



Prof. Annalisa Santucci

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Annalisa Santucci



Curriculum vitae 2013 and specific expertise on Alkaptonuria

PRESENT POSITION

Full Professor of Biochemistry (Professore I fascia di Biochimica), Dipartimento di Biotecnologie, Chimica e Farmacia, Università degli Studi di Siena

EDUCATION AND TRAINING

1984 Laurea in Scienze Biologiche (*magna cum laude*), Siena

1988-1989 University of Edimburgh, UK

1991-1992 University Hospital, Geneva, Switzerland

1994 PhD in Biotechnology, Siena

1997 Hewlett Packard, Palo Alto, CA

1998 Istituto Nazionale per la Ricerca sul Cancro, Genova

1998 Institut für Molekularbiologie und Biophysik, ETH, Zurich

PREVIOUS POSITIONS

1985-1986 FELLOWSHIP Università degli Studi di Siena, CRISMA, Centro Ricerche Sistemi Medici Avanzati

1987-1990 E.N.I. "Albatros" fellowship, Università degli Studi di Siena, Dipartimento di Chimica

1990 FIDIA Fellowship Università degli Studi di Siena, Dipartimento di Biologia Molecolare

1991-1994 PhD Student Biotechnology, Università degli Studi di Siena, Dipartimento di Biologia Molecolare

1995-1996 Fellowships (two) Istituto Superiore di Sanità, Roma

1997-1998 Fellowship Consorzio Siena Ricerche, Siena

1999-2001 Collaboratore Tecnico Laureato 7° livello, Università degli Studi di Siena, Dipartimento di Biologia Molecolare

2001-2002 Ricercatore BIO/10, Facoltà di Farmacia, Università degli Studi di Siena

2002-2004 Associate Professor of Biochemistry, Facoltà di Farmacia, Università degli Studi di Siena

2004-TODAY Full Professor of Biochemistry

OTHER POSITIONS

2003 – 2012 MEMBER OF THE DIRECTIVE COMMITTEE OF THE INTERDEPARTMENTAL CENTRE FOR THE STUDY OF GASTROENTERIC DISEASES ASSOCIATED TO SPYRALIFORM BACTERIA

2008 – 2009 PRESIDENT OF THE COURSE OF BIOTECHNOLOGY FOR HUMAN HEALTH

2009 – 2011 PRESIDENT OF THE COURSE OF PHARMACEUTICAL BIOTECHNOLOGY, FACULTY OF PHARMACY

2008 – 2011 DIRECTOR OF SIENA UNIVERSITY INTERDEPARTMENTAL CENTRE FOR THE BIOCHEMICAL STUDY OF OSTEOARTICULAR PATHOLOGIES

2011- TODAY PRESIDENT OF THE SCIENTIFIC COMMITTEE AND FOUNDER OF AIMAKU (ASSOCIAZIONE ITALINA MALATI DI ALCAPTONURIA)

2008 – TODAY MEMBER OF THE ADVISORY BOARD OF FINDAKURE INTERNATIONAL NETWORK



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SCIENTIFIC EXPERTISE

Protein structure-function relationships

Post-genomic biochemistry

Physiopathological mechanisms of human diseases (gastric cancer, pancreatic cancer, osteosarcoma, alkaptonuria, osteoarthritis)

Microrganism proteomics (*Helicobacter pylori*, *Saccharomyces cerevisiae*, *Neisseria meningitidis*)

PUBLICATIONS

A) Peer- reviewed international publications: 180

B) Patents: 6

C) Editor of books and special issues: 2

REVIEWER FOR INTERNATIONAL JOURNALS LIKE CLINICAL CHEMISTRY, PROTEOMICS, JOURNAL OF PROTEOME RESEARCH, PROTEOME SCIENCE, ELECTROPHORESIS, JOURNAL OF AGRICULTURAL AND FOOD CHEMISTRY, JOURNAL OF CHROMATOGRAPHY, EXPERT REVIEWS OF PROTEOMICS, ACTA PHARMACOLOGICA SINICA, JOURNAL OF DENTAL RESEARCH, BBA – PROTEINS AND PROTEOMICS, BBRC, FEBS LETTERS, ANTI-CANCER DRUGS, EXPERT OPINION ON MEDICAL DIAGNOSTICS, PROTEOMICS CLINICAL APPLICATIONS, AFRICAN HEALTH SCIENCES, JOURNAL OF MEDICAL MICROBIOLOGY, APPLIED MICROBIOLOGY, BMC BIOLOGY, JOURNAL OF PROTEOMICS, JOURNAL OF INHERITED METABOLIC DISEASES, MICROBIAL CELL FACTORIES, JOURNAL OF PHARMACY AND PHARMACOLOGY, BMC GASTROENTEROLOGY, JOURNAL OF METABOLOMICS AND SYSTEMS BIOLOGY, VACCINES, AFRICAN HEALTH SCIENCES, AMINO ACIDS, MICROBIOLOGICA, JOURNAL OF PROTEOME SCIENCE, PLOS ONE, AND SEVERAL OTHER.

REVIEWER OF PROPOSALS FOR:

- "RECHERCHE CLINIQUE TRANSLATIONNELLE 2009", INSERM (FRENCH INSTITUTE OF HEALTH AND MEDICAL RESEARCH) AND FRENCH MINISTRY OF HEALTH

- NATURAL SCIENCES AND ENGINEERING RESEARCH COUNCIL OF CANADA, DISCOVERY GRANT PROPOSAL REVIEWER

- AGENCE NATIONALE DE LA RECHERCHÉ, FRANCE

- MIUR, CINECA

- PROVINCIA AUTONOMA DI BOLZANO

ORGANIZER OF:

1988 – Scuola Nazionale di Scienza delle Proteine, SIB, Pontignano (SI), 22 agosto-2 settembre

1989 - IV Convegno Nazionale "Proteine '89", SIB, Siena, 14-16 settembre

1989 – COMETT Advanced Course on Protein Structural Aspects of Biotechnology, Edinburgh, UK, September

1991 – Corso Teorico Applicativo di Tecniche e Elettroforetiche, Fiorenzuola d'Arda (PC), 21-24 gennaio

1996 – XI Convegno Nazionale "Proteine '96", SIB, Siena, 29-31 maggio

1999 - Workshop CEE "The Immunobiotechnology Workshop on Antibody Technology", Siena, 25th April 1999

2005 – "Analisi Proteomica: dalla metodologia alle applicazioni", serie di seminari, simposi ed attività di orientamento, 4 maggio-21 giugno 2005, Firenze, Pisa, Padova, Roma, Napoli, Milano

2005 – Workshop ENEA "Saccharomyces cerevisiae: sistema modello per l'analisi delle risposte allo stress. Prospettive di ricerca e innovazione", Roma 16 dicembre 2005

2008 – Seldi-ToF Users' Meeting Bio-Rad, Siena 23-24 ottobre 2008

2009 – 3rd International Workshop on Alkaptonuria, Siena, 27th October 2009

2009 – Meeting of the International Advisory Board on AKU, Siena, 28th October 2009

2011 - 4th International Workshop on AKU, Cambridge, UK,

2011- 5th International Workshop on AKU, Liverpool, UK,

2013 - 6th International Workshop on AKU, Piestany, Slovakia,

2012 - Conferenza dei Presidi Regione Toscana – "Centralità della Toscana nella ricerca mondiale su una malattia ultra-rara: l'alcaptonuria", Firenze 3 ottobre

COORDINATOR OF SIENA UNIVERSITY INTERNATIONAL EDUCATIONAL AGREEMENTS:

UNIVERSITY OF LIVERPOOL, LIVERPOOL, UK

PROF. JAMES A. GALLAGHER

SCHOOL OF BIOMEDICAL SCIENCES



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DEPT. BIOMEDICAL SCIENCES
[HTTP://WWW.LIV.AC.UK/HACB](http://www.liv.ac.uk/HACB)

UNIVERSITY OF VIENNA, VIENNA, A
PROF. OSKAR HOFFMANN
INSTITUTE OF PHARMACOLOGY AND TOXICOLOGY
DEPT. OF PHARMACOLOGY AND TOXICOLOGY
[HTTP://WWW.UNIVIE.AC.AT/PTOX/INDEX.HTML](http://www.univie.ac.at/PTOX/INDEX.HTML)

THE SCRIPPS RESEARCH INSTITUTE, LA JOLLA, CA, USA
PROF. LUIGI PULVIRENTI
DEPT. OF NEUROPHARMACOLOGY
[HTTP://WWW.SCRIPPS.EDU/E_INDEX.HTML](http://www.scripps.edu/E_INDEX.HTML)

THE UNIVERSITY OF AUCKLAND, AUCKLAND, NEW ZEALAND
PROF. JILLIAN CORNISH
FACULTY OF MEDICINE AND HEALTH SCIENCES
SCHOOL OF MEDICINE
DEPT. OF MEDICINE
[HTTP://WWW.FMHS.AUCKLAND.AC.NZ/](http://www.fmhs.auckland.ac.nz/)

THE UNIVERSITY OF NEW SOUTH WALES, SYDNEY, AUSTRALIA
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SCHOOL OF BIOTECHNOLOGY AND BIOMOLECULAR SCIENCES
[MARC.WILKINS@UNSW.EDU.AU](mailto:marc.wilkins@unsw.edu.au)

JAGIELLONIAN UNIVERSITY, KRAKOW, POLAND
PROF. TADEUSZ LIBROWSKI
MEDICAL COLLEGE
FACULTY OF PHARMACY
[MFLIBROW@CYF-KR.EDU.PL](mailto:mflibrow@cyf-kr.edu.pl)

KILIS 7 ARALIK UNIVERSITY, KILIS, TURKEY
PROF. NAZIM ŞEKEROĞLU
ART AND SCIENCE FACULTY
BIOLOGY DEPARTMENT
[NSEKEROGLU@GMAIL.COM](mailto:nsekeroglu@gmail.com)

DOMAIN EXPERTISE ON ALKAPTONURIA (AKU)

AS is founder and President of the Scientific Committee of aimAKU (Associazione Italiana Malati di Alcaptonuria).

She is a collaborator with the following associations of AKU patients: AKU Society (UK), ALCAP (France), AKU Society North America (USA and Canada), DSAKU (Germany).

AS is member of the Advisory Board of international network FindAKUre (www.findakure.org), entirely devoted to the study of AKU and including also UK (Liverpool, Cambridge Universities), USA (NIH), Slovakia (Slovak Academy of Sciences), Jordan (Mutah University).

AS research group is at the moment the only team in the world studying AKU at the molecular level.

She has been organizer and member of the Scientific Committee of the following meetings on AKU:

- 3rd International Workshop on AKU, Siena 27 ottobre 2009
- 4th International Workshop on AKU, Cambridge, UK, 2011
- 5th International Workshop on AKU, Liverpool, UK, 2011
- 6th International Workshop on AKU, Piestany, Slovakia, 2013
- Conferenza dei Presidi Regione Toscana – “Centralità della Toscana nella ricerca mondiale su una malattia ultra-rara: l'alcaptonuria”, Firenze 3 ottobre 2013

AS has gained the following grants for AKU research:



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Toscana Life Sciences Foundation - TLS Orphan_01 - Studio proteomico e clinico della fisiopatologia dell'alcaptonuria ed allestimento di una terapia per il trattamento dell'ocronosi

Fondazione MPS 2009-2010 - Studio proteomico e clinico della fisiopatologia dell'alcaptonuria e creazione di un'associazione di pazienti

Fondazione MPS 2011- Identificazione e analisi preclinica di molecole terapeutiche per il trattamento dell'alcaptonuria

TELETHON – grant GGP10058 - Preclinical study of therapeutic agents for the treatment of ochronotic arthropathy

FP7-HEALTH-2012-INNOVATION-1 - Clinical Development of Nitisinone for Alkaptonuria

(Participants: Royal Liverpool University Hospital Trust UK; University of Liverpool, UK; National Institute of Rheumatic Diseases, Slovakia; Assistance Publique Hopitaux de Paris, F; PSR Group BV, NL; Cudos BV, NL; Nordic Bioscience A/S, Denmark; University of Siena, Italy; Alkaptonuria Society Ltd, UK; Swedish Orphan Biovitrum International AB, Sweden; Institute of Molecular Physiology and Genetics, Slovakia; ALCAP, F; Institut Necker, Paris, F).

She has been the Coordinator of 10 Erasmus international ex-changes of students and scientific personnel between Siena University and Liverpool, Cambridge (UK) and Nijmegen (NL) Universities for specific studies on AKU.

Publications on AKU

- [1] D. Braconi, G. Bernardini, C. Bianchini, M. Laschi, L. Millucci, L. Amato, L. Tinti, T. Serchi, F. Chellini, A. Spreafico, **A. Santucci**, Biochemical and proteomic characterization of alkaptonuric chondrocytes, *J Cell Physiol*, (2011) 227(9) (2012) 3333-3343.
- [2] D. Braconi, M. Laschi, L. Amato, G. Bernardini, L. Millucci, R. Marcolongo, G. Cavallo, A. Spreafico, **A. Santucci**, Evaluation of anti-oxidant treatments in an in vitro model of alkaptonuric ochronosis, *Rheumatology (Oxford)*, 49 (2010) 1975-1983.
- [3] D. Braconi, M. Laschi, A.M. Taylor, G. Bernardini, A. Spreafico, L. Tinti, J.A. Gallagher, **A. Santucci**, Proteomic and redox-proteomic evaluation of homogentisic acid and ascorbic acid effects on human articular chondrocytes, *J Cell Biochem*, 111 (2010) 922-932.
- [4] L. Tinti, A. Spreafico, D. Braconi, L. Millucci, G. Bernardini, F. Chellini, G. Cavallo, E. Selvi, M. Galeazzi, R. Marcolongo, J.A. Gallagher, **A. Santucci**, Evaluation of antioxidant drugs for the treatment of ochronotic alkaptonuria in an in vitro human cell model, *J Cell Physiol*, 225 (2010) 84-91.
- [5] L. Tinti, A.M. Taylor, **A. Santucci**, B. Wlodarski, P.J. Wilson, J.C. Jarvis, W.D. Fraser, J.S. Davidson, L.R. Ranganath, J.A. Gallagher, Development of an in vitro model to investigate joint ochronosis in alkaptonuria, *Rheumatology (Oxford)*, 50 (2011) 271-277.
- [6] L. Tinti, A. Spreafico, F. Chellini, M. Galeazzi, **A. Santucci**, A novel ex vivo organotypic culture model of alkaptonuria-ochronosis, *Clin Exp Rheumatol*, 29 (2011) 693-696.
- [7] Spreafico A, Millucci L, Ghezzi L, Geminiani M, Braconi D, Amato L, Chellini F, Frediani B, Moretti E, Collodel G, Bernardini G, **Santucci A**. Anti-oxidants inhibit SAA-amyloid formation and pro-inflammatory cytokines release in a human cell model of alkaptonuria, *Rheumatology (Oxford)*. 2013 May 23. [Epub ahead of print]
- [8] Millucci L, Spreafico A, Tinti L, Braconi D, Ghezzi L, Paccagnini E, Bernardini G, Amato L, Laschi M, Selvi E, Galeazzi M, Mannoni A, Benucci M, Lupetti P, Chellini F, Orlandini M, **Santucci A.**, Alkaptonuria is a novel human secondary amyloidogenic disease. *Biochim Biophys Acta*. 2012 Nov;1822(11):1682-91.
- [9] Laschi M, Tinti L, Braconi D, Millucci L, Ghezzi L, Amato L, Selvi E, Spreafico A, Bernardini G, **Santucci A**. Homogentisate 1,2 dioxygenase is expressed in human osteoarticular cells: implications in alkaptonuria. *J Cell Physiol*. 2012;227(9):3254-7.
- [10] Bernardini G, Braconi D, Spreafico A, **Santucci A**. Post-genomics of bone metabolic dysfunctions and neoplasias. *Proteomics*. 2012 Feb;12(4-5):708-21.
- [11] Braconi D, Bianchini C, Bernardini G, Laschi M, Millucci L, Spreafico A, **Santucci A**. Redox-proteomics of the effects of homogentisic acid in an in vitro human serum model of alkaptonuric ochronosis. *J Inherit Metab Dis*. 2011 Dec;34(6):1163-76.