Università di Genova

Tiziana Bachetti

Contract Professor

EDUCATION AND TRAINING

2006 Award of Specialty in Applied Genetics, University of Pisa, Italy, (110/110 cum laude)
2004 PhD in Biology and Genetics, University of Genova, Italy
2001 Italian Board of Biologists Certification (96/100)
1999 Graduated in Biologic Sciences, University of Genova, Italy (110/110 cum laude)

PROFESSIONAL HISTORY

Present position	Since 2021 Dirigente Biologo at IRCCS Ospedale Policlinico San Martino UO Proteomics and Mass Spectrometry UO Quality Assurance Manager UO Worker Safety Management System
Past positions	2018- 2021: Researcher RTDA at the Neurodevelopmental Biology laboratory, University of Genova (from 2020: agreement as Dirigente Sanitario at IRCCS Gaslini) 2013-2018: Excellence Research contracts, Istituto Giannina Gaslini 2005-2012: Post-doc research contracts, Istituto Giannina Gaslini 2000-2012: Research contracts – Laboratorio di Genetica e Genomica delle Malattie Rare (past UO Genetica Medica), IRCCS Giannina Gaslini, Genova 1999-2000: Training at Lab Immunopathology, Advanced Biotechnology Center, Genova

ACADEMIC APPOINTMENTS

From 2022 to today: Contract Professor - Cell and Developmental Biology-6CFU for Master Degree in Applied and Experimental Biology, University of Genoa
2020: Doctoral program Biotechnology and bioinformatics in plants and animal research and their application in agriculture and farming for Doctorate in Sciences and Technologies for the Environment and the Territory (STAT)
2018-2021: RTDA, Cell and Developmental Biology-6CFU for Degree in Biological Sciences, University of Genoa
2018-2019 Biology II (Cytology, Histology and laboratory) - 4 CFU - for Degree in Biotechnology, University of Genoa
From 2012 to today: tutoring activity for MS students in Biological Sciences and Biotechnology and PhD programs students

EXPERIENCE

MAIN SEMINARS

2023 International Meeting Proteomics Association: Multi-omics analyses reveal altered lipid metabolism and oxidative stress as pathogenic mechanisms in a zebrafish model of Alexander disease. Rome, Nov 29th -1st Dec, 2023 (selected abstract)
2022: Role of GFAP mutations and gene expression in Alexander disease pathogenesis. Istituto Italiano Tecnologia (IIT), Genova
2022: MALDI Spettrometria di massa per immagini: dal tessuto ai pathways molecolari nella ricerca di base e traslazionale. NEUROBLASTOMA: DISCUSSIONI CLINICHE E DI RICERCA ANNO 2022 (invited)
2022 Il gene GFAP: ruolo delle mutazioni e dell' espressione nella patogenesi della malattia di Alexander. 1° Meeting Nazionale sulle Leucodistrofie. Milano, 19-20 settembre 2022
2022 Studio dell'interattoma di PHOX2B. 16° meeting delle famiglie AISICC, 10 aprile 2022, Firenze
2019 Trascrittomica & proteomica per lo studio delle mutazioni NPARMs nelle forme di CCHS sindromica; 15° meeting delle famiglie AISICC, Pratolino (Fi), Italy
2018 Genetic investigation in SIDS. Corso SIDS-ALTE: multidisciplinarietà e sinergie, Istituto Gaslini (invited)
2016 Novel technologies and their applications, CISEF, Gaslini
2014 Update on research and diagnosis activity, 10° meeting AISICC, Pratolino (Fi), Italy
2014 TNF Receptor Associated Periodic Syndrome (TRAPS) as a model linking autophagy and inflammation in protein aggregate diseases. Seminario presso Università degli Studi di Genova, Genova, Italy (invited)
2013 Defective autophagy in TNFR associated periodic syndrome as a player in inflammation in TRAPS. Autoinflammation 2013-ISSAID 2013, Losanna, Switzerland (selected abstract)
2012 Novelty in PHOX2B molecular diagnosis improves detection of congenital central hypoventilation syndrome transmission. Fourth International meeting on Congenital Central Hypoventilation Syndrome (CCHS), Varsavia, Poland.
2012 Defective autophagy in TNFR associated periodic syndrome accounts for tnfr accumulation and enhanced inflammation, Translational and Clinical Issues in Paediatric Rheumatology, PReS Research Course, Genoa, Italy. (selected abstract)
2010 Polyalanine expansions in Congenital Central Hypoventilation Syndrome (CCHS): genetic aspects and possible therapeutic approaches. Società Italiana di Genetica Umana (SIGU), Firenze, Italia (selected abstract)
2009 Heat shock response and misfolded protein elimination associated with PHOX2B polyalanine expansion and GFAP mutations in Congenital Central Hypoventilation Syndrome and Alexander disease. IV Meeting on Molecular Mechanisms of Neurodegeneration, Milano, Italy. (selected abstract)
2008 Characterisation of a SNP in GFAP promoter associated with different allelic transcriptional activity in patients affected with Alexander disease. Società Italiana di Genetica Umana (SIGU), Genova, Italia (selected abstract)
2007 Pathogenetic mechanisms underlying the effects of PHOX2B polyalanine expansions. Third International meeting on Congenital Central Hypoventilation Syndrome (CCHS). Sestri Levante, Italy

SCIENTIFIC RESPONSIBILITY FOR RESEARCH PROJECTS ACCEPTED FOR FUNDING ON THE BASIS OF COMPETITIVE CALLS INVOLVING PEER REVIEW

2025-2026 Scientific Agreement – Agencia Estatal Consejo Superior De Investigaciones Científicas
2025-2027 Ricerca Corrente Ministero della Salute (PI)

2022-2024 Ricerca Corrente Ministero della Salute (PI)
Collaborator in “5x1000” project (PI of the Unit: dr. Paola Monti, Mutagenesis, PI dr Ulrich Pfeffer, IRCCS Osp. Policlinico San Martino)
2018: Fondazione Umberto VeronesiPostdoctoral Fellowships
Project: Combined approaches to identify molecules effective in rescuing gene expression alteration induced by PHOX2B mutations in neuroblastoma
2017: Associazione Più Unici che Rari Onlus “Modello di zebrafish per la malattia di Alexander: nuovo sistema per studiare la patogenesi delle mutazioni nel gene GFAP e per identificare farmaci potenzialmente efficaci nel contrastare l’accumulo della proteina GFAP mutata”
2016: Minigrant Istituto Giannina Gaslini
Project “Development of novel diagnostic methods with NGS technology”
2015: Excellence Research Contract Istituto Giannina Gaslini
Titolo del progetto “Search of genetic factors of susceptibility for the Hischsprung disease associated enterocolitis (HAEC)”
2013-2015:Excellence Research Contract Istituto Giannina Gaslini
Project “high throughput drugs screening to identify drugs able to ounteract PHOX2B overexpression in neuroblastoma”
2013: Research Fellow Fondazione Umberto Veronesi
Project: “High throughput drugs screening to identify PHOX2B posttranscriptional elements as novel therapeutic targets in neuroblastoma”
2012: Associazione Italia per la Ricerca sul Cancro My First AIRC Grant
Project: “A therapeutic approach for neuroblastoma based on reducing PHOX2B overexpression and pathogenetic interactions”.
2009: ELA Foundation for Leukodystrophies (France)Postdoc fellowship
Project: “Effect of curcumin on mutant GFAP aggregation associated with Alexander disease”.

EDITORSHIP OR PARTICIPATION IN EDITORIAL BOARDS OF JOURNALS, PUBLISHING SERIES, ENCYCLOPAEDIAS AND TREATISES

1. **Bachetti T**. and Ceccherini I. In vitro studies of PHOX2B gene mutations in congenital central hypoventilation syndrome. Chapter 6 of the book: “Genetics of respiratory control disorders”. Gaultier Ed., Springer Science, 2008.
2. **Bachetti T**, Ceccherini I . PHOX2B (pairedlike homeobox 2b). Atlas Genet Cytogenet Oncol Haematol. April 2013.

PRIZES AND ACCOLADES FOR SCIENTIFIC ACTIVITY, INCLUDING MEMBERSHIP OF ACADEMIES

2010: AWARD Department Ligure Of Genetics & Lion Club For Genetics Best Publications Yrs 20082010
Member of European Association Human Genetics

OTHER EXPERIENCES

RESEARCH INTERESTS AND SKILLS: long lasting experience in molecular genetics and cell biology to study the pathogenetic molecular mechanisms of neurodegenerative and neurodevelopmental diseases.

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In addition, computational biology skills (PPI netwroks, Gene Ontology analyses, in silico drug repurposing) and experience in cell-based luciferase-reporter platforms for high throughput drug screening targeting gene expression regulation.

22.07.25