PERSONAL INFORMATION

Caterina Garone

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Sex Female | *Date of birth* 27/09/1979 | *Nationality* Italian

Enterprise	University	EPR
☐ Management Level	☐ Full professor	☐ Research Director and 1st level Technologist / First Researcher and 2nd level Technologist / Principal Investigator
☐ Mid-Management Level	XAssociate Professor	☐ Level III Researcher and Technologist
☐ Employee / worker level	☐ Researcher and Technologist of IV, V, VI and VII level / Technical collaborator***	☐ Researcher and Technologist of IV, V, VI and VII level / Technical collaborator

WORK EXPERIENCE

2022-present	**Associate Professor Medical Genetics** Department of Medical and Surgical Sciences, University of Bologna, Italy *Group leader of mitochondrial translational medicine laboratory* *Teaching activity and laboratory training on medical genetics field*
2019-2022	**Senior Assistant Professor Medical Genetics** Department of Medical and Surgical Sciences, University of Bologna, Italy *Group leader of mitochondrial translational medicine laboratory Teaching activity and laboratory training on medical genetics field*
2020-present	**Consultant Child Neurologist** IRCCS Neurological Sciences, Bologna, Italy *Specialized clinics for rare neurogenetics diseases*
2014-2019	**Postdoctoral Scientist** MRC Mitochondrial Biology Unit, University of Cambridge - UK *Discovering disease pathways and novel treatment approaches for mtDNA metabolism disorders*
2015-2019	**Honorary Consultant Pediatric Neurology** Cambridge University Hospital - UK *Dedicated clinics for inherited neuromuscular and neurometabolic disorders*
2010-2014	**Research scientist at Columbia University (US) during Italian PhD program** Columbia University Medical Center, New York (US) *Discovering novel disease causing genes and developing treatment approaches for mitochondrial disorders*

EDUCATION AND TRAINING

2020	**Alumni** MRC Mitochondrial Biology Unit, University of Cambridge http://www.mrc-mbu.cam.ac.uk/alumni	*NA*
2010-2014	**PhD**: **Human Genetics**, University of Turin *The research program was developed abroad in the laboratories of Prof.Billi DiMauro and Michio Hirano, at Columbia University Medical Center, New York (US)	*8*
2005-2010	**Child Neurology and Psychiatry**, University of Bologna - Bologna	*8*
1999-2005	**Medical Doctor Degree**, University of Bologna - Bologna	*7*

WORK ACTIVITIES

Awards	2019 - **Rita Levi Montalcini Award**- Rientro Cervelli – Italian Minister of University and Research (MED03/BIO12) *"Tissue-specificity of mtDNA metabolism disorders"* 2016 - **Marie Skłodowska-Curie Actions** – **European Commission**- Proposal 705560 – MITOBIOPATH *"Discovering new disease pathways affecting mtDNA metabolism"*

	2013 - **Young Investigator at Neurolobiology and Disease in Children** symposium: mitochondrial disease, US
Editorial activity	- **Guest Editor** for "Essay in Biochemistry: Mitochondrial diseases" - **Guest Editor** for Frontiers in Genetics Special Issue "Mitochondrial Genetics and Epigenetics" - **Referee** for EMBO Molecular Medicine, Journal of Pediatric Neurology, Brain, Gene, Gene Review, Archive of Diseases in childhood, Clinical Neurology and Neuroscience, Frontiers in Genetics, Annals of Human Genetics
Invited presentations	**Mitocon** (2018, 2019) **European School of Human Genetics** (2017) **MEET** (Mitochondrial European Training) symposium (2016) **ENMC** Workshop (2017) **British Pediatric Neurology Association** (2017) **Italian Society of Metabolic disease** (2015)
Grants	**2021- Bando Ricerca Traslazionale Carisbo** - "*Studio di cellule staminali di pazienti con disordine del metabolismo del DNA mitocondriale" (*PI role) **2020- Bando Alta Tecnologia Carisbo** - *"Sviluppo di tecnologie all'avanguardia per la sperimentazione traslazionale di approcci terapeutici alle malattie mitocondriali" (*PI role) **2019- Charlie Gard Foundation Grant Award**- *"Development of treatment strategies for mitochondrial dNTP unbalance-related disorders" (*PI role) **2019- Lily Foundation Grant Award** - Universities of Cambridge, Cardiff, Newcastle - *"Towards devising a therapeutic strategy for patients with recessive RRM2B-related mitochondrial disease"(*PI role)
Patents	- **United States Patent No**. **10,292,996**: "Deoxyribonucleoside monophosphate bypass therapy for mitochondrial DNA depletion syndromes" - **United States Patent No. 10,471,087**: "Deoxynucleosides therapy for diseases caused by unbalanced nucleotide pools including mitochondrial DNA depletion syndromes"

ADDITIONAL INFORMATION

Publications	total number of publications in peer-review journals = **64** average IF/paper: **10.04** total number of citations = **2074** H index = **25** 1. Rebelo-Guiomar P…**Garone C**…Minczuk M. A late-stage assembly checkpoint of the human mitochondrial ribosome large subunit. *Nat. Commun.* Epub ahead of print 2. Van Haute L…***Garone C***…Minczuk M. NSUN2 introduces 5-methylcytosines in mammalian mitochondrial tRNAs. *Nucleic Acids Res*. 2019 3. **Garone C**, Viscomi C. Towards a therapy for mitochondrial disease: an update. *Biochem Soc Trans*. 2018 4. Lopez-Gomez C…**Garone C*,** Hirano M*. Deoxycytidine and deoxythymidine treatment for thymidine kinase 2 deficiency. Annals of Neurology, 2017- *Ann Neurol.* 5. **Garone C*** et al. Defective mitochondrial rRNA methyltransferase MRM2 causes MELAS-like clinical syndrome. *Hum Mol Genet.* 2017. 6. Harel T, Yoon WH, **Garone C** et al. Recurrent De Novo and Biallelic Variation of ATAD3A, Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. *Am J Hum Genet*. 2016 7. **Garone C** et al. Deoxypyrimidine monophosphate bypass therapy for thymidine kinase 2 deficiency. *EMBO Mol Med*. 2014. 8. Kariya S, Obis T, **Garone C** et al. Requirement of enhanced Survival Motoneuron protein imposed during neuromuscular junction maturation. *J Clin Invest*. 2014. 9. Quinzii CM*, **Garone C*** et al. M.Tissue- specific oxidative stress and loss of mitochondria in CoQ-deficient Pdss2 mutant mice. *FASEB J.* 2013 10. Calvo SE, Compton AG, Hershman SG, Lim SC, Lieber DS, Tucker EJ, Laskowski A, **Garone C**, Shangtao Liu S, Jaffe DB, Christodoulou J, Fletcher JM, Bruno DL, Goldblatt J, DiMauro S, Thorburn DR, Mootha V. Molecular Diagnosis of Infantile Mitochondrial Disease with Targeted Next-Generation Sequencing. *Science Translational Medicine*, 2012; 4(118): 118ra10.