

Marcella Attimonelli

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Experience

Associate Professor in Molecular Biology

Department of Biosciences,
Biotechnology and Biopharmaceutics,
University Aldo Moro
Bari
- Currently

Chair Academic SpinOff

**BROWSer: Bioinformatics Resource
for Omics Wide Services**
Bari
October-2016 - Currently

BROWSer is a spin-off of the University of Bari, which provides healthcare users with genomics experiences and services function

Member of the MSeqDR Consortium

<https://mseqdr.org>
October-2012 - Currently

MSeqDR: the Mitochondrial Disease Sequence Data Resource Consortium - A global effort, 100+ mitochondrial disease experts.

Member of the Mitochondrial Disease Variant Curation Expert Panel

ClinGen
- Currently

Expert curation to assess variant pathogenicity in the most prevalent and/or actionable causes of Leigh syndrome, Leigh-like syndrome, and pediatric-onset mitochondrial encephalopathy syndromes in both nuclear and mitochondrial DNA will be performed, and facilitated by utilization of the Mitochondrial Disease Sequence Data Resource, MSeqDR. Over time, we will continue to broaden our focus to include variant curation for additional causes of primary mitochondrial diseases.

Delegate for Bari University within Elixir-IIB

Elixir
Europe
- Currently

Elixir-IIB is the Italian node of the European Elixir Infrastructure. Elixir-IIB is a network of universities and research centres; each node of the consortium has nominated a delegate.

Education

Chemistry Degree
Università Aldo Moro Bari
Italy

Thesis in Theoretical Physical Chemistry, vote 110/110 and honors

Skills

Bioinformatic Technologies, Comparative Genomics, In silico analysis to support clinical research, Design and implementation of biological databases

Languages

English

Good

Projects

- Design of Human Mitochondrial Databases: HmtDB and HmtVar
 - Design of the MToolBox package for the assembly and annotation of human mitochondrial genomes starting from Whole Exome data
 - Project coordinator for the implementation of the MEWAs package aimed at the recognition of allelic co-occurrences in human mtDNA sites and of nuclear genes expressed in the mitochondrion.
 - Analysis of human mitochondrial variability in oncological studies
 - Development of compilation of nuclear sequences of mitochondrial origin (NumtS) related to 23 eukaryotic species
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Publications

Author of 70 publication of which 40 open access. Here below a list of the most recent publications featuring major expertise of Marcella Attimonelli

1: Shen L, Attimonelli M, Bai R, Lott MT, Wallace DC, Falk MJ, Gai X. MSeqDR mvTool: A mitochondrial DNA Web and API resource for comprehensive variant annotation, universal nomenclature collation, and reference genome conversion. *Hum Mutat.* 2018 Jun;39(6):806-810. doi: 10.1002/humu.23422. Epub 2018 Apr 6. PubMed PMID: 29539190; PubMed Central PMCID: PMC5992054.

2: Kürschner G, Zhang Q, Clima R, Xiao Y, Busch JF, Kilic E, Jung K, Berndt N, Bulik S, Holzhütter HG, Gasparre G, Attimonelli M, Babu M, Meierhofer D. Renal oncocytoma characterized by the defective complex I of the respiratory chain boosts the synthesis of the ROS scavenger glutathione. *Oncotarget.* 2017 Nov 11;8(62):105882-105904. doi: 10.18632/oncotarget.22413. eCollection 2017 Dec 1. PubMed PMID: 29285300; PubMed Central PMCID: PMC5739687.

3: Calabrese FM, Balacco DL, Preste R, Diroma MA, Forino R, Ventura M, Attimonelli M. NumtS colonization in mammalian genomes. *Sci Rep.* 2017 Nov 27;7(1):16357. doi: 10.1038/s41598-017-16750-2. PubMed PMID: 29180746; PubMed Central PMCID: PMC5703718.

4: Diroma MA, Lubisco P, Attimonelli M. A comprehensive collection of annotations to interpret sequence variation in human mitochondrial transfer RNAs. *BMC Bioinformatics*. 2016 Nov 8;17(Suppl 12):338. doi: 10.1186/s12859-016-1193-4. PubMed PMID: 28185569; PubMed Central PMCID: PMC5123245.

5: Clima R, Preste R, Calabrese C, Diroma MA, Santorsola M, Scioscia G, Simone D, Shen L, Gasparre G, Attimonelli M. HmtDB 2016: data update, a better performing query system and human mitochondrial DNA haplogroup predictor. *Nucleic Acids Res*. 2017 Jan 4;45(D1):D698-D706. doi: 10.1093/nar/gkw1066. Epub 2016 Nov 28. PubMed PMID: 27899581; PubMed Central PMCID: PMC5210550.

6: Shen L, Diroma MA, Gonzalez M, Navarro-Gomez D, Leipzig J, Lott MT, van Oven M, Wallace DC, Muraresku CC, Zolkipli-Cunningham Z, Chinnery PF, Attimonelli M, Zuchner S, Falk MJ, Gai X. MSeqDR: A Centralized Knowledge Repository and Bioinformatics Web Resource to Facilitate Genomic Investigations in Mitochondrial Disease. *Hum Mutat*. 2016 Jun;37(6):540-548. doi: 10.1002/humu.22974. Epub 2016 Mar 21. PubMed PMID: 26919060; PubMed Central PMCID: PMC4846568.

7: Fedi S, Barberi TT, Nappi MR, Sandri F, Booth S, Turner RJ, Attimonelli M, Cappelletti M, Zannoni D. The Role of cheA Genes in Swarming and Swimming Motility of *Pseudomonas pseudoalcaligenes* KF707. *Microbes Environ*. 2016 Jun 25;31(2):169-72. doi: 10.1264/jjsme2.ME15164. Epub 2016 May 3. PubMed PMID: 27151656; PubMed Central PMCID: PMC4912153.

8: Santorsola M, Calabrese C, Girolimetti G, Diroma MA, Gasparre G, Attimonelli M. A multi-parametric workflow for the prioritization of mitochondrial DNA variants of clinical interest. *Hum Genet*. 2016 Jan;135(1):121-36. doi: 10.1007/s00439-015-1615-9. Epub 2015 Nov 30. PubMed PMID: 26621530; PubMed Central PMCID: PMC4698288.

9: Vidone M, Clima R, Santorsola M, Calabrese C, Girolimetti G, Kurelac I, Amato LB, Iommarini L, Trevisan E, Leone M, Soffietti R, Morra I, Faccani G, Attimonelli M, Porcelli AM, Gasparre G. A comprehensive characterization of mitochondrial DNA mutations in glioblastoma multiforme. *Int J Biochem Cell Biol*. 2015 Jun;63:46-54. doi: 10.1016/j.biocel.2015.01.027. Epub 2015 Feb 7. PubMed PMID: 25668474.

10: Diroma MA, Calabrese C, Simone D, Santorsola M, Calabrese FM, Gasparre G, Attimonelli M. Extraction and annotation of human mitochondrial genomes from 1000 Genomes Whole Exome Sequencing data. *BMC Genomics*. 2014;15 Suppl 3:S2. doi: 10.1186/1471-2164-15-S3-S2. Epub 2014 May 6. PubMed PMID: 25077682; PubMed Central PMCID: PMC4083402.

11: Falk MJ, Shen L, Gonzalez M, Leipzig J, Lott MT, Stassen AP, Diroma MA, Navarro-Gomez D, Yeske P, Bai R, Boles RG, Brilhante V, Ralph D, DaRe JT, Shelton R, Terry SF, Zhang Z, Copeland WC, van Oven M, Prokisch H, Wallace DC, Attimonelli M, Krotoski D, Zuchner S, Gai X; MSeqDR Consortium Participants; MSeqDR Consortium participants: Sherri Bale, Jirair Bedoyan, Doron Behar, Penelope Bonnen, Lisa Brooks, Claudia Calabrese, Sarah Calvo, Patrick Chinnery, John Christodoulou, Deanna Church,; Rosanna Clima, Bruce H. Cohen, Richard G. Cotton, IFM de Coo, Olga Derbenevoa, Johan T. den Dunnen, David Dimmock, Gregory Enns, Giuseppe Gasparre,; Amy Goldstein, Iris Gonzalez, Katrina Gwinn, Sihoun Hahn, Richard H. Haas, Hakon Hakonarson, Michio Hirano, Douglas Kerr, Dong Li, Maria Lvova, Finley Macrae, Donna Maglott, Elizabeth McCormick, Grant Mitchell, Vamsi K. Mootha, Yasushi Okazaki,; Aurora Pujol, Melissa Parisi, Juan Carlos Perin, Eric A. Pierce, Vincent Procaccio, Shamima Rahman, Honey Reddi, Heidi Rehm, Erin Riggs, Richard Rodenburg, Yaffa Rubinstein, Russell Saneto, Mariangela Santorsola, Curt Scharfe,; Claire Sheldon, Eric A. Shoubridge, Domenico Simone, Bert Smeets, Jan A. Smeitink, Christine Stanley, Anu Suomalainen, Mark Tarnopolsky, Isabelle Thiffault, David R. Thorburn, Johan Van Hove, Lynne Wolfe, and Lee-Jun Wong. Mitochondrial Disease Sequence Data Resource (MSeqDR): a global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. *Mol Genet Metab.* 2015 Mar;114(3):388-96. doi: 10.1016/j.ymgme.2014.11.016. Epub 2014 Dec 4. Review. PubMed PMID: 25542617; PubMed Central PMCID: PMC4512182.

12: Calabrese C, Simone D, Diroma MA, Santorsola M, Guttà C, Gasparre G, Picardi E, Pesole G, Attimonelli M. MToolBox: a highly automated pipeline for heteroplasmy annotation and prioritization analysis of human mitochondrial variants in high-throughput sequencing. *Bioinformatics.* 2014 Nov 1;30(21):3115-7. doi: 10.1093/bioinformatics/btu483. Epub 2014 Jul 14. PubMed PMID: 25028726; PubMed Central PMCID: PMC4201154.

13: Calabrese C, Mangiulli M, Manzari C, Paluscio AM, Caratozzolo MF, Marzano F, Kurelac I, D'Erchia AM, D'Elia D, Licciulli F, Liuni S, Picardi E, Attimonelli M, Gasparre G, Porcelli AM, Pesole G, Sbisà E, Tullo A. A platform independent RNA-Seq protocol for the detection of transcriptome complexity. *BMC Genomics.* 2013 Dec 5;14:855. doi: 10.1186/1471-2164-14-855. PubMed PMID: 24308330; PubMed Central PMCID: PMC4046740.

14: Tommaseo-Ponzetta M, Mona S, Calabrese F, Konrad G, Vacca E, Attimonelli M. Mountain pygmies of Western New Guinea: a morphological and molecular approach. *Hum Biol.* 2013 Feb-Jun;85(1-3):285-308. PubMed PMID: 24297230.