Mariarita Bertoldi
Dipartimento di Neuroscienze, Biomedicina e Movimento, Section of Biological Chemistry, University of Verona
email: mita.bertoldi@univr.it

Education:
-1994, University of Parma, Italy, graduated in Biomolecular Sciences
-1999 University of Verona, Italy, PhD in Biochemistry

Positions and Employment
-1994, Sept-Nov: Research Fellowship in Biochemistry, Institute of Biological Chemistry at the University of Verona, Faculty of Medicine
-1994, Nov-1999 March: PhD student in Biochemistry, Division of Biological Chemistry at the University of Verona, Faculty of Medicine
-1999-2004: Research Assistant at the University of Verona, Division of Biological Chemistry, Faculty of Medicine
-2005-present Associate Professor of Biochemistry at the Division of Biological Chemistry, Faculty of Medicine, University of Verona
-2017-National Qualification for Full Professor of Biochemistry

Teaching Courses:
-1999 until now: Chemistry and Structural Biochemistry for the School of Medicine, University of Verona
-2001-until now: Biochemistry for several 3-years bachelor courses (Physiotherapy, Laboratory Technician, Maternal Care and Specialization, Sport Sciences), University of Verona
-From 2006-2015: elected member of the Council of the School of Doctorate in Life and Health Sciences.
-until now: member of the teaching staff of the Doctorate in Biomolecular Medicine, University of Verona

-cooperation project of the University of Verona with the University of Ngozi (Burundi): teaching courses from 2007 to 2019 in Chemistry and Biochemistry for the course in Nursing, Ngozi, Burundi

Scientific activity and honours:
The scientific interest spans from protein chemistry to enzymology. In detail she has been interested in: a) pyridoxal 5'-phosphate enzymes, b) oligomers of bovine pancreatic ribonuclease that could represent a model for protein aggregation, c) peroxiredoxin-2: a novel essential redox enzyme for erythrocytes and central nervous system.
In particular, by means of a combination of biophysical and biochemical approaches and methods, she studies the structure-function relationships of proteins, the enzyme kinetics and the mechanisms of inhibition.
She is member of the Italian Biochemistry and Molecular Biology since 1997.
In 2002: Young Investigator Prize by Mc Graw-Hill and The Italian Biochemistry and Molecular Biology Society.
She has been invited speaker in several international and national scientific meetings.
She was part of the International Advisory Panel of next International Meeting on vitamins and cofactors which took place in Athens, Georgia, USA from 26 to 31 Oct., 2008.
She serves as reviewer for Biochemistry Journals.

-organizzazione Belgrado 2022
-invito proteine Pisa 2022
-invito alle guidelines Praga 2023 per aadc (revisione) e th (dicembre 2024 belgrado)
-invito a gerusalemme 2023 per SSIEM
-invito a co-chair a FEBS 2024
-organizzazione YSF Verona 2023
-guided poster SSIEM 2024 Porto

-clingen VCEP

Valutatore per:
Service des Programmes Incitatifs Scientifiques (SPIS) / Incentive Scientific Programs Division Institut Pasteur (2020)
FRRB Early Career (2024) grant

Fundings and grants:
-187,499 euro 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-National Coordinator (individual project: 97,499 euro) 2023-2025 (with Eva Trevisson, UNIPD)
-140,000 euro PTC Therapeutics 2023-2025 for the Project 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, PI of the Unit of Verona
-1-yr Research Grant (Euro 35,500) by GC Pharma for the project “Molecular basis for enzyme replacement therapy in SSADH deficiency” (February 2022-March 2023).
-2-years Research Grant ((€ 78,300) by PTC Therapeutics for the project A molecular approach of AADC deficiency as a prerequisite to personalized therapy (May 2021-April 2023), as Principal Investigator.
-2-years Research Grant (€ 70,000) by the AADC Research Trust and Agilent Biopharma for a project entitled " Molecular insights into AADC deficiency" (July 2017-June 2019), as Principal Investigator.
-2-years Research Grant (€ 17,534) Joint Project, University of Verona, title. "Aromatic amino acid decarboxylase deficiency is strictly connected to pyridoxine-related seizures: a biochemical approach to gain insight into inherited neurotransmitters disorders" (January 2018-December 2019), as Principal Investigator.
-From 2010 until now: each year: grant from the University of Verona (ex60% now FUR). The last were FUR2016, FUR2017, FUR2018, FUR2019; FUR2020, FUR2021, FUR2022 for basic research activies, as Principal Investigator.
-2001-2003 Italian National Research Council N°CU01.00922.CT26 (€ 6,000) title: "Interaction of Dopa decarboxylase with serotonin" as Principal Investigator.
-Several 2-year national research projects PRIN as participant (1999, 2001, 2005, 2007).

Incarichi istitutzionali
Componente del PdQ dal 2014 al 2019 (controlla) come rappresentante di area scienze vita e salute
Presidente della commissione riconoscimento carriere per il CdS in medicina
componente della commissione didattica per scienze motorie (dal 2014? al 2024)
Referente per la qualità del dottorato di Medicina Biomolecolare
Docente referente del 1 anno di medicina e selezionato per visita anvur

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.
50) Lupo F, Tibaldi E, Matte A, Sharma AK, Brunati AM, Alper SL, Zancanaro C, Benati D, Siciliano A, Bertoldi M, Zonta F, Storch A, Walker RH, Danek A, Bader B, Hermann A, De Franceschi L. A new molecular link between defective autophagy and erythroid abnormalities in chorea-acanthocytosis. **Blood**. 2016 Dec 22;128(25):2976-2987. doi: 10.1182/blood-2016-07-727321.
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.
57) Himmelreich N, Montioli R, Bertoldi M, Carducci C, Leuzzi V, Gemperle C, Berner T, Hyland K, Thöny B, Hoffmann GF, Voltattorni CB, Blau N. Aromatic amino acid decarboxylase deficiency: Molecular and metabolic basis and therapeutic outlook. **Mol Genet Metab**. 2019 May;127(1):12-22. doi: 10.1016/j.ymgme.2019.03.009. Epub 2019 Mar 27. Review.
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
66) Bisello G, Kusmierska K, Verbeek MM, Sykut-Cegielska J, Willemsen MAAP, Wevers RA, Szymańska K, Poznanski J, Drozak J, Wertheim-Tysarowska K, Rygiel AM, Bertoldi M*. The novel P330L pathogenic variant of aromatic amino acid decarboxylase maps on the catalytic flexible loop underlying its crucial role. **Cell Mol Life Sci**. 2022 May 20;79(6):305
67) Bisello G, Bertoldi M*., Compound heterozygosis in AADC deficiency and its complex phenotype in terms of AADC protein population, **Int. J. Mol. Sci** 2022, 23(19):11238. https://doi.org/10.3390/ijms231911238
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
69) Tokatly Latzer I, Hanson E, Bertoldi M, García-Cazorla À, Tsuboyama M, MacMullin P, Rotenberg A, Roullet JB, Pearl PL; SSADH Deficiency Investigators Consortium. Autism spectrum disorder and GABA levels in children with succinic semialdehyde dehydrogenase deficiency. **Dev Med Child Neurol**. 2023 May 28. doi: 10.1111/dmcn.15659. Online ahead of print. PMID: 37246331
70) Nastassja Himmelreich, Mariarita Bertoldi, Majid Alfadhel, Malak Ali Alghamdi, Yair Anikster, Xinhua Bao, Fahad A. Bashiri, Bruria Ben Zeev, Giovanni Bisello, Ahmet Cevdet Ceylan, Yin-Hsiu Chien, Yew Sing Choy, Sarah H. Elsea, Lisa Flint, Àngels García-Cazorla, Charul Gijavanekar, Emel Yılmaz Gümüş, Muddathir H. Hamad, Burcu Hişmi, Tomas Honzik, Oya Kuseyri Hübschmann, Wuh-Liang Hwu, Salvador Ibáñez-Micó, Kathrin Jeltsch, Natalia Juliá-Palacios, Çiğdem Seher Kasapkara, Manju A. Kurian, Katarzyna Kusmierska, Ning Liu, Lock Hock Ngu, John D. Odom, Winnie Petee Ong, Thomas Opladen, Mari Oppeboen, Phillip L. Pearl, Belén Pérez, Roser Pons, Agnieszka Magdalena Rygiel, Tan Ee Shien, Robert Spaull, Jolanta Sykut-Cegielska, Brahim Tabarki, Trine Tangeraas, Beat Thöny, Tessa Wassenberg, Yongxin Wen, Yusnita Yakob, Jasmine Goh Chew Yin, Jiri Zeman, Nenad Blau Prevalence of *DDC* genotypes in patients with aromatic L-amino acid decarboxylase (AADC) deficiency and *in silico* prediction of

structural protein changes. **Mol Genet Metab** 2023 Jul;139(3):107624. doi: 10.1016/j.ymgme.2023.107624. Epub 2023 Jun 2. PMID: 37348148-**first co-author**

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Publications (at Oct 2024)
- Number of total Medline publications in peer-review journals: 81
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Book chapters/Editorials/Letters:
- 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