 inhibits proliferation and N-acetylaspartate synthesis in Neuro2A cells. Biochim Biophys Acta Mol Basis Dis. 2017 Jun;1863(6):1422-1435. doi: 10.1016/j.bbadis.2017.02.022. Epub 2017 Feb 21. PMID: 28235644.

57. Mansueto G, Armani A, Viscomi C, D'Orsi L, De Cegli R, Polishchuk EV, Lamperti C, Di Meo I, Romanello V, Marchet S, Saha PK, Zong H, Blaauw B, Solagna F, Tezze C, Grumati P, Bonaldo P, Pessin JE, Zeviani M, Sandri M, Ballabio A. Transcription Factor EB Controls Metabolic Flexibility during Exercise. Cell Metab. 2017 Jan 10;25(1):182-196. doi: 10.1016/j.cmet.2016.11.003. Epub 2016 Dec 20. PMID: 28011087; PMCID: PMC5241227.

58. Viscomi C. Toward a therapy for mitochondrial disease. Biochem Soc Trans. 2016 Oct 15;44(5):1483-1490. doi: 10.1042/BST20160085. PMID: 27911730; PMCID: PMC5095900.

59. Pitceathly RD, Viscomi C. Effects of ketosis in mitochondrial myopathy: potential benefits of a mitotoxic diet. EMBO Mol Med. 2016 Nov 2;8(11):1231-1233. doi: 10.15252/emmm.201606933. PMID: 27729389; PMCID: PMC5090656.

60. Lehtonen JM, Forsström S, Bottani E, Viscomi C, Baris OR, Isoniemi H, Höckerstedt K, Österlund P, Hurme M, Jylhävä J, Leppä S, Markkula R, Heliö T, Mombelli G, Uusimaa J, Laaksonen R, Laaksovirta H, Auranen M, Zeviani M, Smeitink J, Wiesner RJ, Nakada K, Isohanni P, Suomalainen A. FGF21 is a biomarker for mitochondrial translation and mtDNA maintenance disorders. Neurology. 2016 Nov 29;87(22):2290-2299. doi: 10.1212/WNL.0000000000003374. Epub 2016 Oct 28. PMID: 27794108; PMCID: PMC5270510.

61. Kovářová N, Pecina P, Nůsková H, Vrbacký M, Zeviani M, Mráček T, Viscomi C, Houštěk J. Data on cytochrome c oxidase assembly in mice and human fibroblasts or tissues induced by SURF1 defect. Data Brief. 2016 Mar 26;7:1004-9. doi: 10.1016/j.dib.2016.03.065. PMID: 27408912; PMCID: PMC4927972.

62. Sahebekhtiari N, Thomsen MM, Sloth JJ, Stenbroen V, Zeviani M, Gregersen N, Viscomi C, Palmfeldt J. Quantitative proteomics suggests metabolic reprogramming during ETHE1 deficiency. Proteomics. 2016 Apr;16(7):1166-76. doi: 10.1002/pmic.201500336. Epub 2016 Mar 16. PMID: 26867521.

63. Kovářová N, Pecina P, Nůsková H, Vrbacký M, Zeviani M, Mráček T, Viscomi C, Houštěk J. Tissue- and species-specific differences in cytochrome c oxidase assembly induced by SURF1 defects. Biochim Biophys Acta. 2016 Apr;1862(4):705-715. doi: 10.1016/j.bbadis.2016.01.007. Epub 2016 Jan 13. PMID: 26804654; PMCID: PMC4793088.

64. Brunetti D, Torsvik J, Dallabona C, Teixeira P, Sztromwasser P, Fernandez- Vizarra E, Cerutti R, Reyes A, Preziuso C, D'Amati G, Baruffini E, Goffrini P, Viscomi C, Ferrero I, Boman H, Telstad W, Johansson S, Glaser E, Knappskog PM, Zeviani M, Bindoff LA. Defective PITRM1 mitochondrial peptidase is associated with Aβ amyloidotic neurodegeneration. EMBO Mol Med. 2016 Mar 1;8(3):176-90. doi: 10.15252/emmm.201505894. PMID: 26697887; PMCID: PMC4772954.

65. Civiletto G, Varanita T, Cerutti R, Gorletta T, Barbaro S, Marchet S, Lamperti C, Viscomi C, Scorrano L, Zeviani M. Opa1 overexpression ameliorates the phenotype of two mitochondrial disease mouse models. Cell Metab. 2015 Jun 2;21(6):845-54. doi: 10.1016/j.cmet.2015.04.016. PMID: 26039449; PMCID: PMC4457891.

66. Varanita T, Soriano ME, Romanello V, Zaglia T, Quintana-Cabrera R, Semenzato M, Menabò R, Costa V, Civiletto G, Pesce P, Viscomi C, Zeviani M, Di Lisa F, Mongillo M, Sandri M, Scorrano L. The OPA1-dependent mitochondrial cristae remodeling pathway controls atrophic, apoptotic, and ischemic tissue damage. Cell Metab. 2015 Jun 2;21(6):834-44. doi: 10.1016/j.cmet.2015.05.007. PMID: 26039448; PMCID: PMC4457892.

67. Viscomi C, Bottani E, Zeviani M. Emerging concepts in the therapy of mitochondrial disease. Biochim Biophys Acta. 2015 Jun-Jul;1847(6-7):544-57. doi: 10.1016/j.bbabio.2015.03.001. Epub 2015 Mar 10. PMID: 25766847.

68. Pulliam DA, Deepa SS, Liu Y, Hill S, Lin AL, Bhattacharya A, Shi Y, Sloane L, Viscomi C, Zeviani M, Van Remmen H. Complex IV-deficient Surf1(-/-) mice initiate mitochondrial stress responses. Biochem J. 2014 Sep 1;462(2):359-71. doi: 10.1042/BJ20140291. PMID: 24911525; PMCID: PMC4145821.

69. Cerutti R, Pirinen E, Lamperti C, Marchet S, Sauve AA, Li W, Leoni V, Schon EA, Dantzer F, Auwerx J, Viscomi C, Zeviani M. NAD(+)-dependent activation of Sirt1 corrects the phenotype in a mouse model of mitochondrial disease. Cell Metab. 2014 Jun 3;19(6):1042-9. doi: 10.1016/j.cmet.2014.04.001. Epub 2014 May 8. PMID: 24814483; PMCID: PMC4051987.

70. Pirinen E, Cantó C, Jo YS, Morato L, Zhang H, Menzies KJ, Williams EG, Mouchiroud L, Moullan N, Hagberg C, Li W, Timmers S, Imhof R, Verbeek J, Pujol A, van Loon B, Viscomi C, Zeviani M, Schrauwen P, Sauve AA, Schoonjans K, Auwerx J. Pharmacological Inhibition of poly(ADP-ribose) polymerases improves fitness and mitochondrial function in skeletal muscle. Cell Metab. 2014 Jun 3;19(6):1034-41. doi: 10.1016/j.cmet.2014.04.002. Epub 2014 May 8. PMID: 24814482; PMCID: PMC4047186.

71. Torres-Torronteras J, Viscomi C, Cabrera-Pérez R, Cámara Y, Di Meo I, Barquinero J, Auricchio A, Pizzorno G, Hirano M, Zeviani M, Martí R. Gene therapy using a liver-targeted AAV vector restores nucleoside and nucleotide homeostasis in a murine model of MNGIE. Mol Ther. 2014 May;22(5):901-7. doi: 10.1038/mt.2014.6. Epub 2014 Jan 22. PMID: 24448160; PMCID: PMC4015233.

72. Bottani E, Giordano C, Civiletto G, Di Meo I, Auricchio A, Ciusani E, Marchet S, Lamperti C, d'Amati G, Viscomi C, Zeviani M. AAV-mediated liver- specific MPV17 expression restores mtDNA levels and prevents diet-induced liver failure. Mol Ther. 2014 Jan;22(1):10-7. doi: 10.1038/mt.2013.230. Epub 2013 Jul 3. PMID: 24247928; PMCID: PMC3880585.

73. Lin AL, Pulliam DA, Deepa SS, Halloran JJ, Hussong SA, Burbank RR, Bresnen A, Liu Y, Podlutskaya N, Soundararajan A, Muir E, Duong TQ, Bokov AF, Viscomi C, Zeviani M, Richardson AG, Van Remmen H, Fox PT, Galvan V. Decreased in vitro mitochondrial function is associated with enhanced brain metabolism, blood flow, and memory in Surf1-deficient mice. J Cereb Blood Flow Metab. 2013 Oct;33(10):1605-11. doi: 10.1038/jcbfm.2013.116. Epub 2013 Jul 10. PMID: 23838831; PMCID: PMC3790931.

74. Hildebrandt TM, Di Meo I, Zeviani M, Viscomi C, Braun HP. Proteome adaptations in Ethe1-deficient mice indicate a role in lipid catabolism and cytoskeleton organization via post-translational protein modifications. Biosci Rep. 2013 Jul 25;33(4):e00052. doi: 10.1042/BSR20130051. PMID: 23800285; PMCID: PMC3827611.

75. Deepa SS, Pulliam D, Hill S, Shi Y, Walsh ME, Salmon A, Sloane L, Zhang N, Zeviani M, Viscomi C, Musi N, Van Remmen H. Improved insulin sensitivity associated with reduced mitochondrial complex IV assembly and activity. FASEB J. 2013 Apr;27(4):1371-80. doi: 10.1096/fj.12-221879. Epub 2012 Dec 14. PMID: 23241310.

76. Di Meo I, Auricchio A, Lamperti C, Burlina A, Viscomi C, Zeviani M. Effective AAV-mediated gene therapy in a mouse model of ethylmalonic encephalopathy. EMBO Mol Med. 2012 Sep;4(9):1008-14. doi: 10.1002/emmm.201201433. Epub 2012 Aug 20. PMID: 22903887; PMCID: PMC3491831.

77. Giordano C, Viscomi C, Orlandi M, Papoff P, Spalice A, Burlina A, Di Meo I, Tiranti V, Leuzzi V, d'Amati G, Zeviani M. Morphologic evidence of diffuse vascular damage in human and in the experimental model of ethylmalonic encephalopathy. J Inherit Metab Dis. 2012 May;35(3):451-8. doi: 10.1007/s10545-011-9408-3. Epub 2011 Oct 22. PMID: 22020834.

78. **Viscomi C, Bottani E, Civiletto G, Cerutti R, Moggio M, Fagiolari G, Schon EA, Lamperti C, Zeviani M. In vivo correction of COX deficiency by activation of the AMPK/PGC-1α axis. Cell Metab. 2011 Jul 6;14(1):80-90. doi: 10.1016/j.cmet.2011.04.011. PMID: 21723506; PMCID: PMC3130927**.

79. Moreno-Loshuertos R, Ferrín G, Acín-Pérez R, Gallardo ME, Viscomi C, Pérez- Martos A, Zeviani M, Fernández-Silva P, Enríquez JA. Evolution meets disease: penetrance and functional epistasis of mitochondrial tRNA mutations. PLoS Genet. 2011 Apr;7(4):e1001379. doi: 10.1371/journal.pgen.1001379. Epub 2011 Apr 21. PMID: 21533077; PMCID: PMC3080857.

80. Baruscotti M, Bucchi A, Viscomi C, Mandelli G, Consalez G, Gnecchi-Rusconi T, Montano N, Casali KR, Micheloni S, Barbuti A, DiFrancesco D. Deep bradycardia and heart block caused by inducible cardiac-specific knockout of the pacemaker channel gene Hcn4. Proc Natl Acad Sci U S A. 2011 Jan 25;108(4):1705-10. doi: 10.1073/pnas.1010122108. Epub 2011 Jan 10. PMID: 21220308; PMCID: PMC3029742.

81. Stettner GM, Viscomi C, Zeviani M, Wilichowski E, Dutschmann M. Hypoxic and hypercapnic challenges unveil respiratory vulnerability of Surf1 knockout mice, an animal model of Leigh syndrome. Mitochondrion. 2011 May;11(3):413-20. doi: 10.1016/j.mito.2010.12.011. Epub 2010 Dec 16. PMID: 21167962.

82. Di Meo I, Fagiolari G, Prelle A, Viscomi C, Zeviani M, Tiranti V. Chronic exposure to sulfide causes accelerated degradation of cytochrome c oxidase in ethylmalonic encephalopathy. Antioxid Redox Signal. 2011 Jul 15;15(2):353-62. doi: 10.1089/ars.2010.3520. Epub 2011 Feb 25. PMID: 20812865.

83. Viscomi C, Burlina AB, Dweikat I, Savoiardo M, Lamperti C, Hildebrandt T, Tiranti V, Zeviani M. Combined treatment with oral metronidazole and N-acetylcysteine is effective in ethylmalonic encephalopathy. Nat Med. 2010 Aug;16(8):869-71. doi: 10.1038/nm.2188. Epub 2010 Jul 25. PMID: 20657580.

84. Tzoulis C, Neckelmann G, Mørk SJ, Engelsen BE, Viscomi C, Moen G, Ersland L, Zeviani M, Bindoff LA. Localized cerebral energy failure in DNA polymerase gamma-associated encephalopathy syndromes. Brain. 2010 May;133(Pt 5):1428-37. doi: 10.1093/brain/awq067. Epub 2010 Apr 16. PMID: 20400524.

85. Zecic A, Smet JE, De Praeter CM, Vanhaesebrouck P, Viscomi C, Van Den Broecke C, De Paepe B, Lohse P, Martin JJ, Jackson JG, Campbell CR, De Meirleir LJ, Zeviani M, Seneca SH, Lissens W, Van Coster RN. Lactic acidosis in a newborn with adrenal calcifications. Pediatr Res. 2009 Sep;66(3):317-22. doi: 10.1203/PDR.0b013e3181b40a80. PMID: 19581830; PMCID: PMC7101825.

86. Tiranti V, Viscomi C, Hildebrandt T, Di Meo I, Mineri R, Tiveron C, Levitt MD, Prelle A, Fagiolari G, Rimoldi M, Zeviani M. Loss of ETHE1, a mitochondrial dioxygenase, causes fatal sulfide toxicity in ethylmalonic encephalopathy. Nat Med. 2009 Feb;15(2):200-5. doi: 10.1038/nm.1907. Epub 2009 Jan 11. Erratum in: Nat Med. 2009 Feb;15(2):220. PMID: 19136963.

87. Ghezzi D, Viscomi C, Ferlini A, Gualandi F, Mereghetti P, DeGrandis D, Zeviani M. Paroxysmal non-kinesigenic dyskinesia is caused by mutations of the MR-1 mitochondrial targeting sequence. Hum Mol Genet. 2009 Mar 15;18(6):1058-64. doi: 10.1093/hmg/ddn441. Epub 2009 Jan 5. PMID: 19124534.

88. Viscomi C, Spinazzola A, Maggioni M, Fernandez-Vizarra E, Massa V, Pagano C, Vettor R, Mora M, Zeviani M. Early-onset liver mtDNA depletion and late-onset proteinuric nephropathy in Mpv17 knockout mice. Hum Mol Genet. 2009 Jan 1;18(1):12-26. doi: 10.1093/hmg/ddn309. Epub 2008 Sep 24. PMID: 18818194; PMCID: PMC2644642.

89. Cardaioli E, Da Pozzo P, Gallus GN, Malandrini A, Gambelli S, Gaudiano C, Malfatti E, Viscomi C, Zicari E, Berti G, Serni G, Dotti MT, Federico A. A novel heteroplasmic tRNA(Ser(UCN)) mtDNA point mutation associated with progressive external ophthalmoplegia and hearing loss. Neuromuscul Disord. 2007 Oct;17(9-10):681-3. doi: 10.1016/j.nmd.2007.05.001. Epub 2007 Jul 5. PMID: 17614276.

90. Spinazzola A, Viscomi C, Fernandez-Vizarra E, Carrara F, D'Adamo P, Calvo S, Marsano RM, Donnini C, Weiher H, Strisciuglio P, Parini R, Sarzi E, Chan A, DiMauro S, Rötig A, Gasparini P, Ferrero I, Mootha VK, Tiranti V, Zeviani M. MPV17 encodes an inner mitochondrial membrane protein and is mutated in
91. infantile hepatic mitochondrial DNA depletion. Nat Genet. 2006 May;38(5):570-5. doi: 10.1038/ng1765. Epub 2006 Apr 2. PMID: 16582910.

92. Gravante B, Barbuti A, Milanesi R, Zappi I, Viscomi C, DiFrancesco D. Interaction of the pacemaker channel HCN1 with filamin A. J Biol Chem. 2004 Oct 15;279(42):43847-53. doi: 10.1074/jbc.M401598200. Epub 2004 Jul 30. PMID:15292205.

93. Altomare C, Terragni B, Brioschi C, Milanesi R, Pagliuca C, Viscomi C, Moroni A, Baruscotti M, DiFrancesco D. Heteromeric HCN1-HCN4 channels: a comparison with native pacemaker channels from the rabbit sinoatrial node. J Physiol. 2003 Jun 1;549(Pt 2):347-59. doi: 10.1113/jphysiol.2002.027698. Epub 2003 Apr 17. PMID: 12702747; PMCID: PMC2342966.

94. Moroni A, Viscomi C, Sangiorgio V, Pagliuca C, Meckel T, Horvath F, Gazzarrini S, Valbuzzi P, Van Etten JL, DiFrancesco D, Thiel G. The short N-terminus is required for functional expression of the virus-encoded miniature K(+) channel Kcv. FEBS Lett. 2002 Oct 23;530(1-3):65-9. doi:10.1016/s0014-5793(02)03397-5. PMID: 12387867.

Book Chapters

Viscomi, C, Ardissone A, Zeviani M. Mitochondrial genes and neurodegenerative diseas, in Mitochondrial Dysfunction in Neurodegenerative Disorders, Editors: Reeve, A.K., Simcox, E., Duchen, M.R., Turnbull, D.M. (Eds.), Springer, 2016, ISBN 978-3-319-28637-2.

Viscomi C and Zeviani M. Mitochondria in health and disease, in Clinical Mitochondrial Medicine – P.F Chinnery and M Keogh (Eds), Cambridge University Press, ISBN 9781139192460.