



Curriculum Vitae Europass

Personal data

Name / Surname

Address

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Citizenship

Date of birth

Sex

Massimo Zeviani

Veneto Institute of Molecular Medicine,
Department of Neurosciences
University of Padova, Italy
massimo.zeviani@unipd.it

Italian

5th November 1955

Male

Professional sector

Health – Research

Professional Experience

2019 - present

Professor of Neurology, University of Padova, Italy

2013 - 2019

Professor of Mitochondrial Medicine, University of Cambridge, and Director of the MRC Mitochondrial Biology Unit, Cambridge, UK

2011 - 2013

Director of the Department of Molecular Medicine at the Istituto Neurologico "Carlo Besta", Milan (Italy)

2003 - 2013

Director of the Unit of Molecular Neurogenetics at the Istituto Neurologico "Carlo Besta"

1998 – 2002

Director of the Unit of Biochemistry and Genetics at the Istituto Neurologico "Carlo Besta".

1996 – 1997

Director of the Unit of Molecular Medicine at the Children's Hospital "Bambino Gesù" in Rome (Italy), and also Consultant at the Casa Sollievo della Sofferenza in San Giovanni Rotondo (Foggia-Italy) as Neurogeneticist

1993 – 1996

Associate of Neurology at the Istituto Neurologico "C. Besta", Milan (Italy)

1990 – 1993

Assistant of Neurology at the Department of Biochemistry and Genetics, and also Director of the Laboratory of Molecular Pathology of the Istituto Neurologico "C. Besta", directed by Prof. Stefano Di Donato.

Education

1997	PhD in Genetics "magna cum laude", registered 25 February 1997, University of Paris
1989	Specialization in Neurology "magna cum laude", registered 26 July 1989, University of Verona
1983	Specialization in Endocrinology "magna cum laude", registered 30 July 1983, University of Padova
1980	MD degree "magna cum laude", University of Padova
1974	High School Diploma ("Lyceum Classicum")

Professional Interests

- Muscle physiopathology
- Biochemistry and genetics of muscle metabolism
- Genetics of mitochondrial disorders and energy metabolism
- Genetics of inherited neurodegenerative disease

Teaching Experience

Held numerous seminars and conferences (>500) in National and International Congresses and in several University Institutions in Italy, Europe, Japan, Canada, Brazil, Argentina, Saudi Arabia, Israel, and the US. Opponent for PhD dissertations in Molecular Genetics at Helsinki University (1993, Anu Suomalainen), University of Paris (1994, Thomas Bourgeron), University of Glasgow (1994, Fiona Reid), Karolinska Institutet (2000, Jang-Minh), University of Tampere (2001, Olli Kajander); Karolinska Institutet (2005, Matt Ekstrand); University of Maastricht (2005; Bianca van der Bosch).

Under his supervision, numerous students graduated in Biology, Biotechnology and Medicine by carrying out experimental work for the final dissertation.

2007-2012	Course on Mitochondrial Medicine to undergraduate students of the Istituto Univesitario Studi Superiori, University of Pavia School of Medicine
2002	Course on Neurogenetics to undergraduate students of the University of Padua School of Medicine
1998	FEBS advanced course in Oxidative Phosphorylation
1996	European School of Medical Genetics, III Course. Lecture on mtDNA genetics and related human pathology
1993-1995	Course on Neurogenetics at the School of Specialization in Medical Genetics of the University of Milano
1990-1995	European School of Medical Genetics, III Course. Lecture on mtDNA genetics and related human pathology
1983	Institute of General Pathology, University of Padova. Taught histopathology to 3rd year medical students
1981-1982	Institute of Medical Semeiotics, University of Padova. Taught physical examination to 3rd year medical students

Honours

"The Annual George Komrower Honorary Lecture" SSIEM annual Meeting, Rome, September 2016

"The George Karpati Honorary Lecture" Institute of Neurology, McGill University, Montreal, CA, May 2016

Prix de la Fondation NRJ 2013" (Institut de France) « Génétique des maladies dégénératives » - June 2013

"Adam Barski Honorary Lecture in Mitochondrial Disease" The Sick kids Hospital, University of Toronto, ON, CA, April 2009

"Sir William Dunn" Scholar at The Dunn Human Nutrition Unit – MRC, Cambridge, UK, August-September 2008

"Gaetano Conte" Prize of the Mediterranean Society of Myology

"René Descartes" EU award for European transnational research, Prague, December 2004

"Brain" award for research on Neurogenetics, May 2000

"Late breaking news" Lecture at the Annual Meeting of the American Society of Human Genetics, Denver, USA, October 1998

"Anita Harding" Memorial Lecture, IX^o Meeting of the European Neurological Society, June 1998.

"Best Presentation" award at the 4^o Meeting of the European Neurological Society, June 1995.

INSERM "Post Vert" award for Visiting Researchers, 1994.

Associazione Italiana Ricerche sull' Handicap (AIRH) Award, 1991

Young Researcher Award at the 4^o National Congress of the Italian Federation for the Study of Inherited Disorders (FISME), Milano, September 1990.

"Valigia dell'Intelletto" Award by A.R.I.N. (Associazione Italiana per la Promozione della Ricerca neurologica) for Clinical Research in Neurology, January 1986

Young Researcher Award at the 1st International Congress on "Molecular genetics of neurological and neuromuscular diseases", Saint Vincent, September 1986

Selected publications (20 out of 421).

- Protasoni M, Pérez-Pérez R, Lobo-Jarne T, Harbour ME, Ding S, Peñas A, Diaz F, Moraes CT, Fearnley IM, Zeviani M, Ugalde C, Fernández-Vizarra E. [Respiratory supercomplexes act as a platform for complex III-mediated maturation of human mitochondrial complexes I and IV](#). EMBO J. 2020 020 Jan 8:e102817. doi: 10.15252/embj.2019102817.
- Costa R, Peruzzo R, Bachmann M, Montà GD, Vicario M, Santinon G, Mattarei A, Moro E, Quintana-Cabrera R, Scorrano L, Zeviani M, Vallese F, Zoratti M, Paradisi C, Argenton F, Brini M, Calì T, Dupont S, Szabò I, Leanza L. **Impaired Mitochondrial ATP Production Downregulates Wnt Signaling via ER Stress Induction**. Cell Rep. 2019 Aug 20;28(8):1949-1960.e6.
- Mohanraj K, Wasilewski M, Benincá C, Cysewski D, Poznanski J, Sakowska P, Bugajska Z, Deckers M, Dennerlein S, Fernandez-Vizarra E, Rehling P, Dadlez M, Zeviani M, Chacinska A. **Inhibition of proteasome rescues a pathogenic variant of respiratory chain assembly factor COA7**. EMBO Mol Med. 2019 May;11(5). pii: e9561.
- Posse V, Al-Behadili A, Uhler JP, Clausen AR, Reyes A, Zeviani M, Falkenberg M, Gustafsson CM. **RNase H1 directs origin-specific initiation of DNA replication in human mitochondria**. PLoS Genet. 2019 Jan 3;15(1):e1007781.

- Signes A, Cerutti R, Dickson AS, Benincá C, Hinchey 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). pii: e9582. doi: 10.15252/emmm.201809582.
- Civiletti 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). pii: e8799.
- Reyes A, Melchionda L, Burlina A, Robinson AJ, Ghezzi D, Zeviani M. **Mutations in TIMM50 compromise cell survival in OxPhos-dependent metabolic conditions.** EMBO Mol Med. 2018 Oct;10(10). pii: e8698.
- 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.
- Al-Behadili A, Uhler JP, Berglund AK, Peter B, Doimo M, Reyes A, Wanrooij S, Zeviani M, Falkenberg M. **A two-nuclease pathway involving RNase H1 is required for primer removal a human mitochondrial OriL.** Nucleic Acids Res. 2018 Oct 12;46(18):9471-9483.
- 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.
- 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 UQCRC1 Turnover in the Biogenesis of Mitochondrial Respiratory Complex III.** Mol Cell. 2017 Jul 6;67(1):96-105.
- Vidoni S, Harbour ME, Guerrero-Castillo S, Signes A, Ding S, Fearnley IM, Taylor RW, Tiranti V, Arnold S, Fernandez-Vizarra E, Zeviani M. **MR-1S Interacts with PET100 and PET117 in Module-Based Assembly of Human Cytochrome c Oxidase.** Cell Rep. 2017 Feb 14;18(7):1727-1738.
- 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.
- 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.
- Reyes A, Melchionda L, Nasca A, Carrara F, Lamantea E, Zanolini A, Lamperti C, Fang M, Zhang J, Ronchi D, Bonato S, Fagiolari G, Moggio M, Ghezzi D, Zeviani M. **RNASEH1 Mutations Impair mtDNA Replication and Cause Adult-Onset Mitochondrial Encephalomyopathy.** Am J Hum Genet. 2015 Jul 2;97(1):186-93.
- Civiletti 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 Metabolism. 2015 Jun; 21(6): 845-854.
- Steenweg ME, Ghezzi D, Haack T, Abbink TEM, Martinelli D, van Berkel CGM, Bley A, Diogo L, Grillo E, Te Water Naude J, Strom TM, Bertini E, Prokisch H, van der Knaap MS, Zeviani M. **Leukoencephalopathy with thalamus and brainstem involvement and high lactate "LTBL" caused by EARS2 mutations.** Brain. 2012 Apr; 135(5): 1387-1394.

- Ghezzi D, Arzuffi P, Zordan M, Da Re C, Lamperti C, Benna C, D'Adamo P, Diodato D, Costa R, Mariotti C, Uziel G, Smiderle C, Zeviani M. **Mutations in TTC19 cause mitochondrial complex III deficiency and neurological impairment in humans and flies.** Nature Genetics. 2011 Jan; 43(3): 259–263.
- Ghezzi D, Goffrini P, Uziel G, Horvath R, Klopstock T, Lochmüller H, D'Adamo P, Gasparini P, Strom TM, Prokisch H, Invernizzi F, Ferrero I, Zeviani M. **SDHAF1, encoding a LYR complex-II specific assembly factor, is mutated in SDH-defective infantile leukoencephalopathy.** Nature Genetics. 2009 May; 41(6): 654–656.
- Tiranti V, Viscomi C, Hildebrandt T, Di Meo I, Mineri R, Tiveron C, D Levitt M, Prella A, Fagioli G, Rimoldi M, Zeviani M. **Loss of ETHE1, a mitochondrial dioxygenase, causes fatal sulfide toxicity in ethylmalonic encephalopathy.** Nature Medicine. 2009 Jan; 15(2): 200–205.

Publications 2015-2020

75 results

1. PMID: 32632204

Loss of function of the mitochondrial peptidase PITRM1 induces proteotoxic stress and Alzheimer's disease-like pathology in human cerebral organoids.

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. *Mol Psychiatry*. 2020 Jul 7. doi: 10.1038/s41380-020-0807-4. Online ahead of print. PMID: 32632204

2. PMID: 32562616

Opa1 Overexpression Protects from Early-Onset Mpv17^{-/-}-Related Mouse Kidney Disease.

Luna-Sanchez M, Benincá C, Cerutti R, Brea-Calvo G, Yeates A, Scorrano L, Zeviani M, Viscomi C. *Mol Ther*. 2020 Jun 12;S1525-0016(20)30300-2. doi: 10.1016/j.jymthe.2020.06.010. Online ahead of print. PMID: 32562616

3. PMID: 32478122

A Single Intravenous Injection of AAV-PHP.B-*hNDUFS4* Ameliorates the Phenotype of *Ndufs4*^{-/-} Mice.

Silva-Pinheiro P, Cerutti R, Luna-Sanchez M, Zeviani M, Viscomi C. *Mol Ther Methods Clin Dev*. 2020 May 4;17:1071-1078. doi: 10.1016/j.omtm.2020.04.026. eCollection 2020 Jun 12. PMID: 32478122 **Free PMC article.**

4. PMID: 32439808

Mutation in the MICOS subunit gene *APOO* (MIC26) associated with an X-linked recessive mitochondrial myopathy, lactic acidosis, cognitive impairment and autistic features.

Benincá C, Zanette V, Brischigliaro M, Johnson M, Reyes A, Valle DAD, J Robinson A, Degiorgi A, Yeates A, Telles BA, Prudent J, Baruffini E, S F Santos ML, R de Souza RL, Fernandez-Vizarra E, Whitworth AJ, Zeviani M. *J Med Genet*. 2020 May 21:jmedgenet-2020-106861. doi: 10.1136/jmedgenet-2020-106861. Online ahead of print. PMID: 32439808

5. PMID: 32344152

A homozygous MRPL24 mutation causes a complex movement disorder and affects the mitoribosome assembly.

Di Nottia M, Marchese M, Verrigni D, Mutti CD, Torraco A, Oliva R, Fernandez-Vizarra E, Morani F, Trani G, Rizza T, Ghezzi D, Ardisson A, Nesti C, Vasco G, Zeviani M, Minczuk M, Bertini E, Santorelli FM, Carrozzo R. *Neurobiol Dis.* 2020 Jul;141:104880. doi: 10.1016/j.nbd.2020.104880. Epub 2020 Apr 25. PMID: 32344152

6. PMID: 32219868

ATPase Domain AFG3L2 Mutations Alter OPA1 Processing and Cause Optic Neuropathy.

Caporali L, Magri S, Legati A, Del Dotto V, Tagliavini F, Balistreri F, Nasca A, La Morgia C, Carbonelli M, Valentino ML, Lamantea E, Baratta S, Schöls L, Schüle R, Barboni P, Cascavilla ML, Maresca A, Capristo M, Ardisson A, Pareyson D, Cammarata G, Melzi L, Zeviani M, Peverelli L, Lamperti C, Marzoli SB, Fang M, Synofzik M, Ghezzi D, Carelli V, Taroni F. *Ann Neurol.* 2020 Jul;88(1):18-32. doi: 10.1002/ana.25723. Epub 2020 Apr 21. PMID: 32219868

7. PMID: 32100338

Strategies for fighting mitochondrial diseases.

Viscomi C, Zeviani M. *J Intern Med.* 2020 Jun;287(6):665-684. doi: 10.1111/joim.13046. Epub 2020 Apr 13. PMID: 32100338

8. PMID: 32082360

RNase H1 Regulates Mitochondrial Transcription and Translation *via* the Degradation of 7S RNA.

Reyes A, Rusecka J, Tońska K, Zeviani M. *Front Genet.* 2020 Jan 31;10:1393. doi: 10.3389/fgene.2019.01393. eCollection 2019. PMID: 32082360 **Free PMC article.**

9. PMID: 32042910

Expanding the molecular and phenotypic spectrum of truncating *MT-ATP6* mutations.

Bugiardini E, Bottani E, Marchet S, Poole OV, Beninca C, Horga A, Woodward C, Lam A, Hargreaves I, Chalasani A, Valerio A, Lamantea E, Venner K, Holton JL, Zeviani M, Houlden H, Quinlivan R, Lamperti C, Hanna MG, Pitceathly RDS. *Neurol Genet.* 2020 Jan 7;6(1):e381. doi: 10.1212/NXG.0000000000000381. eCollection 2020 Feb. PMID: 32042910 **Free PMC article.**

10. PMID: 31989346

Awareness of rare and genetic neurological diseases among italian neurologist. A national survey.

Mancuso M, Filosto M, Lamperti C, Musumeci O, Santorelli FM, Servidei S, Valente EM, Zeviani M, Mancardi G, Tedeschi G, Federico A. *Neurol Sci.* 2020 Jun;41(6):1567-1570. doi: 10.1007/s10072-020-04271-2. Epub 2020 Jan 27. PMID: 31989346

11. PMID: 31912925

Respiratory supercomplexes act as a platform for complex III-mediated maturation of human mitochondrial complexes I and IV.

Protasoni M, Pérez-Pérez R, Lobo-Jarne T, Harbour ME, Ding S, Peñas A, Diaz F, Moraes CT, Fearnley IM, Zeviani M, Ugalde C, Fernández-Vizarra E. *EMBO J.* 2020 Feb 3;39(3):e102817. doi: 10.15252/embj.2019102817. Epub 2020 Jan 8. PMID: 31912925 **Free PMC article.**

12. PMID: 31787496

Novel compound heterozygous pathogenic variants in nucleotide-binding protein like protein (NUBPL) cause leukoencephalopathy with multi-systemic involvement.

Protasoni M, Bruno C, Donati MA, Mohamoud K, Severino M, Allegri A, Robinson AJ, Reyes A, Zeviani M, Garone C. *Mol Genet Metab.* 2020 Jan;129(1):26-34. doi: 10.1016/j.ymgme.2019.11.003. Epub 2019 Nov 21. PMID: 31787496

13. PMID: 31577932

Breathe: Your Mitochondria Will Do the Rest... If They Are Healthy!

Viscomi C, Zeviani M. *Cell Metab.* 2019 Oct 1;30(4):628-629. doi: 10.1016/j.cmet.2019.09.004. PMID: 31577932

14. PMID: 31555154

Knockdown of *APOPT1/COA8* Causes Cytochrome c Oxidase Deficiency, Neuromuscular Impairment, and Reduced Resistance to Oxidative Stress in *Drosophila melanogaster*.

Brischigliaro M, Corrà S, Tregnago C, Fernandez-Vizarra E, Zeviani M, Costa R, De Pittà C. *Front Physiol.* 2019 Sep 6;10:1143. doi: 10.3389/fphys.2019.01143. eCollection 2019. PMID: 31555154 **Free PMC article.**

15. PMID: 31433973

Impaired Mitochondrial ATP Production Downregulates Wnt Signaling via ER Stress Induction.

Costa R, Peruzzo R, Bachmann M, Montà GD, Vicario M, Santinon G, Mattarei A, Moro E, Quintana-Cabrera R, Scorrano L, Zeviani M, Vallese F, Zoratti M, Paradisi C, Argenton F, Brini M, Calì T, Dupont S, Szabò I, Leanza L. *Cell Rep.* 2019 Aug 20;28(8):1949-1960.e6. doi: 10.1016/j.celrep.2019.07.050. PMID: 31433973

16. PMID: 30979712

miR-181a/b downregulation exerts a protective action on mitochondrial disease models.

Indrieri A, Carrella S, Romano A, Spaziano A, Marrocco E, Fernandez-Vizarra E, Barbato S, Pizzo M, Ezhova Y, Golia FM, Ciampi L, Tammara R, Henao-Mejia J, Williams A, Flavell RA, De Leonibus E, Zeviani M, Surace EM, Banfi S, Franco B. *EMBO Mol Med.* 2019 May;11(5):e8734. doi: 10.15252/emmm.201708734. PMID: 30979712 **Free PMC article.**

17. PMID: 30885959

Inhibition of proteasome rescues a pathogenic variant of respiratory chain assembly factor COA7.

Mohanraj K, Wasilewski M, Benincà C, Cysewski D, Poznanski J, Sakowska P, Bugajska Z, Deckers M, Dennerlein S, Fernandez-Vizarra E, Rehling P, Dadlez M, Zeviani M, Chacinska A. *EMBO Mol Med.* 2019 May;11(5):e9561. doi: 10.15252/emmm.201809561. PMID: 30885959 **Free PMC article.**

18. PMID: 30873109

Lipomatosis Incidence and Characteristics in an Italian Cohort of Mitochondrial Patients.

Musumeci O, Barca E, Lamperti C, Servidei S, Comi GP, Moggio M, Mongini T, Siciliano G, Filosto M, Pegoraro E, Primiano G, Ronchi D, Vercelli L, Orsucci D, Bello L, Zeviani M, Mancuso M, Toscano A. *Front Neurol.* 2019 Feb 27;10:160. doi: 10.3389/fneur.2019.00160. eCollection 2019. PMID: 30873109 **Free PMC article.**

19. PMID: 30605451

[RNase H1 directs origin-specific initiation of DNA replication in human mitochondria.](#)

Posse V, Al-Behadili A, Uhler JP, Clausen AR, Reyes A, Zeviani M, Falkenberg M, Gustafsson CM. *PLoS Genet.* 2019 Jan 3;15(1):e1007781. doi:

10.1371/journal.pgen.1007781. eCollection 2019 Jan. PMID: 30605451 **Free PMC article.**

20. PMID: 30552096

[APOPT1/COA8 assists COX assembly and is oppositely regulated by UPS and ROS.](#)

Signes A, Cerutti R, Dickson AS, Benincá C, Hinchey EC, Ghezzi D, Carrozzo R, Bertini E, Murphy MP, Nathan JA, Viscomi C, Fernandez-Vizarra E, Zeviani M. *EMBO Mol Med.* 2019 Jan;11(1):e9582. doi: 10.15252/emmm.201809582. PMID: 30552096 **Free PMC article.**

21. PMID: 30309855

[Rapamycin rescues mitochondrial myopathy via coordinated activation of autophagy and lysosomal biogenesis.](#)

Civiletto G, Dogan SA, Cerutti R, Fagiolaro G, Moggio M, Lamperti C, Benincá C, Viscomi C, Zeviani M. *EMBO Mol Med.* 2018 Nov;10(11):e8799. doi:

10.15252/emmm.201708799. PMID: 30309855 **Free PMC article.**

22. PMID: 30250142

[Genome editing in mitochondria corrects a pathogenic mtDNA mutation in vivo.](#)

Gammage PA, Viscomi C, Simard ML, Costa ASH, Gaude E, Powell CA, Van Haute L, McCann BJ, Rebelo-Guimaraes P, Cerutti R, Zhang L, Rebar EJ, Zeviani M, Frezza C, Stewart JB, Minczuk M. *Nat Med.* 2018 Nov;24(11):1691-1695. doi: 10.1038/s41591-018-0165-9. Epub 2018 Sep 24. PMID: 30250142 **Free PMC article.**

23. PMID: 30190335

[Mutations in *TIMM50* compromise cell survival in OxPhos-dependent metabolic conditions.](#)

Reyes A, Melchionda L, Burlina A, Robinson AJ, Ghezzi D, Zeviani M. *EMBO Mol Med.*

2018 Oct;10(10):e8698. doi: 10.15252/emmm.201708698. PMID: 30190335 **Free PMC article.**

24. PMID: 30122554

[Perturbed Redox Signaling Exacerbates a Mitochondrial Myopathy.](#)

Dogan SA, Cerutti R, Benincá C, Brea-Calvo G, Jacobs HT, Zeviani M, Szibor M, Viscomi C. *Cell Metab.* 2018 Nov 6;28(5):764-775.e5. doi: 10.1016/j.cmet.2018.07.012. Epub

2018 Aug 16. PMID: 30122554 **Free PMC article.**

25. PMID: 30102370

[A two-nuclease pathway involving RNase H1 is required for primer removal at human mitochondrial OriL.](#)

Al-Behadili A, Uhler JP, Berglund AK, Peter B, Doimo M, Reyes A, Wanrooij S, Zeviani M, Falkenberg M. *Nucleic Acids Res.* 2018 Oct 12;46(18):9471-9483. doi:

10.1093/nar/gky708. PMID: 30102370 **Free PMC article.**

26. PMID: 30030362

[Human diseases associated with defects in assembly of OXPHOS complexes.](#)

Ghezzi D, Zeviani M. *Essays Biochem.* 2018 Jul 20;62(3):271-286. doi: 10.1042/EBC20170099. Print 2018 Jul 20. PMID: 30030362 **Free PMC article.** Review.

27. PMID: 30025539

Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?

Repp BM, Mastantuono E, Alston CL, Schiff M, Haack TB, Rötig A, Ardisson A, Lombès A, Catarino CB, Diodato D, Schottmann G, Poulton J, Burlina A, Jonckheere A, Munnich A, Rolinski B, Ghezzi D, Rokicki D, Wellesley D, Martinelli D, Wenhong D, Lamantea E, Ostergaard E, Pronicka E, Pierre G, Smeets HJM, Wittig I, Scurr I, de Coo IFM, Moroni I, Smet J, Mayr JA, Dai L, de Meirleir L, Schuelke M, Zeviani M, Morscher RJ, McFarland R, Seneca S, Klopstock T, Meitinger T, Wieland T, Strom TM, Herberg U, Ahting U, Sperl W, Nassogne MC, Ling H, Fang F, Freisinger P, Van Coster R, Strecker V, Taylor RW, Häberle J, Vockley J, Prokisch H, Wortmann S. *Orphanet J Rare Dis.* 2018 Jul 19;13(1):120. doi: 10.1186/s13023-018-0784-8. PMID: 30025539 **Free PMC article.**

28. PMID: 29764912

Mitochondrial *PITRM1* peptidase loss-of-function in childhood cerebellar atrophy.

Langer Y, Aran A, Gulsuner S, Abu Libdeh B, Renbaum P, Brunetti D, Teixeira PF, Walsh T, Zeligson S, Ruotolo R, Beerli R, Dweikat I, Shahrour M, Weinberg-Shukron A, Zahdeh F, Baruffini E, Glaser E, King MC, Levy-Lahad E, Zeviani M, Segel R. *J Med Genet.* 2018 Sep;55(9):599-606. doi: 10.1136/jmedgenet-2018-105330. Epub 2018 May 15. PMID: 29764912

29. PMID: 29601977

SURF1 knockout cloned pigs: Early onset of a severe lethal phenotype.

Quadalti C, Brunetti D, Lagutina I, Duchi R, Perota A, Lazzari G, Cerutti R, Di Meo I, Johnson M, Bottani E, Crociara P, Corona C, Grifoni S, Tiranti V, Fernandez-Vizarra E, Robinson AJ, Viscomi C, Casalone C, Zeviani M, Galli C. *Biochim Biophys Acta Mol Basis Dis.* 2018 Jun;1864(6 Pt A):2131-2142. doi: 10.1016/j.bbadis.2018.03.021. Epub 2018 Mar 28. PMID: 29601977 **Free PMC article.**

30. PMID: 29577824

Cavitating Leukoencephalopathy With Posterior Predominance Caused by a Deletion in the *APOPT1* Gene in an Indian Boy.

Sharma S, Singh P, Fernandez-Vizarra E, Zeviani M, Van der Knaap MS, Saran RK. *J Child Neurol.* 2018 May;33(6):428-431. doi: 10.1177/0883073818760875. Epub 2018 Mar 26. PMID: 29577824

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Compound heterozygous missense and deep intronic variants in *NDUFAF6* unraveled by exome sequencing and mRNA analysis.

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Selected Grants.

Title	Funding Agency	Duration	Contract No.	Total Amount (k€)
Identification and Characterization of Nuclear Genes Responsible for Human Mitochondrial Disorders (Coordinator)	Telethon-IT	2012-14	GPP11011	389
Definition and characterization of disease genes in mitochondrial disorders (Coordinator)	Fondazione CARIPLO	2012-13	2011-0526	300
Mitochondrial Disorders – Connecting biobanks, empowering diagnostics and exploring disease models (Partner)	E-Rare	2012-14	JTC2011	128
Combating mitochondrial disorders (Coordinator)	Telethon-IT	2011-13	GPP 10005	1.235
Establishing a Center for Mitochondrial Disorders of Infancy and Childhood (Individual grant)	Fondazione Mariani	2009-13	Ricerca2000	750

MitCare: Mitochondrial Medicine: developing treatments of OxPhos-defects in recombinant mammalian models.	ERC	2013-2018	FP7-322424	2.500
MitoFight: Experimental strategies to combat Pearson's syndrome	Fondazione Renato Comini Onlus	2017-2022		500
Mito-ND, Mitochondrial Neurodegeneration	Centres of Excellence in Neurodegeneration (COEN)	2016-2018		350
GGP19007: Experimental gene therapy in mitochondrial disorders	Telethon-IT	2019-22		300