

Replace with First name(s) Surname(s)

PERSONAL INFORMATION

# Barbara Cellini



- <u>https://www.unipg.it/personale/barbara.cellini</u>
- Replace with type of IM service

Sex Female | Date of birth 02/08/1976 | Nationality Italian

2017-present	Associate professor of Biochemistry, Department of Medicine and Surgery, University of Perugia, Italy				
Aprile 2018	Visiting researcher at "Dresden University of Technology", Germany, under the program Erasmus+ "Staff mobility for training"				
2005-2016	Assistant professor of Biochemistry, Department of neurosciences, Biomedicine and Movement Sciences, University of Verona, Italy.				
07/2010, 09/2010, 04/2011	Visiting scientist, Department of Cell and Developmental Biology University College London, UK				
EDUCATION AND TRAINING 2001-2004	[ PhD in Biochemical Sciences, Department of Neurological Sciences and Visione, University of Verona Supervisor: Prof. Carla Voltattorni	Replace with European Qualification Framework (or other) level if relevant			
2000	Degree in Biological Sciences University of Urbino, score 110/110				

Scientific High School Diploma at the Liceo Scientifico Statale "L. 1995 Laurana" of Urbino with a vote of 60/60

PERSONAL SKILLS
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Mother tongue(s) Italian

cum laude

Supervisor: Prof. Vilberto Stocchi

Other language(s)	UNDERSTANDING		SPEAKING		WRITING
-	Listening	Reading	Spoken interaction	Spoken production	
English	B2	B2	B2	B2	B2
French	A2	A2	A2	A2	A2

Organization/managerial skills

Prof. Cellini is Head of the Physiology and Biochemistry Section of the Department of Medicine and Surgery of the University of Perugia. She is coordinator of numerous national and international projects.



Prof. Cellini manages a group made up of one university researcher, two post-doc fellow, one laboratory technicians, one PhD student, two post-graduate fellows.

# Participation in scientific committees of conferences:

Chair of the 15° International PH Workshop, Perugia June 23-24 2023

Organizer of the OxalEurope Workshop and First European Patient Meeting, Pozzuoli (NA), 8-9 giugno 2018

Chair of the Round Table on "Structural and molecular bases of pathologies", 61° SIB Meeting Virtual Edition

### Scientific activity:

Prof. Cellini's scientific activity mainly concerns the study of the molecular basis of diseases involving enzymes dependent on pyridoxal-5' phosphate and the development of new therapeutic approaches. Her research work concerns studies both on proteins in purified recombinant form and on cellular models. Prof. Cellini worked on cystalysin, a hemolytic toxin produced by Treponema denticola, the etiological agent of adult periodontitis, and on human Dopa decarboxylase, an enzyme involved both in Parkinson's disease, a neurodegenerative disease, and in "AADC deficiency", a neurometabolic syndrome. Since 2007, research activity has been mainly focused on the study of pathogenic variants of alanine:glyoxylate aminotransferase, a liver enzyme whose deficiency causes primary hyperoxaluria type I, and on the development of new therapeutic approaches. You currently also deal with mitochondrial ornithine aminotransferase, the deficiency of which causes gyrate atrophy of the choroid and retina. In recent years, Prof. Cellini has also been working on enzymes involved in the metabolism of sphingolipids as pharmacological targets in the context of fungal infections in patients suffering from cystic fibrosis.

Participation in editorial committees:

Associate editor of Frontiers in Molecular Biosciences Member of the Editorial Board of BBA-Proteins and Proteomics Associate editor of World Research Journal of Biotechnology Guest Editor of Journal of Biomedicine and Biotechnology-Special Issue on PLP-dependent Enzymes

Teaching and academic activities:

Teaching activities in the Biochemistry sector for the Degree Course in Medicine and Surgery and Degree Courses in the Health Professions. Details can be found at the following link: https://www.unipg.it/personale/barbara.cellini

Member of the Quality Assurance Committee of the University of Perugia

Tutoring activities

Member of the Teaching Board of the PhD course in Biosciences and then of that in Biomolecular Medicine of the University of Verona from the academic year 2006-2007 to 2017-2018. Since 2019, member of the Teaching Board of the PhD course in Systems Biology in immune and infectious pathologies, now PhD course in Inflammatoy and Infectious Pathologies, Therapeutical Strategies and Bio-Law

Supervisor of 6 three-year degree theses, 4 specialist degree theses and 7 master's degree theses. She is tutor and supervisor of 3 PhD students in Biosciences, 2 PhD students in Biomolecular Medicine, 1 student in Systems Biology in Immune and Infectious Pathologies, and 1 student in Inflammatoy and Infectious Pathologies, Therapeutical Strategies and Bio-Law

# Job related skills

Member of the Selection Committee for the achievement of the PhD title in:

-Biochemistry and Molecular Biology" 24th cycle a.y. 2011-2012 University of Parma,

- Pharmaceutical Sciences, Biomolecules and Health Products" 31st cycle a.y. 2017-2018 University of Parma
- -Escuela Internacional de Posgrado, Granada, Spain, 6 September 2019
- Biomolecular Medicine" 32nd cycle a.y. 2019-2020 of the University of Verona on 28 May 2020
- Molecular and Cellular Biology" 33rd cycle a.y. 2020-2021 of the University of Milan

- Systems Biology in Immune and Infectious Pathologies" - 33rd cycle a.a. 2020-2021 of the University of Perugia on 6 April 2021

- Life Sciences, Health and Biotechnology" - 34th cycle a.y. 2021-2022 of the University of Urbino on 15 December 2021



Driving licence	Replace with	driving licence	category/-ies.	Example:
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# ADDITIONAL INFORMATION

Publications Dr. Cellini's scientific production is documented by 96 publications in international journals, in which she appears in 47 cases as first author, last author or corresponding author. H-index = 25 (Scopus)

Complete list of publications at : https://pubmed.ncbi.nlm.nih.gov/?term=cellini+barbara&sort=date

Relevant publications of the last 5 years:

-Cellini B, Pampalone G, Camaioni E, Pariano M, Catalano F, Zelante T, Dindo M, Macchioni L, Di Veroli A, Galarini R, Paoletti F, Davidescu M, Stincardini C, Vascelli G, Bellet MM, Saba J, Giovagnoli S, Giardina G, Romani L, Costantini C. Dual species sphingosine-1-phosphate lyase inhibitors to combine antifungal and anti-inflammatory activities in cystic fibrosis: a feasibility study. Sci Rep. 2023 Dec 20;13(1):22692

-Ragavan VN, Nair PC, Jarzebska N, Angom RS, Ruta L, Bianconi E, Grottelli S, Tararova ND, Ryazanskiy D, Lentz SR, Tommasi S, Martens-Lobenhoffer J, Suzuki-Yamamoto T, Kimoto M, Rubets E, Chau S, Chen Y, Hu X, Bernhardt N, Spieth PM, Weiss N, Bornstein SR, Mukhopadhyay D, Bode-Böger SM, Maas R, Wang Y, Macchiarulo A, Mangoni AA, Cellini B, Rodionov RN. A multicentric consortium study demonstrates that dimethylarginine dimethylaminohydrolase 2 is not a dimethylarginine dimethylaminohydrolase. Nat Commun. 2023 Jun 9;14(1):3392. doi: 10.1038/s41467-023-38467-9

-Boffa I, Polishchuk E, De Stefano L, Dell'Aquila F, Nusco E, Marrocco E, Audano M, Pedretti S, Caterino M, Bellezza I, Ruoppolo M, Mitro N, Cellini B, Auricchio A, Brunetti-Pierri N. Liver-directed gene therapy for ornithine aminotransferase deficiency. EMBO Mol Med. 2023 Apr 11;15(4):e17033 -Gatticchi L, Dindo M, Pampalone G, Conter C, Cellini B, Takayama T. Biochemical and cellular effects of a novel missense mutation of the AGXT gene associated with Primary Hyperoxaluria Type 1. Biochem Biophys Res Commun. 2023 Feb 19;645:118-123

-Grottelli S, Annunziato G, Pampalone G, Pieroni M, Dindo M, Ferlenghi F, Costantino G, Cellini B. Identification of Human Alanine-Glyoxylate Aminotransferase Ligands as Pharmacological Chaperones for Variants Associated with Primary Hyperoxaluria Type 1. J Med Chem. 2022 Jul 28;65(14):9718-9734

-Gatticchi L, Grottelli S, Ambrosini G, Pampalone G, Gualtieri O, Dando I, Bellezza I, Cellini B. CRISPR/Cas9-mediated knock-out of AGXT1 in HepG2 cells as a new in vitro model of Primary Hyperoxaluria Type 1. Biochimie. 2022 Nov;202:110-122

-Dindo M, Pascarelli S, Chiasserini D, Grottelli S, Costantini C, Uechi GI, Giardina G, Laurino P, Cellini B. (2022) "Structural dynamics shape the fitness window of alanine:glyoxylate aminotransferase" Protein Sci 31(5):e4303

-F.L. van de Veerdonk, G. Renga, M. Pariano, M. M. Bellet, G. Servillo, F. Fallarino, A. De Luca, R. G. Iannitti, D. Piobbico, M. Gargaro, G. Manni, F. D'Onofrio, C. Stincardini, L. Sforna, M. Borghi, M. Castelli, S. Pieroni, V. Oikonomou, V.R. Villella, M. Puccetti, S. Giovagnoli, R. Galarini, C. Barola, L. Maiuri, M. A. Della Fazia, B. Cellini, V. N. Talesa, C. A. Dinarello, C. Costantini, and L. Romani (2022) "Anakinra restores cellular proteostasis by coupling mitochondrial redox balance to autophagy" J. Clin. Invest. doi 10.1172/JCl144983

-G. Pampalone, S. Grottelli, L. Gatticchi, E.M. Lombardi, I. Bellezza, B. Cellini (2021) "Role of misfolding in rare enzymatic deficits and use of pharmacological chaperones as therapeutic approach" Frontiers in Biosciences-Landmark Edition 26(12):1627-1642

-R. Montioli, G. Sgaravizzi, M.A. Desbats, S. Grottelli, C. Borri Voltattorni, L. Salviati, B. Cellini (2021) "Molecular and Cellular Studies Reveal Folding Defects of Human Ornithine Aminotransferase Variants Associated With Gyrate Atrophy of the Choroid and Retina" Frontiers in Molecular Biosciences 8:695205

-M. Dindo, G. Ambrosini, E. Oppici, AL. Pey, P.J. O'Toole, J.L. Marrison, I.E.G. Morrison, E. Butturini, S. Grottelli, C. Costantini, B. Cellini (2021) "Dimerization Drives Proper Folding of Human Alanine:Glyoxylate Aminotransferase But Is Dispensable for Peroxisomal Targeting" J. Pers. Med. 11(4):273

-R. Montioli, I. Bellezza, M.A. Desbats, C. Borri Voltattorni, L. Salviati, B. Cellini (2021) "Deficit of human ornithine aminotransferase in gyrate atrophy: molecular, cellular and clinical aspects" BBA Proteins and Proteomics, 1869, 140555

-B. Cellini, T. Zelante, M. Dindo, M.M. Bellet, G. Renga, L. Romani, C. Costantini (2020) "Pyridoxal 5'-Phosphate-Dependent Enzymes at the Crossroads of Host-Microbe Tryptophan Metabolism" Int J Mol



# Sci. 21(16): 5823

-M. Dindo, G. Mandrile, C. Conter, R. Montone, D. Giachino, A. Pelle, C. Costantini, B. Cellini (2020)
"The ILE56 mutation on different genetic backgrounds of alanine: Glyoxylate aminotransferase: Clinical features and biochemical characterization" Mol Genet Metab S1096-7192(20)30181-5
-M. Dindo, S. Grottelli, G. Annunziato, G. Giardina, M. Pieroni, G. Pampalone, A. Faccini, F. Cutruzzolà, P. Laurino, G. Costantino, B. Cellini (2019) "Cycloserine enantiomers are reversible

inhibitors of human alanine:glyoxylate aminotransferase: implications for primary hyperoxaluria type 1" Biochem. J.476, 3751-68

-C. Conter, E. Oppici, M. Dindo, L. Rossi, M. Magnani, B. Cellini "Biochemical properties and oxalatedegrading activity of oxalate decarboxylase from bacillus subtilis at neutral pH" IUBMB Life (2019) 71(7):917-927

-M. Dindo, E. Costanzi, M. Pieroni, C. Costantini, G. Annunziato, A. Bruno, N.P. Keller, L. Romani, T. Zelante, B. Cellini "Biochemical characterization of Aspergillus fumigatus AroH, a putative aromatic amino acid aminotransferase" Frontiers in Molecular Biosciences (2019) 5, 104.

-M. Dindo, C. Conter, E. Oppici, V. Ceccarelli, L. Marinucci, B. Cellini "Molecular basis of primary Hyperoxaluria: clues to innovative treatments" Urolithiasis (2019) 47(1):67-78

**Presentations** 

Presentations, Projects, Conferences, Seminars Honours and awards Memberships

"Molecular pathogenesis of PH" International Collaborative Network Kidney Stones Online Academy November 16th, 2023

• "A molecular look on AGT variants" Kidney Stone Research Day October 6th, 2023

•"Molecular pathogenesis of Primary Hyperoxaluria" International Symposium on Primary Hyperoxaluria" Bern (Switzerland), September 8, 2023

• "A biochemical perspective on the pathogenesis of gyrate atrophy "International Hybrid GACR Symposium", Amsterdam March 29<sup>th</sup>, 2023

•"Exploring the dual targeting of host and microbial sphingosine-1-phosphate lyaseas antimicrobial strategy in cystic fibrosis" XX Convention d'autunno dei ricercatori in fibrosi cistica, Verona, November 24-26, 2022

•"Latest from the Lab: How research unlocks the answers to a cure" Oxalosis & Hyperoxaluria Foundation Patient Meeting November 12 2022

• "Preclinical Research" International Hyperoxaluria Conference endosed by OxalEurope, Berlino September 29-October 1st 2022

• "Exploring the dual targeting of host and microbial sphingosine-1-phosphate lyase" XIX Convention d'autunno dei ricercatori in fibrosi cistica, Verona, November 25-26, 2021

•"Intrinsically disordered regions of alanine:glyoxylate aminotransferase shape its fitness and function" WebPro, Proteins on the Web, May 20-21, 2021

• "The role of Vitamin B6 in Primary Hyperoxaluria Type I" OxalEurope meeting, December 1st 2020, online meeting

• "Development of a small-molecule therapy for PH1 based on the combined administration of B6 vitamers and pharmacological chaperones" 13th OHF Hyperoxaluria Workshop", June 21-22, 2019, Boston (USA)

• "The dual coenzyme/chaperone role of pyridoxal 5'-phosphate: Primary hyperoxaluria as a paradigm" Proteins: Structure, Function and Biomedical Applications, Parma, March 29, 2019 • "Molecular basis of primary hyperoxaluria: clues to innovative

treatments" OxalEurope/OxalAfrica MEETING , December 6th-7th, 2018, Cairo, Egypt

• "Clues on Vitamin B6" Italian OxalEurope Workshop and First European Patient Meeting, Pozzuoli (NA), June 8-9 2018

• "Use Of Polymer Conjugates For The Intraperoxisomal Delivery Of Engineered Human Alanine:Glyoxylate Aminotransferase As A Protein Therapy For Primary Hyperoxaluria Type I"59" Congresso Nazionale SIB, Caserta, September 20-22 2017

• "Pharmacological chaperones rescue AGT folding defects in Primary Hyperoxaluria Type I" 12th International Primary Hyperoxaluria Workshop, Tenerife, July 14-16 2017

• "Use Of Polymer Conjugates For The Intraperoxisomal Delivery Of Engineered Human Alanine:Glyoxylate Aminotransferase As A Protein Therapy For Primary Hyperoxaluria Type I" OxalEurope Meeting, November 28, 2016, Amsterdam

• "Pathogenic mutations of alanine-glyoxylate aminotransferase" Cofactor-dependent proteins: evolution, chemical diversity and bioapplications, August 25-28, 2014, Parma, Italy

 "Molecular insights into the S187F variant of human liver alanine:glyoxylate aminotransferase associated with Primary Hyperoxaluria Type I: structural, biochemical and bioinformatic approaches" 57° Congresso Nazionale SIB, Ferrara, September 18-20 2013

• "Effects of polymorphic and pathogenic mutations on the structural and functional properties of human alanine:glyoxylate aminotransferase" Federation of European Biochemical Societies FEBS CONGRESS 2013 "Mechanisms in Biology" July 6th - 11th 2013, St. Petersburg, RUSSIA

• "Primary Hyperoxaluria Type I: identification of small molecules for an enzyme enhancement



therapy" Italian Forum on Industrial Biotechnology and Bioeconomy, Milano, October 23-24 2012 • "Impact of the mutations on alanine:glyoxylate aminotransferase variants associated with Primary Hyperoxaluria Type I: a biochemical approach" 10th International Primary Hyperoxaluria Workshop, Bonn, June 22-23 2012

• "Glycine 41 variants of alanine:glyoxylate aminotransferase: molecular analyses reveal the enzymatic defect leading to PH1" Ninth International Primary Hyperoxaluria Workshop, New York, August 28-29, 2010

• "Molecular defects of the glycine 41 variants of alanine:glyoxylate aminotransferase associated with Primary Hyperoxaluria Type I" P-21 Congresso Proteine 2010, Parma, 8-10 2010

• "Human alanine:glyoxylate aminotransferase in its wild-type and G82E pathogenic form: structurefunction relationship" P 08.11 52° Congresso Nazionale SIB, Riccione September 26-28 2007 • "The role of pyridoxal 5'-phosphate on the dimerization and folding processes of Treponema

denticola cystalysin:" P 13, Congresso Proteine 2006, Novara, June 1-3 2006

• "Mutation of Tyr 64 provides new insight on the cofactor binding and catalytic mechanism of Treponema denticola cystalysin" (S03M) International Interdisciplinary conference on Vitamins, Coenzymes, and Biofactors 2005 Awaji, Japan 6-11 November 2005

• "Spectroscopic and kinetic properties of the pyridoxal 5'-phosphate-dependent enzyme cystalysin from Treponema denticola" 15th National Meeting "A. Castellani" of PhD students in biochemical disciplines, Brallo di Pregola (PV) 11-14 June 2002

# Projects

•Holder of a grant from the "Ministry of University and Research" as part of the PRIN 2022 call for a project entitled "Oxalate metabolism and hyperoxaluria: from genetic predisposition and biochemical analyzes to the development of new treatment strategies" € 81097 (Oct2023 -Sep2025)

• Co-holder of a grant from the "Oxalosis and Hyperoxaluria Foundation" for a project entitled "Exploring the role of commensal bacterial and fungal communities and their metabolites in the handling of oxalate in primary and secondary hyperoxaluria" (May2022-Apr2024) € 196000 (Co-PI with Prof. Luigina Romani, University of Perugia)

• Holder of a grant from the "Cystic Fibrosis Research Foundation" for a project entitled "Exploring the dual targeting of host and microbial sphingosine-1-phosphate lyase as antimicrobial strategy in cystic fibrosis, FFC#19/2021" (Oct 2021-Sep 2022)

• Responsible owner of the Perugia Unit in a grant from the "Ministry of University and Research" as part of the FISR 2019 call for a project entitled "Cell-mediated enzymatic therapy for the treatment of gyrate atrophy", FISR2019\_04835, Coordinator Prof. Mauro Magnani (Dec 2020-Nov 2022),

• Holder of a grant from the "Cystic Fibrosis Research Foundation" for a project entitled "Exploring the dual-targeting of host and microbial sphingosine-1-phosphate lyase as antimicrobial strategy in cystic fibrosis, FFC#16/2020" (Sep 2020- Aug 2021),

• Co-holder of a grant from the "Oxalosis and Hyperoxaluria Foundation" for a project entitled "Understanding the pathomechanisms undelying PH3" (Apr2020-Mar2023) (Co-PI with Prof. Nicola Brunetti-Pierri, TIGEM Napoli)

• Holder of a grant from the "Oxalosis and Hyperoxaluria Foundation" for a project entitled "Development of a small-molecule therapy for Primary Hyperoxaluria Type I based on the combined administration of B6 vitamers and pharmacological chaperones" (Oct 2017-Sep 2019)

• Holder of funding from the Telethon Foundation for a project entitled "Comprehensive analysis of the molecular pathogenesis of gyrate atrophy towards the rationalization and the optimization of the therapy with vitamin B6" (Jan 2016- Dec 2018) (GGP 15114)

• Holder of ministerial funding within the SIR (Scientific Independence for Young Researchers) Projects for a project entitled "Erythrocytes as carriers of oxalate-degrading enzymes: an innovative approach for the treatment of hyperoxaluria" (RBSI148BK3) (September 2015- Aug 2018)

Holder of a university loan as part of the "Joint project 2012" for a project entitled "Biosynthetic activity of trophectoderm: new tools for embryo selection techniques in vitro" (Jan 2014-Dec 2015)
Holder of funding from the Inter-University Consortium for Biotechnology for a project entitled "Development of new therapeutic strategies for Dopa decarboxylase deficiency syndrome: a molecular approach" (Dec 2013-Nov 2014)

Holder of a grant from the "Oxalosis and Hyperoxaluria Foundation" for a project entitled "A combined biochemical and cell biology approach to improve the pharmacological treatment of Primary Hyperoxaluria type I: from pyridoxine therapy to proteostasis regulators" (Oct 2012-September 2014)
Holder of a university loan as part of the "Joint project 2010" for a project entitled "Type I Primary Hyperoxaluria: identification of small molecules to be used as "enzyme enhancement therapy" (Jan 2011 - Dec 2012)

• Holder of a grant from the "Oxalosis and Hyperoxaluria Foundation" for a project entitled "Combination of biophysical enzymology and molecular cell biology in the multidisciplinary study of primary hyperoxaluria type I (PH1)" (Jul 2010- Apr 2011)

• Holder of a university loan as part of the "Joint projects 2008" for a project entitled "A molecular



approach to the study of severe congenital neutropenia" (Jan 2009-Dec 2010)

# Seminars

. Cellini "Type I primary hyperoxaluria: from molecular mechanisms to the development of new therapeutic approaches" Department of Neurological Sciences, Biomedicine and Movement, University of Verona, 19 May 2020

• B. Cellini "Type I primary hyperoxaluria: from molecular mechanisms to the development of new therapeutic approaches" Department of Experimental Medicine, University of Perugia, 25 November 2016

• B. Cellini "A biochemical perspective on Primary Hyperoxaluria Type I: exploring new therapeutic

strategies from pharmacological chaperones to protein therapeutics", TIGEM, Naples, 29 June 2016 • B. Cellini "Molecular approaches to the study of primary Type I Hyperoxaluria and development of new therapeutic strategies", Department of Biomolecular Sciences, University of Urbino, 14 November 2013

• B. Cellini "Molecular approaches to the study of primary Type I Hyperoxaluria and development of new therapeutic strategies", Department of Clinical and Biological Sciences, University of Turin, 10 December 2012

• B. Cellini "Alanine:glyoxylate aminotransferase variants responsible for Type I Primary Hyperoxaluria: biochemical and bioinformatic analyses" PhD in Biochemistry, Molecular Biology and Biotechnology, University of Ferrara, 23 June 2010

# Membership

Appointed member of the "Scientific Advisory Board" of the "Oxalosis and Hyperoxaluria Foundation" from 2015 to 2019

Member of the Steering Committee of the OxalEurope consortium since 2017

Member of the "Scientific Advisory Coucil" of the "Oxalosis and Hyperoxaluria Foundation" from 2013 Member of the Italian Society of Biochemistry and Molecular Biology since 2005

Part of the "Kidney Health Initiative" of the American Society of Nephrology for the project entitled "Endpoints for Hyperoxaluria Clinical Trials"