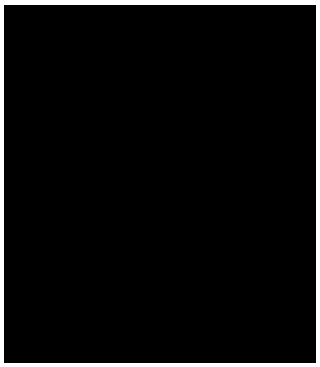
PERSONAL INFORMATION

Caterina Garone



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WORK EXPERIENCE

2022-present	**Associate Professor Medical Genetics** Department of Medical and Surgical Sciences, University of Bologna, 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, Bologna (Italy) *Group leader of mitochondrial translational medicine laboratory* *Teaching activity and laboratory training on medical genetics field*
2020-present	**Consultant Neurogeneticist** IRCCS Neurological Sciences, Bologna, Italy *Specialized clinics for rare neurogenetics diseases*
2022-present	**Scientific Advisor for Glycomine** - US
2019-2022	**Scientific Advisor for Modis Therapeutics** – (US)
2014-2019	**Postdoctoral Scientist** MRC Mitochondrial Biology Unit, University of Cambridge**,** 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

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)
2005-2010	**Child Neurology and Psychiatry**, University of Bologna – Bologna
1999-2005	**Medical Doctor Degree**, University of Bologna – Bologna

WORK ACTIVITIES

Teaching activities	**2022-present Medical Genetics Course,** Facoltà di Medicina e Chirurgia
	2020- present: Medical Genetics Course, Scuola di Specializzazione Medica in Genetica Medica – UNIBO
	2020- present - Medical Genetics Course, Scuola di Specializzazione Non Medica in Genetica Medica – UNIBO – continuativamente dal 2020
	2021-2022 - Master Metabolic Diseases – Lesson on "Mitochondrial Medicine" and "Gene therapy and mRNA drugs"
	2019- present: Supervisor "Tesi in Genetica Medica per Laura Magistrale in Medicina e Chirurgia" n.2 students
	2019- present: Supervisor "Tesi per Laura Magistrale in Biologia Molecolare" n.3 students
	2019- present: Supervisor "Tesi per Laura Magistrale in Biotecnologie mediche" n.2 students
	2019-present: Supervisor "Tesi in Neuropsichiatria Infantile per Laura Magistrale in Medicina" n.2 students
	Joint Seminars with John Hopkins University on Medical Genetics (2022)
	Seminar on Mitochondrial Medicine Scuola di Specializzazione in Neuropsichiatria Infantile – UNIBO 2020
	European School of Human Genetics (2017) – Lesson on Mitochondrial Medicine
Istitutional Board Committees	Head of Research (Delegato Ricerca) for the Department of Medical and Surgical Sciences
	Membro del Consiglio di Sorveglia per il Centro Nazionale Terapia Genica e Farmaci mRNA (CN3)
	Gruppo di Lavoro PNRR per Centro Nazionale Terapia Genica e Farmaci mRNA (CN3)
	Gruppo di Lavoro UNIBO per la realizzazione del progetto "Torre Biomedica"
Member of Scientific Committee	2016- present Italian Society of Human Genetics (Società Italiana di Genetica Umana) (SIGU)
Commission of Trust	2016 - present Referee for Italian Minister of Health (grant application - PRIN), Insubria university (Italy), Rita Levi Montalcini program (Italy), Biotechnology and Biological Sciences Research Council Fellowship Scheme (UK)
Awards	2019 - **Rita Levi Montalcini Award**- Rientro Cervelli – Italian Minister of University and Research (MED03/BIO12) *"Tissue-specificity of mtDNA metabolism disorders"*

	2015 - **in-year Special Award Scheme (SAS)** - Medical Research Council Mitochondrial Biology Unit, Cambridge
	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
	2013 - **Young Investigator at NDC meeting**. Selected as young investigator at Neurolobiology and Disease in Children symposium: mitochondrial disease.
	2013 - **Travel Grant at 18th International World Muscle Society Congress**, Asilomar (US)
	2010-2011 - **Fellowship AMMeC** (Associazione Malattie Metaboliche e congenite ereditarie) Project: "Treatment with dNTP pool in Tk2 knock-in mouse model" developed at Columbia University (New York)
Editorial activity	**Associated Editor for** "Cellular Metabolism Therapy" section in Journal of Translational Medicine
	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** (2019, 2018, 2017)
	MEET (Mitochondrial European Training) symposium (2016)
	ENMC Workshop Recommendations for treatment of Mitochondrial DNA maintenance disorders" (Heemskerk, The Netherland) (2017)
	British Pediatric Neurology Association - Cambridge, UK (2017)
	Italian Society of Metabolic disease, National Meeting – Firenze, Italy (2015)
	Italian Society of Pediatric Neurology, National Meeting – Bologna, Italy (2015)
	Ground Round, Department of Neurology Columbia University- New York, US (2012)
Grants	*2022* ***PNRR****- National Center for gene therapy and mRNA drugs- Spoke Genetics (PI role)*
	2022 ***PNRR****- PE12 (Collaborator Role)*
	2021 - ALMarie Curie SUPER *(*PI role)
	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"
Book	- Corinne Quadalti & **Caterina Garone.** The Human Mitochondrial Genome. Chapter 20: mtDNA maintenance: disease and therapy. EMSS 2020.
	- Tay Stacey, **Caterina Garone**, Salvatore DiMauro. Mitochondrial Encephalomyopathy. Chapter 168: 673-686. Book: International Neurology, Edited by Robert P. Lisak, Daniel Truong, William M. Carroll, Roongroj Bhidayasiri, 2nd Revised edition, 2016, New York, United States, John Wiley & Sons Inc.

Publications

1. **Garone C**, Pietra A, Nesci S. From the structural and (Dys)Function of ATP Synthase to Deficiency in Age-Related Diseases. Life (Basel). 2022 Mar 10;12(3):401.
2. Rebelo-Guiomar P, Pellegrino S, Dent KC, Sas-Chen A, Miller-Fleming L, **Garone C**, Van Haute L, Rogan JF, Dinan A, Firth AE, Andrews B, Whitworth AJ, Schwartz S, Warren AJ, Minczuk M.A late-stage assembly checkpoint of the human mitochondrial ribosome large subunit. Nat Commun. 2022 Feb 17;13(1):929.
3. Mancuso M, La Morgia C, Valentino ML, Ardissone A, Lamperti C, Procopio E, **Garone C**, Siciliano G, Musumeci O, Toscano A, Primiano G, Servidei S, Carelli V.SARS-CoV-2 infection in patients with primary mitochondrial diseases: Features and outcomes in Italy. Mitochondrion. 2021 May;58:243-245.
4. Boschetti E, D'Angelo R, Tardio ML, Costa R, Giordano C, Accarino A, Malagelada C, Clavenzani P, Tugnoli V, Caio G, Righi V, **Garone C**, D'Errico A, Cenacchi G, Dotti MT, Stanghellini V, Sternini C, Pironi L, Rinaldi R, Carelli V, De Giorgio R.Evidence of enteric angiopathy and neuromuscular hypoxia in patients with mitochondrial neurogastrointestinal encephalomyopathy. Am J Physiol Gastrointest Liver Physiol. 2021 May 1;320(5):G768-G779.
5. Mercatelli D, Balboni N, Giorgio F, Aleo E, **Garone C**, Giorgi FM.The Transcriptome of SH-SY5Y at Single-Cell Resolution: A CITE-Seq Data Analysis Workflow. Methods Protoc. 2021 May 6;4(2):28.
6. Protasoni M, Bruno C , Donati MA, Mohamoud K, Severino M, Allegri A, Robinson AJ, Reyes A, Zeviani M, ***Garone C***. Novel Compound Heterozygous Pathogenic Variants in Nucleotide-Binding Protein Like Protein (NUBPL) Cause Leukoencephalopathy With Multi-Systemic Involvement. *Mol Genet Metab*, 129 (1), 26-34. Jan 2020
7. Van Haute L, Lee SY, McCann BJ, Powell CA, Bansal D, Vasiliauskaitė L, ***Garone C***, Shin S, Kim JS, Frye M, Gleeson JG, Miska EA, Rhee HW, Minczuk M. NSUN2 introduces 5-methylcytosines in mammalian mitochondrial tRNAs. *Nucleic Acids Res*. 2019 Jul 5.
8. Domínguez-González C , Madruga-Garrido M, Mavillard F, **Garone C** et al. Deoxynucleoside Therapy for Thymidine Kinase 2-Deficient Myopathy *Ann Neurol*, 86 (2), 293-303. August 2019.

9. Manoli I, Sysol JR, Epping MW, Li L, Wang C, Sloan JL, Pass A, Gagné J, Ktena YP, Li L, Trivedi NS, Ouattara B, Zerfas PM, Hoffmann V, Abu-Asab M, Tsokos MG, Kleiner DE, ***Garone C***, Cusmano-Ozog K, Enns GM, Vernon HJ, Andersson HC, Grunewald S,Elkahloun AG, Girard CL, Schnermann J, DiMauro S, Andres-Mateos E, Vandenberghe LH, Chandler RJ, Venditti CP. FGF21 underlies a hormetic response to metabolic stress in methylmalonic acidemia. *JCI Insight.* 2018 Dec 6;3(23). pii: 124351.
10. **Garone C**, Viscomi C. Towards a therapy for mitochondrial disease: an update. *Biochem Soc Trans*. 2018 Oct 19;46(5):1247-1261.
11. **Garone C**, Taylor RW, Nascimento A, Poulton J et al. Retrospective natural history of thymidine kinase 2 deficiency. *J Med Genet*. 2018 Mar 30. pii: jmedgenet-2017-105012.
12. Kullar PJ, Gomez-Duran A, Gammage PA, **Garone C**, Minczuk M, Golder Z, Wilson J, Montoya J, Häkli S, Kärppä M, Horvath R, Majamaa K, Chinnery PF. Heterozygous SSBP1 start loss mutation co-segregates with hearing loss and the m.1555A>G mitochondrial DNA variant in a large multigenerational family. *Brain.* 2018 Jan 1;141(1):55-62.
13. Hamilton EMC, van der Lei HDW, Vermeulen G, Gerver JAM, Lourenço CM, Naidu S, Mierzewska H, Gemke RJBJ, de Vet HCW, Uitdehaag BMJ, Lissenberg-Witte BI; **VWM Research Group**, van der Knaap MS. Natural History of Vanishing White Matter. *Ann Neurol.* 2018 Aug;84(2):274-288.
14. Pacitti D, Levene M, **Garone C**, Nirmalananthan N, Bax BE. Mitochondrial Neurogastrointestinal Encephalomyopathy: Into the Fourth Decade, What We Have Learned So Far. *Front Genet.* 2018 Dec 21;9:669.
15. Feichtinger RG*, Oláhová M*, Kishita Y*, **Garone C*** et al. Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. The American Journal of Human Genetics (2017). The *American Journal of Human Genetics* (2017). 101(4):525-538.
16. **Garone C*,** D'Souza AR, Dallabona C et al. Defective mitochondrial rRNA methyltransferase MRM2 causes MELAS-like clinical syndrome. *Hum Mol Genet.* 2017 Nov 1;26(21):4257-4266.
17. Lopez-Gomez C, Levy R, Sanchez-Quintero M, Juanola-Falgarona M, Barca E, Garcia-Diaz B, Tadesse S, **Garone C*,** Hirano M*. Deoxycytidine and deoxythymidine treatment for thymidine kinase 2 deficiency. Annals of Neurology, 2017- *Ann Neurol.* 2017 May;81(5):641-652.
18. **Garone C**, Gurgel-Giannetti J, Sanna-Cherchi S, Krishna S, Naini A, Quinzii CM, Hirano M. A novel SUCLA2 mutation presenting as a complex childhood movement disorder. *J Child Neurol*. 2016 Sep 20.
19. 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 Oct 6;99(4):831-845.

20. Emmanuele V, Kubota A, Garcia-Diaz B, **Garone C**, Akman HO, Sánchez-Gutiérrez D, Escudero LM, Kariya S, Homma S, Tanji K, Quinzii CM, Hirano M. Fhl1 W122S causes loss of protein function and late-onset mild myopathy. *Hum Mol Genet.* 2015 Feb 1;24(3):714-26.

21. Kariya S, Obis T, **Garone C**, Akay T, Sera F, Iwata S, Homma S, Monani UR. Requirement of enhanced Survival Motoneuron protein imposed during neuromuscular junction maturation.*J Clin Invest*. 2014 Feb 3;124(2):785-800.

22. Garcia-Diaz B, **Garone C**, Barca E, Mojahed H, Gutierrez P, Pizzorno G, Tanji K, Arias- Mendoza F, Quinzii CM, Hirano M. Deoxynucleoside stress exacerbates the phenotype of a mouse model of mitochondrial neurogastrointestinal encephalopathy. *Brain.* 2014 May;137(Pt 5):1337-49.

23. **Garone C**, Garcia-Diaz B, Emmanuele V, Lopez LC, Tadesse S, Akman HO, Tanji K, Quinzii CM, Hirano M. Deoxypyrimidine monophosphate bypass therapy for thymidine kinase 2 deficiency. *EMBO Molecular Medicine*, EMBO Mol Med. 2014; 6(8):1016-2.

24. Yien Y, Robledo R, Schultz I, Takahashi-Makise N, Gwynn B, Bauer D, Dass A, Yi G, Li L, Hildick-Smith G, Cooney G, Pierce E, Mohler K, Dailey T, Miyata N, Kingsley P, **Garone C**, Hattangadi S, Huang H, Chen W, Keenan E, Shah D, Schlaeger T, DiMauro S, Orkin SH, Cantor A, Palis J, Koehler C, Lodish HF, Kaplan J, Ward D, Dailey H, Phillips J, Peters LL, Paw B. TMEM14C is required for erythroid mitochondrial heme metabolism. *J Clin Invest*. 2014 Oct;124(10):4294-304.

25. Quinzii CM*, **Garone C*,** Emmanuele V, Tadesse S, Krishna S, Dorado B, Hirano M.Tissue- specific oxidative stress and loss of mitochondria in CoQ-deficient Pdss2 mutant mice. *FASEB J.* 2013 Feb;27(2):612-21.

26. **Garone C**, Donati MA, Sacchini M, Garcia-Diaz B, Bruno C, Calvo S, Mootha VK, DiMauro S. Mitochondrial encephalomyopathy due to a novel mutation in ACAD9. *JAMA Neurology* 2013 Sep 1;70(9):1177-9.

27. Manoli I, Sysol JR, Li L, Houillier P, **Garone C,** Wang C, Zerfas PM, Cusmano-Ozog K, Young S, Trivedi NS, Cheng J, Sloan JL, Chandler RJ, Abu-Asab M, Tsokos M, Elkahloun AG, Rosen S, Enns GM, Berry GT, Hoffmann V, Dimauro S, Schnermann J, Venditti CP. Targeting proximal tubule mitochondrial dysfunction attenuates the renal disease of methylmalonic acidemia. *Proc Natl Acad Sci U S A.* 2013 Aug 13;110(33):13552-7.

28. Hildick-Smith GJ*, Cooney JD*, **Garone C*** et al. Macrocytic anemia and mitochondriopathy resulting from a defect in sideroflexin 4. *Am J Hum Genet.* 2013 Nov 7;93(5):906-14.

29. **Garone C,** Rubio JC, Calvo SE, Naini A, Tanji K, Dimauro S, Mootha VK, Hirano M. MPV17 Mutations Causing Adult-Onset Multisystemic Disorder With Multiple Mitochondrial DNA Deletions. *Arch Neurol.* 2012 Dec;69(12):1648-51.

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31. 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.

32. Cordelli DM, Aldrovandi A, Gentile V, **Garone C**, Conti S, Aceti A, Gennaro E, Zara F, Franzoni E. Fever as a seizure precipitant factor in Panayiotopoulos syndrome: a clinical and genetic study. *Seizure.* 2012 Mar;21(2):141-3.

33. Anderson BH, Kasher PR, Mayer J, Szynkiewicz M, Jenkinson EM, Bhaskar SS, Urquhart JE, Daly SB, Dickerson JE, O'Sullivan J, Leibundgut EO, J Muter J, Abdel-Salem GMH, Babul- Hirji R, Baxter P, Berger A, Bonafé L, Brunstom-Hernandez JE, Buckard JA, Chitayat D, Chong WK, Cordelli DM, Ferreira P, Fluss J, Forrest EH, Franzoni E, **Garone C**, Hammans SR, Houge G, Hughes I, Jacquemont S, Jeannet PY, Jefferson RJ Kumar R, Kutschke G, Lundberg S, Lourenço CM, Ramesh Mehta R, Naidu S, Ken K Nischal, Nunes L, Õunap K, Philippart M,Prabhakar P, Risen SR, Schiffmann R, Soh C, Stephenson JBP, Stewart H, Stone J, Tolmie JL, van der Knaap MS, Vieira JP, Vilain CN, Wakeling EL, Wermenbol V, Whitney A, Lovell SC, Meyer S, Livingston JH, Baerlocher GM, Black GCM, Rice GI& Crow J. Mutations in CTC1,encoding conserved telomere maintenance component 1, cause Coats plus. *Nature Genetics*, 2012; 44(3): 338-42.

34. Hirano M, **Garone C**, Quinzii CM. CoQ10 deficiencies and MNGIE: Two Treatable Mitochondrial Disorders *Biochimica Biophysica* Acta, 2012; 1820(5): 625-31.

35. DiMauro S, **Garone C.** Metabolic disorders of fetal life: Glycogenoses and mitochondrial defects of the mitochondrial respiratory chain. *Semin Fetal Neonatal Med.* 2011; 16(4): 181-9.

36. **Garone** C, Pippucci T, Cordelli DM, Zuntini R, Castegnaro G, Marconi C, Graziano C, Marchiani V, Verrotti A, Seri M, Franzoni E. FA2H-related disorders: a novel c.270+3A>T splice-site mutation leads to a complex neurodegenerative phenotype. *Dev Med Child Neurol*. 2011; 50(10): 958-61.

37. **Garone C**, Tadesse S, Hirano M. Clinical and genetic spectrum of mitochondrial neurogastrointestinal encephalomyopathy. *Brain* 2011; 134: 326-32.

38. Cordelli DM*, **Garone C*,** Marchiani V, Lodi R, Tonon C, Ferrari S, Seri M, Franzoni E. Progressive cerebral white matter involvement in a patient with Congenital Cataracts Facial Dysmorphisms Neuropathy (CCFDN). *Neuromuscul Disord*. 2010; 20(5): 343-5.

39. Franzoni E, **Garone C**, Marchiani V, Brunetto D, Tonon C, Lodi R, Bernardi B. A new case of idiopathic hemiplegia hemiconvulsion syndrome. *Neurol Sci.* 2010 Dec;31(6):799-805.

40. Ramesh V, Bernardi B, Stafa A, **Garone C**, Franzoni E, Abinun M, Mitchell P, Mitra D, Friswell M, Nelson J, Shalev SA, Rice GI, Gornall H, Szynkiewicz M, Aymard F, Ganesan V, Prendiville J, Livingston JH, Crow YJ. Intracerebral large artery disease

	in Aicardi-Goutie res syndrome implicates SAMHD1 in vascular homeostasis. *Dev Med Child Neurol*. 2010; 52(8): 725-32. ***41.*** DiMauro S, **Garone C**, Naini A. Metabolic myopathies. *Curr Rheumatol Rep. 2010*; 12(5): 386-93. **42.** DiMauro S, **Garone C**. Historical perspective on mitochondrial medicine. *Developmental Disabilities Research Review*, 2010; 16(2): 106 – 113. **43.** Franzoni E, Monti M, Pellicciari A, Muratore C, Verrotti A, **Garone C**, Cecconi I, Iero L, Gualandi S, Savarino F, Gualandi P. SAFA: A new measure to evaluate psychiatric symptoms detected in a sample of children and adolescents affected by eating disorders. Correlations with risk factors. *Neuropsychiatr Dis Treat*. 2009;5:207-14. **44.** Franzoni E, Gentile V, Pellicciari A, **Garone C**, Iero L, Gualandi S, Cordelli DM, Cecconi I, Moscano FC, Marchiani V, Errani A. Prospective study on long-term treatment with oxcarbazepine in pediatric epilepsy. *J Neurol.* 2009 Sep;256(9):1527-32. **45.** Coppola G, Franzoni E, Verrotti A, **Garone C**, Sarajlija J, Operto FF, Pascotto A. Levetiracetam or oxcarbazepine as monotherapy in newly diagnosed benign epilepsy of childhood with centrotemporal spikes (BECTS): an open-label, parallel group trial. *Brain Dev.* 2007 Jun;29(5):281-4. **46.** Franzoni E, Sarajlija J, **Garone C**, Malaspina E, Marchiani V. No kinetic interaction between levetiracetam and cyclosporine: a case report. *J Child Neurol.* 2007 Apr;22(4):440-2. **47.** Franzoni E, Verrotti A, Sarajlija J, **Garone C**, Matricardi S, Salerno GG, Monti M, Chiarelli F. Topiramate: effects on serum lipids and lipoproteins levels in children. *Eur J Neurol.* 2007 Dec;14(12):1334-7. **48.** Franzoni E, **Garone C**, Sarajlija J, Gualandi S, Malaspina E, Cecconi I, Moscano FC, Marchiani V. Open prospective study on oxcarbazepine in epilepsy in children: a preliminary report. *Seizure.* 2006 Jul;15(5):292-8. **49.** Franzoni E, Marchiani V, Cecconi I, Moscano FC, Gualandi S, **Garone C**,Sarajlija J, Malaspina E. Preliminary report on effects of oxcarbazepine-treatment on serum lipid levels in children. *Eur J Neurol.* 2006 Dec;13(12):1389-91. **50.** Franzoni E, Van der Knaap MS, Errani A, Colonnelli MC, Bracceschi R, Malaspina E, Moscano FC, **Garone C**, Sarajlija J, Zimmerman RA, Salomons GS, Bernardi B. Unusual diagnosis in a child suffering from juvenile Alexander disease: clinical and imaging report. *J Child Neurol*. 2006 Dec;21(12):1075-80.
Public engamente activities	2019 - Scientific Committee for the meeting “Science, Policy and the Public in Italy 2019”, Cambridge University Italian Society, Cambridge, UK https://spp2019.github.io/conference/
	2019 - Charlie Gard Foundation fundraising event in London, UK
	2019 - European Scientist Night, University for Bologna, Bologna, Italy
	2018 - Athena Swan representative for the MRC Mitochondrial Biology Unit, University of Cambridge, Cambridge, UK

	2017/2018 - “Mitochondrial Disease Awareness Day” and “Rare Disease Awareness Day”, MRC Mitochondrial Biology Unit, University of Cambridge, Cambridge, UK
	2016 - “Science Day”, Anglia Ruskin University, Cambridge, UK
	2015/ 2016 - “Cambridge Science Festival”, MRC Mitochondrial Biology Unit, University of Cambridge, Cambridge, UK http://www.mrc-mbu.cam.ac.uk/public-engagement
	2014- “Tk2cure” fundraising event in Miami, US