

# **CRISTINA BOMBIERI**

## **CURRICULUM VITAE**

### **CURRENT POSITION**

Associate Professor of Applied Biology, Dpt of Neurological, Biomedical and Movement Sciences, University of Verona.

Coordinator of the Cystic Fibrosis and Respiratory Diseases research team.

### **PREVIOUS POSITIONS**

2005-2015: Assistant Professor at the University of Verona, Dpt. of Life and Reproduction Sciences; Verona (today: Dpt of Neurological, Biomedical and Movement Sciences), Italy.

1998-2004: Fellowship Owner at the University of Verona, Dpt. of Mother and Child and of Biology and Genetics (today: Dpt of Neurological, Biomedical and Movement Sciences), Verona, Italy.

### **EDUCATIONAL BACKGROUND**

- Degree on Biological Sciences, summa cum laude, University of Padova, Italy, 1992.
- PhD on Molecular and Cellular Biology and Pathology, University of Verona, Italy, 1998.

### **STAGE IN FOREIGN INSTITUTES**

- Stage in the Lab. of Molecular and Epidemiology Genetics, INSERM U613, University of Brest, France, directed by prof. Claude Ferec (9-19 march 2004): Analysis of large CFTR gene rearrangements by Quantitative Multiplex PCR of Short fluorescent Fragments
- Visiting Researcher in the Lab. of Molecular Genetics, INSERM U827, Institute of Clinical Research, Univ. Montpellier, France (october 2007 - september 2008), directed by prof. Mireille Claustris (Dr. M-C Romey's research group) to study CFTR gene expression regulation and functional characterization of promoter mutations.

### **RESEARCH ACTIVITY**

Human Molecular Genetic researches, involving also national and international collaborations with relevant European and American Research Groups, to study

- development of 3D cellular models to study the molecular basis of neurodegenerative, muscular and bone diseases
- mechanisms of post-transcriptional regulation of gene expression and role of ribonucleoproteins in the synthesis regulation of the TAU protein tissue specific RNA
- the genetic causes of Cystic Fibrosis and CFTR-Related Diseases and involvement of ENAC subunit genes in CF development and clinical expression, and for the identification of genetic factors involved in development and/or severity of some complex diseases and traits (bronchiectasis, rhinitis, COPD, asthma and occupational asthma, T2D, BMI and anthropometric traits).

Mainly used techniques include mutation analysis, high-throughput genotyping, Sanger and NGS sequencing, genome-wide and differential gene expression analyses, genotype-phenotype correlations, linkage and association analysis, candidate gene analysis, case-control studies, minigene transfection in cellular model to analyse gene expression and post-transcriptional regulation.

Person in charge for the preparation of the GEIRD biobank of phenotypically well-characterized subjects for the study of the role of genes and environment on respiratory diseases. Coordinator, prof. De Marco, University of Verona.

Laboratory jointly responsible for the preparation of a DNA-biobank including a population representative of Italian Veneto Region enrolled for the INCipe study (Project Coordinators: Prof. A. Lupo, University of Verona and Prof. G. Gambaro, Cattolica University of Rome), concerning chronic kidney disease.

## RECENT GRANTS

- Member of the thematic area “From Genomics to Function in rare diseases” in the “Project of Departmental Excellence (2023-2027): Cellular plasticity and degeneration from molecules to individuals- mechanisms, environment and therapy”.
- Grants for University Research, University of Verona (from 2005)
- Principal Investigator of the Local Research Unit dedicated to genetic analysis of the 3-years (January 2012 to December 2014) project titled "OXYDATIVE STRESS, AGEING, AND RESPIRATORY DISEASES. A multicentre epidemiological and genetic study on the general adult and elderly population in Italy". Project Coordinator, prof. De Marco, University of Verona; Italy. Granting Agency: Italian Ministry of Health, grant #RF-2009-1471235.
- Principal Investigator of a Local Research Unit of the 2-yrs project (september 2012 – september 2014): “Study of the pathogenetic and therapeutic role of the Epithelial Na<sup>+</sup> channel (ENaC) in CF and CF-like disease”. Italian CF Research Foundation (grant #FFC 3/12). Project Coordinator prof. Marco Lucarelli, Sapienza University Rome; Italy.
- Project coordinator of the 1-yr project (september 2010 – september 2011): “Molecular and functional study of the Epithelial Na<sup>+</sup> channel (ENaC) in CF and CF-like disease”. Italian CF Research Foundation (grant #FFC 1/10).

## INVITED PRESENTATIONS

- CFTR related diseases. 4th International Symposium for Cystic Fibrosis. Budapest, Hungary, 2-3 December, 2000.
- Genetic Tests in Cystic Fibrosis. Medical Genetics Specialization School, University Magna Graecia, Catanzaro, Italy. 24 February 2009.
- Recommendations for the classification of the diseases as CFTR-related disorders. European Cystic Fibrosis Society - Cystic Fibrosis Diagnostic Network Working Group Meeting. Verona, Italy, 10-12 February 2012
- Geni, malattie respiratorie e cardiovascolari. Meeting on COPD and cardiovascular system “Broncopneumopatia cronica ostruttiva: la complessa interazione con l’apparato cardiovascolare”. Verona, Italy, 13 December 2012
- Diagnosis of CF and related disorders in the older child and adult. Symposium 21 - Genetics and Diagnosis, 36th European Cystic Fibrosis Conference, Lisbon, Portugal, 12-15 June 2013.
- CFTR related diseases. CF course for early career medical and paramedical health care workers involved in CF care and/or CF research, 38th European Cystic Fibrosis Conference, Brussels, Belgium, 10-13 June 2015.
- Geni Modificatori in CF. Meeting of Italian CF Society. Bari, Italy, 11-14 June 2015.

## OTHER SCIENTIFIC ACTIVITIES

- Member of the Italian Society of Biology and Genetics (AIBG)
- Member of the Italian Society of Human Genetics (SIGU)
- From 2007 to 2008, member of the international study team on CFTR-related disease for the "FP6-funded EurocareCF project. CFTR Related Diseases Subgroup in Workpackage 3 - Coordination of Clinical Research" and organiser of the First International Workshop (Garda Lake, Verona, Italy, 22-23 Mars 2007).
- From 2002 to 2010 member of the Steering Committee at the Italian National Institute of Health for the Italian External Quality Control Program for Cystic Fibrosis Genetic Testing.

## PEER-REVIEWER

From 2001, Peer-reviewer for several scientific journals (American Journal of Respiratory Critical Care Medicine, Human Genetics, European Journal of Human Genetics, Journal of Cystic Fibrosis, Acta Pediatrica, Gene, Annals of human biology, Biotechniques, Genetic Testing and Molecular Biomarkers, Molecular Genetics and Metabolism, Journal of molecular diagnostics, etc).

## DIDACTICS AND TUTORIAL

- From 2010: Teacher of General Biology or Applied Biology in the Combined Bachelor's + Master's degrees in Pharmacy and in Dentistry and Dental Prosthetics, and in the Bachelor's Degrees in Physiotherapy, Osteopathy, Nurse, and Midwifery, at the University of Verona.
- From 2016: Teacher of Human Genetics and Fundamental of Medical Genetics, Medical Bioinformatics Master-Degree, University of Verona
- From 2010-2016: Teacher of Molecular Diagnosis of Genetic Diseases, Bioinformatics and Medical Biotechnologies Master-Degree, University of Verona
- From 2005: Assistant Professor in the Human Molecular Genetics course, Medicine and Surgery Degree, University of Verona
- From 2005: Member of the Teaching Staff of the PhD on Translational Biomedicine, in the PhD School on Translational Biomedicine, University of Verona
- From 2014: Member of the Teaching Staff of the PhD on Applied Life and Health Sciences, in the PhD School on Life and Health Sciences, University of Verona
- From 2009 to 2011: Teacher of General Biology, Bioinformatics Degree, University of Verona
- From 2006 to 2011: Teacher of "Molecular Diagnosis of Genetic Diseases" in the School of Specialization in Medical Genetics, University of Verona
- From 2009 to 2011: Teacher of "Population Genetics" in the School of Specialization in Medical Genetics, University of Verona
- Tutorial activity for Biological Sciences, Medicine and Surgery, and Biomedical Laboratory Assistant degrees.

## POSITIONS OF RESPONSIBILITY

- Member of the Faculty of Medicine and Surgery as representative of the Associate professors of the Dept. of Neurosciences, Biomedicine and Movement Sciences.
- President of the Student File Commission – Physiotherapy Bachelor's Degree
- Member of the Student File Commission for the Combined Bachelor's + Master's degrees in Medicine and Surgery and in Technological Medicine and Surgery
- Member of Teaching Commission in the Bachelor's degree in Osteopathy.
- Member of the Quality Insurance Commission in the Master's degree in Medical Bioinformatics
- Member of the FUR Commission for the grant distribution in the Department of Neurosciences, Biomedicine and Movement Sciences.
- Member of Fellowship commissions; University of Verona.
- Member of commissions for Bachelor's and Master's degrees in Medicine and Surgery, Bioinformatics, Medical Bioinformatics, Physiotherapy and for the PhD thesis discussion.
- Member of admission test commissions for Bachelor's and Master's degrees in Medicine and Surgery and in Health Professions, and in the PhD School.

## PUBLICATIONS AND BIBLIOMETRICS

Author of several articles published on international peer reviewed journals

Author of a contribution on Molecular Diagnosis of Genetic Diseases ("Diagnosi molecolare delle malattie genetiche" - "Genetica Medica e Umana") for the 3rd Update of the Italian Medical Encyclopaedia (UTET, Torino, Italy: col. 1447- col. 1465, 2007).