Mariarita Bertoldi
Dipartimento di Neuroscienze, Biomedicina e Movimento, Sezione di Chimica Biologica, Università di Verona
email: mita.bertoldi@univr.it

Formazione:
-1994, Università di Parma, Italia, laurea in Scienze Biologiche-Biomolecolari
-1999 Università di Verona, Italia, Dottorato di Ricerca in Biochimica

Posizione:
-1994, Set-Nov: Borsa di Ricerca, Istituto di Chimica Biologica, Università di Verona, Facoltà di Medicina e Chirurgia
- 1994, Nov-1999 Mar: Studente di Dottorato in Biochimica, Sezione di Chimica Biologica, Università di Verona, Facoltà di Medicina e Chirurgia
-1999-2004: Ricercatore Universitario di Biochimica BIO/10, Sezione di Chimica Biologica, Università di Verona, Facoltà di Medicina e Chirurgia
-2005-tutt'oggi Professore Associato di Biochimica BIO/10, Sezione di Chimica Biologica, Università di Verona, Facoltà di Medicina e Chirurgia
-2017-Abilitazione Nazionale al Ruolo di Professore Ordinario di Biochimica BIO/10

Corsi di insegnamento:
-1999 a tutt'oggi: Chimica e propedeutica Biochimica, Corso di Laurea Magistrale a ciclo unico in Medicina e Chirurgia, Università di Verona
-2001- a tutt'oggi: Corso di Biochimica per parecchie lauree triennali (Fisioterapia, Tecnico di Laboratorio, Ostetricia, Scienze Motorie, Igiene Dentale, Scienze Nutraceutiche e della Salute Alimentare), Università di Verona
-dal 2015 a tutt'oggi: Componente della Commissione didattica di Science Motorie
-dal 2023: Referente della Commissione di Pratiche Riconoscimento Crediti Studenti per il corso di laurea in Medicina e Chirurgia.
-Dal 2006-2015: membro eletto del consiglio della scuola di dottorato in Scienze della Vita e della Salute.
-A tutt'oggi: membro del collegio docenti del Corso di Dottorato in Medicina Biomolecolare, Università di Verona.
-E' stata ed è supervisore di una decina di studenti di dottorato negli ultimi 8 anni.

Attività istituzionale:
-2014-2019: Rappresentante dell'Area Medica per il Presidio di Qualità di Ateneo.

Scientific activity and honours:
Gli interessi scientifici sono focalizzati sulla chimica delle proteine e l'enzimologia. In dettaglio è interessata a: 1) enzimi dipendenti dal piridossal 5'-fosfato, 2) oligomeri di ribonucleasi pancreatica bovina che rappresentano modelli di aggregazione proteica, 3) perossiredossina 2, un nuovo essenziale enzima redox per gli eritrociti e il sistema nervoso centrale. Negli ultimi anni si interessa di enzimi quali la decarbossilasi degli amino acidi aromatici e la succinico semialdeide deidrogenasi le cui mutazioni provocano malattie genetiche dei neurotrasmettitori. Dal punto di vista metodologico la prof. Bertoldi studia le relazioni struttura-funzione di proteine, enzimi, la cinetica enzimatica e i meccanismi di inibizione utilizzando una combinazione di approcci e metodi biofisici e biochimici quali la spettroscopia in assorbimento, fluorescenza e dicroismo circolare, la cinetica allo stato prestazionario, la separazione cromatografica ed elettroforetica, oltre alla tecniche di base di biologia molecolare.
E' membro della Società Italiana di Biochimica e Biologia Molecolare dal 1997.
Nel 2002 ha vinto il Young Investigator Prize di Mc Graw-Hill-The Italian Biochemistry and Molecular Biology Society.
E' stata invitata a tenere presentazioni orali in parecchi meeting nazionali e internazionali e ha partecipato al comitato scientifico del International Advisory Panel of next International Meeting on vitamins and cofactors which took place in Athens, Georgia, USA from 26 to 31 Oct., 2008 e a quello organizzativo di Proteine 2018, Verona, Maggio 2018.

E’ revisore per molte riviste biochimiche di medio-alto impact factor (come esempio: Protein Science, Biochem. Biophys. Acta, Archives of Biochemistry and Biophysics, Biochemical Journal)
E’ attualmente supervisore di un gruppo composto da 5 post-docs, 2 dottoranti e 2 tesisti magistrali e 2 tesisti triennali.

Finanziamenti:
-187,499 euro finanziamento biennale PRIN2022 2022BTMTP8-A cross-sectional integrated approach to enable genotype/phenotype correlations in AADC deficiency: from protein analysis to an in vivo worm model towards precision medicine-MUR grant-Coordinatore Nazionale (individual project: 97,499 euro) 2023-2025 (with Eva Trevisson, UNIPD)
-140,000 euro PTC Therapeutics 2023-2025 per il progetto: A tool for severity prediction in AADC deficiency by analysis of the AADC protein population and correlation with the genotype.
-European Joint Programme-Rare Diseases-NSS 40-46300-98-1042: (Euro 30,000) 2021-2022 Rare neurotransmitter diseases: from novel research to focused treatments through iNTD network strengthening and patient empowerment, Principale Investigatore per l’Unità di Verona
-Research Grant (Euro 35,500) da GC Pharma per il progetto “Molecular basis for enzyme replacement therapy in SSADH deficiency” (Feb 2022-Mar 2023).
-Research Grant ((€ 78,300) da PTC Therapeutics per il progetto A molecular approach of AADC deficiency as a prerequisite to personalized therapy (Mag 2021-Apr 2023), come Investigatore Principale.
-Research Grant (€ 70,000) da AADC Research Trust and Agilent Biopharma per il progetto " Molecular insights into AADC deficiency" (Lug 2017-Giu 2019), come Investigatore Principale.
-Research Grant (€ 17,534) Joint Project, Università di Verona. "Aromatic amino acid decarboxylase deficiency is strictly connected to pyridoxine-related seizures: a biochemical approach to gain insight into inherited neurotransmitters disorders" (Gen 2018-Dic 2019), come Investigatore Principale.
-Dal 2010 a tutt’oggi: finanziamento annuale da Università di Verona (ex60% now FUR). Gli ultimi sono stati: FUR2016, FUR2017, FUR2018, FUR2019; FUR2020, FUR2021, FUR2022 e FUR2023 come Investigatore Principale.
-2001-2003 Finanziamento CNR N°CU01.00922.CT26 (equivalente di € 6,000): "Interaction of Dopa decarboxylase with serotonin" come Investigatore Principale.
-Parecchi progetti PRIN come partecipante (1999, 2001, 2005, 2007).

List of publications
1) M. Bertoldi, P.S. Moore, B. Maras, P. Dominici and C. Borri Voltattorni "Mechanism-based inactivation of Dopa decarboxylase by serotonin" **J. Biol. Chem**. (1996), 271, 23954-23959
2) M. Libonati, M. Bertoldi and S. Sorrentino "The activity on double-stranded RNA of aggregates of ribonuclease A higher than dimers increases as a function of the size of the aggregates" **Biochem. J.** (1996), 318, 287-290
3) M. Bertoldi, G. Gotte, S. Sorrentino and M. Libonati "Artificial Ribonuclease A oligomers degrade double-stranded RNA with efficiency that increases as a function of the size of the oligomer" **It. J. Biochem**. (1996), 45, 186-187
4) P. Dominici, P.S. Moore, S. Castellani, M. Bertoldi and C. Borri Voltattorni "Mutation of cysteine 111 in Dopa decarboxylase leads to active site perturbation" **Protein Sci.** (1997)**,** 6**,** 2007-2015
5) P.S. Moore, M. Bertoldi, P. Dominici and C. Borri Voltattorni "Aromatic amino acid methyl ester analogs form quinonoidal species with Dopa decarboxylase" **FEBS Lett**. (1997), 412, 245-248
6) M Bertoldi, P. Dominici, P.S. Moore, B. Maras and C. Borri Voltattorni "Reaction of Dopa decarboxylase with α-methyldopa leads to an oxidative deamination producing 3,4-dihydroxyphenylacetone, an active site-directed affinity label" **Biochemistry** (1998), 37, 6552-6561
7) M. Bertoldi, P. Frigeri, M. Paci and C. Borri Voltattorni “Reaction specificity of native and nicked 3, 4 dihydroxyphenylalanine decarboxylase” **J. Biol. Chem**. (1999), 274, 5514-5521
8) M. Bertoldi, V. Carbone and C. Borri Voltattorni “Ornithine and glutamate decarboxylase catalyse an oxidative deamination of their alpha-methyl substrates” **Biochem. J.** (1999)**,** 342, 507-510
9) G. Gotte, M. Bertoldi and M. Libonati “Structure versatility of bovine ribonuclease A: distinct conformers of trimeric and tetrameric aggregates of the enzyme” **Eur. J. Biochem**. (1999), 265, 680-687
10) M. Bertoldi and C. Borri Voltattorni “Reaction of Dopa decarboxylase with L-aromatic amino acids under aerobic and anaerobic conditions” **Biochem. J.** (2000)**,** 352, 533-538

11) M. Bertoldi, S. Castellani and C. Borri Voltattorni "Mutation of residues in the coenzyme binding pocket of Dopa decarboxylase: effects on catalytic properties" **Eur. J. Biochem**. (2001), 268, 2975-81
12) M. Bertoldi, M. Gonsalvi and C. Borri Voltattorni "Green tea polyphenols: novel irreversible inhibitors of Dopa decarboxylase" **Biochem. Biophys. Res. Commun**. (2001), 284, 90-93
13) M. Bertoldi and C. Borri Voltattorni " Dopa decarboxylase exhibits low pH half transaminase and high pH oxidative deaminase activities toward serotonin (5-hydroxytryptamine)" **Protein Sci.** (2001), 10, 1178-86
14) A. Nenci, G. Gotte, M. Bertoldi and M. Libonati "Structural properties of trimers and tetramers of Ribonuclease A" **Protein Sci.** (2001), 10, 2017-2027
15) C. Borri Voltattorni and M. Bertoldi "Dopa decarboxylase" in **Encyclopedia of Molecular Medicine** (T. Creighton, ed.) (2002), vol.2 pp.1089-1093, Wiley and Sons, Inc.
16) M. Bertoldi, B. Cellini, T. Clausen and C. Borri Voltattorni "Spectroscopic and kinetic analyses reveal the pyridoxal 5'-phosphate binding mode and the catalytic features of Treponema denticola cystalysin" **Biochemistry** (2002), 41, 9153-9164
17) C. Borri Voltattorni, M. Bertoldi, S. Bianconi, W. Deng, K. Wong, I. Kim, B. Herbert and K.L. Kirk "Behavior of fluorinated analogs of L-(3,4-dihydroxyphenyl)alanine and L-threo-(3,4-dihydroxyphenyl)serine as substrates for Dopa decarboxylase" **Biochem. Biophys. Res. Commun.** (2002), 295, 107-111
18) M. Bertoldi, M. Gonsalvi, R. Contestabile, and C. Borri Voltattorni "Mutation of tyrosine 332 to phenylalanine converts Dopa decarboxylase into a decarboxylation-dependent oxidative deaminase" **J. Biol. Chem.** (2002), 277, 36357-36362
19) M. Bertoldi and C. Borri Voltattorni "Reaction and substrate specificity of pig kidney dopa decarboxylase under aerobic and anaerobic conditions" **Biochem.Biophys. Acta** (2003), 1647, 42-47.
20) M. Bertoldi, B. Cellini, A. Paiardini, M.L. di Salvo and C. Borri Voltattorni. "*Treponema denticola* cystalysin exhibits a significant alanine racemase activity accompanied by transamination: mechanistic implications." **Biochem J.** (2003) 371, 473-483.
21) M. Bertoldi, B. Cellini, S. D'Aguanno, and C. Borri Voltattorni "Lysine 238 is an essential residue for alfa, beta-elimination catalyzed by Treponema denticola cystalysin" **J. Biol Chem**. (2003), 278, 37336-37343
22) B. Cellini, M. Bertoldi and C. Borri Voltattorni "Treponema denticola cystalysin catalyzes beta-desulfination of L-cysteine sulfinic acid and beta-decarboxylation of L-aspartate and oxalacetate**" FEBS Lett.** (2003), 554, 306-310.
23) B. Cellini, M.Bertoldi, A.Paiardini, S.D'Aguanno and C. Borri Voltattorni "Site-directed mutagenesis provides insight into racemization and transamination of alanine catalyzed by Treponema denticola cystalysin". **J Biol Chem**. (2004), 279, 36898-36905
24) M. Bertoldi, B. Cellini, D.V. Laurents and C. Borri Voltattorni "Folding pathway of the pyridoxal 5'-phosphate C-S lyase MalY from Escherichia coli" **Biochem J**. (2005) 389, 885-898
25) B. Cellini, M. Bertoldi, R. Montioli and C. Borri Voltattorni "Probing the role of Tyr 64 of Treponema denticola cystalysin by site-directed mutagenesis and kinetic studies" **Biochemistry** (2005) 44, 13970-13980-**first co-author**
26) M. Bertoldi, B. Cellini, B. Maras and C. Borri Voltattorni "A quinonoid is an intermediate of oxidative deamination reaction catalyzed by Dopa decarboxylase" **FEBS Lett** (2005) 579, 5175-5180
27) B. Cellini, R. Montioli, A. Bossi, M. Bertoldi, D.V. Laurents, and C. Borri Voltattorni "Holo- and apo-cystalysin from Treponema denticola: two different conformations" **Arch. Biochem. Biophys**. (2006) 455, 31-39
28) B. Cellini, M. Bertoldi, R. Montioli, D.V. Laurents, A. Paiardini and C. Borri Voltattorni "Dimerization and folding processes of Treponema denticola cystalysin: the role of pyridoxal 5'-phosphate" **Biochemistry** (2006) 45, 14140-14154
29) Candeloro P, Voltattorni CB, Perniola R, Bertoldi M, Betterle C, Mannelli M, Giordano R, De Bellis A, Tiberti C, Laureti S, Santeusanio F, Falorni A. "Mapping of human autoantibody epitopes on aromatic l-amino acid decarboxylase**. J Clin Endocrinol Metab**. (2007), Mar;92(3):1096-105
30) A. Amadasi, M. Bertoldi, R. Contestabile S. Bettati, B. Cellini, M.L. di Salvo, C. Borri-Voltattorni, F. Bossa and A. Mozzarelli "Pyridoxal 5'-Phosphate Enzymes as Targets for Therapeutic Agents" Current Medicinal Chemistry, (2007), 14, **Curr Med Chem**. 2007;14(12):1291-324. Review.
31) Cellini B, Bertoldi M, Montioli R, Paiardini A, Borri Voltattorni C. Human wild-type alanine: glyoxylate aminotransferase and its naturally occurring G82E variant: functional properties and physiological implications **Biochem J.** 2007, 408, 39-50

32) Bertoldi M; Cellini B; Paiardini A.; Montioli R and Borri Voltattorni C. “Reactions of human liver peroxisomal alanine:glyoxylate aminotransferase with beta-chloro-L-alanine and L-cysteine: spectroscopic and kinetic analysis. **Biochimica Biophysica Acta**, 2008, 1784(9):1356-62
33) Bertoldi M*, Cellini B, Montioli R and Borri Voltattorni C. “Insights into the mechanism of oxidative deamination catalyzed by Dopa decarboxylase”. **Biochemistry,** 2008 47(27):7187-95
34) Montioli R, Cellini B, Bertoldi M, Paiardini A and Borri Voltattorni C “An engineered folded PLP-bound monomer of Treponema denticola cystalisin reveleals the effect of the dimeric structure on the catalytic properties of the enzyme” **Proteins** 2009, 74(2):304-17.
35) Bertoldi M. * and Borri Voltattorni C. “Multiple roles of the active site lysine of Dopa decarboxylase” **Arch. Biochem. Biophys** 2009, 488, 130-9
36) A. Siciliano, F. Turrini, M. Bertoldi, A. Matte’, A. Pantaleo, O. Olivieri and L. De Franceschi “Dexygenation affects tyrosine phosphoproteome of red cell membrane from patients with sickle cell disease” **Blood Cell Mol & Dis.** 2010, 44, 233-242
37) A. Matté, P.S. Low, F. Turrini, M. Bertoldi, M.E. Campanella, D. Spano, A. Siciliano and L. De Franceschi “Peroxiredoxin-2 (Prx2) expression is increased in β thalassemic mouse red cells but displaced from the membrane as a marker of oxidative stress” **Free Rad. Biol. Med.** 2010, 49(3):457-66
38) De Franceschi L, Bertoldi M, De Falco L, Santos Franco S, Ronzoni L, Turrini F, Colancecco A, Camaschella C, Cappellini MD, Iolascon A. "Oxidative stress modulates heme synthesis and induces peroxiredoxin-2 as a novel cytoprotective response in beta thalassemic erythropoiesis" **Haematologica.** 2011 96(11):1595-604
39) De Franceschi L, Tomelleri C, Matte A, Brunati AM, Bovee-Geurts PH, Bertoldi M, Lasonder E, Danek A , Walker RH , Jung HH, Bader B, Siciliano A, Ferru E, Mohandas N, Bosman GJCGM "Erythrocyte membrane changes of chorea-acanthocytosis are the result of altered lyn-kinase activity (2011) **BLOOD** 118(20):5652-63.
40) Hoefig CS, Renko K, Piehl S, Scanlan TS, Bertoldi M, Opladen T, Hoffmann GF, Klein J, Blankenstein O, Schweizer U, Köhrle J. Does the aromatic L-amino acid decarboxylase contribute to thyronamine biosynthesis? **Mol Cell Endocrinol**. 2012 Feb 26;349(2):195-201
41) De Franceschi L, Franco RS, Bertoldi M, Brugnara C, Matté A, Siciliano A, Wieschhaus AJ, Chishti AH, Joiner CH. Pharmacological inhibition of calpain-1 prevents red cell dehydration and reduces Gardos channel activity in a mouse model of sickle cell disease. **FASEB J**. 2013 Feb;27(2):750-9
42) Matte A, Bertoldi M*, Mohandas N, An X, Bugatti A, Maria Brunati A, Rusnati M, Tibaldi E, Siciliano A, Turrini F, Perrotta S, De Franceschi L. Membrane association of peroxiredoxin-2 in red cells is mediated by the N-terminal cytoplasmic domain of band 3. **Free Radic Biol Med**. 2013 Feb;55:27-35
43) De Franceschi L, Bertoldi M, Matte A, Santos Franco S, Pantaleo A, Ferru E, Turrini F. Oxidative stress and β-thalassemic erythroid cells behind the molecular defect. **Oxid Med Cell Longev**. 2013;2013:985210.. Review
44) Franco SS, De Falco L, Ghaffari S, Brugnara C, Sinclair DA, Matte' A, Iolascon A, Mohandas N, Bertoldi M, An X, Siciliano A, Rimmelé P, Cappellini MD, Michan S, Zoratti E, Anne J, De Franceschi L Resveratrol accelerates erythroid maturation by activation of FoxO3 and ameliorates anemia in beta-thalassemic mice. .
Haematologica. 2014 Feb;99(2):267-75.
45) Bertoldi M.* Mammalian Dopa decarboxylase: structure, catalytic activity and inhibition. Arch Biochem Biophys. 2014 Mar 15;546:1-7
46) Matté A, Pantaleo A, Ferru E, Turrini F, Bertoldi M, Lupo F, Siciliano A, Ho Zoon C, De Franceschi L. The novel role of peroxiredoxin-2 in red cell membrane protein homeostasis and senescence. **Free Radic Biol Med.** 2014 Nov;76:80-8. doi: 10.1016/j.freeradbiomed.2014.08.004. Epub 2014 Aug 21.
47) Bertoldi M* Human Peroxiredoxins 1 and 2 and Their Interacting Protein Partners; Through Structure Toward Functions of Biological Complexes. **Protein Pept Lett**. 2016;23(1):69-77
48) Montioli R, Voltattorni CB, Bertoldi M.* Parkinson's Disease: Recent Updates in the Identification of Human Dopa Decarboxylase Inhibitors. **Curr Drug Metab**. 2016;17(5):513-8. Review.
49) Montioli R, Paiardini A, Kurian MA, Dindo M, Rossignoli G, Heales SJ, Pope S, Voltattorni CB, Bertoldi M*.
The novel R347g pathogenic mutation of aromatic amino acid decarboxylase provides additional molecular insights into enzyme catalysis and deficiency. **Biochim Biophys Acta Protein and Proteomics.** 2016 Jun;1864(6):676-82. doi: 10.1016/j.bbapap.2016.03.011.

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51) Paiardini A, Giardina G, Rossignoli G, Voltattorni CB, Bertoldi M*. New insights emerging from recent investigations on human group II pyridoxal 5'-phosphate Decarboxylases. **Curr Med Chem**. 2017, 24 (3), 226-244
52) Astegno A, Maresi E, Bertoldi M, La Verde V, Paiardini A, Dominici P. Unique substrate specificity of ornithine aminotransferase from Toxoplasma gondii. **Biochem J**. 2017 Jan 26. pii: 474(6):939-955
53) Montioli R, Janson G, Paiardini A, Bertoldi M, Borri Voltattorni C Heterozygosis in aromatic amino acid decarboxylase deficiency: Evidence for a positive interallelic complementation between R347Q and R358H mutations.
IUBMB Life. 2018 Mar;70(3):215-223. doi: 10.1002/iub.1718. Epub 2018 Jan 22.
54) Rossignoli G, Phillips RS, Astegno A, Menegazzi M, Voltattorni CB, Bertoldi M*. Phosphorylation of pyridoxal 5'-phosphate enzymes: an intriguing and neglected topic. **Amino Acids**. 2018 Feb;50(2):205-215. doi: 10.1007/s00726-017-2521-3. Epub 2017 Dec 4. Review.
55) Rossignoli G, Grottesi A, Bisello G, Montioli R, Borri Voltattorni C, Paiardini A, Bertoldi M*. Cysteine 180 Is a Redox Sensor Modulating the Activity of Human Pyridoxal 5'-Phosphate Histidine Decarboxylase. **Biochemistry**. 2018 Nov 6;57(44):6336-6348. doi: 10.1021/acs.biochem.8b00625. Epub 2018 Oct 29.
56) Montioli R, Battini R, Paiardini A, Tolve M, Bertoldi M, Carducci C, Leuzzi V, Borri Voltattorni C. A novel compound heterozygous genotype associated with aromatic amino acid decarboxylase deficiency: Clinical aspects and biochemical studies. **Mol Genet Metab**. 2019 Jun;127(2):132-137. doi: 10.1016/j.ymgme.2019.05.004. Epub 2019 May 10.
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58) Montioli R, Bisello G, Dindo M, Rossignoli G, Voltattorni CB, Bertoldi M* New variants of AADC deficiency expand the knowledge of enzymatic phenotypes. **Arch Biochem Biophys**. 2020 Mar 30;682:108263. doi: 10.1016/j.abb.2020.108263. Epub 2020 Jan 15. PMID: 31953134
59) Menegazzi M, Campagnari R, Bertoldi M, Crupi R, Di Paola R, Cuzzocrea S. Protective Effect of Epigallocatechin-3-Gallate (EGCG) in Diseases with Uncontrolled Immune Activation: Could Such a Scenario Be Helpful to Counteract COVID-19? **Int J Mol Sci.** 2020 Jul 21;21(14):5171. doi: 10.3390/ijms21145171. PMID: 32708322 Free PMC article. Review.
60) Bisello G, Longo C, Rossignoli G, Phillips RS, Bertoldi M*. Oxygen reactivity with pyridoxal 5'-phosphate enzymes: biochemical implications and functional relevance. **Amino Acids.** 2020 Aug;52(8):1089-1105. doi: 10.1007/s00726-020-02885-6. Epub 2020 Aug 25.
61) Brennenstuhl H, Didiasova M, Assmann B, Bertoldi M, Molla G, Jung-Klawitter S, Kuseyri Hübschmann O, Schröter J, Opladen T, Tikkanen R Succinic Semialdehyde Dehydrogenase Deficiency: in vitro and in silico characterization of a novel pathogenic missense variant and analysis of the mutational spectrum of ALDH5A1-**Int. J. Mol. Sci.** 2020 doi: 10.3390/ijms21228578. PMID: 33203024
62) Astegno A., Conter C., Bertoldi M., and Dominici P. Structural insights into heme pocket and oligomeric state of non-symbiotic hemoglobins from Arabidopsis thaliana- **Biomolecules** 2020, 10, 1615-33
63) Mattè A, Federti E, Tibaldi E, Di Paolo ML, Bisello G, Bertoldi M*, Carpentieri A, Pucci P, Iatchencko I, Wilson AB, Riccardi V, Siciliano A, Turrini F, Kim DW, Choi SY, Brunati AM, De Franceschi L. Tyrosine Phosphorylation Modulates Peroxiredoxin-2 Activity in Normal and Diseased Red Cells. **Antioxidants (Basel)**. 2021 Feb 1;10(2):206.
64) Rossignoli R., Kramer K., Lugarà E., Alrashidi H., Pope S., De La Fuente Barrigon C., Barwick K., Bisello G., Ng J., Counsell J., Lignani G., Heales S.J.R., Bertoldi M*., Barral S., Kurian M.A. Aromatic L-amino acid decarboxylase deficiency: a patient-derived neuronal model for precision therapies, **BRAIN (Oxford)**, 2021, open access, DOI: 10.1093/brain/awab123 Brain. 2021 Sep 4;144(8):2443-2456
65) Longo C, Montioli R, Bisello G, Palazzi L, Mastrangelo M, Brennenstuhl H, de Laureto PP, Opladen T, Leuzzi V, Bertoldi M.* Compound heterozygosis in AADC deficiency: A complex phenotype dissected through comparison among heterodimeric and homodimeric AADC proteins**. Mol Genet Metab.** 2021 Aug 28:S1096-7192(21)00775-7

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68) Tokatly Latzer I, Bertoldi M, DiBacco ML, Arning E, Tsuboyama M, MacMullin P, Sachee D, Rotenberg A, Lee HHC, Aygun D, Opladen T, Jeltsch K, García-Cazorla À, Roullet JB, Gibson KM, Pearl PL. The presence and severity of epilepsy coincide with reduced γ-aminobutyrate and cortical excitatory markers in succinic semialdehyde dehydrogenase deficiency. **Epilepsia**. 2023 Jun;64(6):1516-1526. doi: 10.1111/epi.17592. Epub 2023 Apr 4. PMID: 36961285
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*corresponding or co-corresponding author

Publications (at 2023)
- Number of total Medline publications in peer-review journals: 75
- Total number of citations 1969 (Scopus)
- H index 26 (Scopus)
She also authored 3 chapters of book.

Chapters in books:

- C. Borri Voltattorni and M. Bertoldi "Dopa decarboxylase" in **Encyclopedia of Molecular Medicine** (T. Creighton, ed.) (2002), vol.2 pp.1089-1093, Wiley and Sons, Inc.

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*corresponding author