

Paola Riva CV

Dipartimento di Biotecnologie Mediche e Medicina Traslazionale – Università degli studi di Milano

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EDUCATION AND TRAINING

1987 – Degree in Biological Sciences - Università degli Studi di Milano.

1988 – Attends the 1st Course of European School of Medical Genetics, Sestri Levante (Ge)

1992 - PhD in Molecular and Cellular Biology - Università degli Studi di Milano, Roma

1992 – Attends the EMBO practical Course "Cloning into YAC vectors 2", Napoli

1993-1994 – Post-Doctoral Fellow (Università degli Studi di Milano).

1997 - Diploma in Medical Genetics following four-year attendance to the Post Lauream School in Medical Genetics - Università degli Studi di Milano.

EMPLOYMENT AND EXPERIENCE

2022 to date – Coordinator of the Advanced Course: The cytogenetic lab applied to diagnostics: from theory to practice-Università degli Studi di Milano

2022 to date - Member of the directory board of Associazione Italiana Biologia e Genetica

2019 to date - Professor of the PhD School in Molecular Genetics, Biotechnology and Experimental Medicine – Università degli Studi di Brescia

2015 - Full Professor in Experimental Biology (BIO/13 group) - Università degli Studi di Milano

2015 -2018 - Professor of the PhD School in Experimental Medicine and Medical Biotechnology

2014 to date – member of the Independent Ethics Committee of Derming srl

2013 –2015 - Professor of the PhD School in Molecular and Cellular Biology and member of the directors' board – Università degli Studi di Milano

2012 to date – Director of the Laboratory “Service for study and diagnosis of genomic anomalies” – Università degli Studi di Milano, certified as “SmEL” by Regione Lombardia

2012 -2018 - Professor of the PhD/MD School in Biomedical Research associated to the single cycle degree course of Medicine – Università degli Studi di Milano

2009 -2014 - Professor of the PhD School in Molecular and Biological Sciences and member of the directors' board – Università degli Studi di Milano

May 2008 – April 2012 – Director of the Department of Biology and Genetics for Medical Sciences – Università degli Studi di Milano

2007 to date - Affiliation to the Stem Cells Research Center “UniStem”

2004–2007 – Professor of the International PhD program – EMBL – Heidelberg associated with the PhD course in “Genetic and Biomolecular Sciences” - Università degli Studi di Milano

2003–2009- Professor of the PhD Course “Molecular and Cellular Biology” - Università degli Studi di Milano”

2001 –2015 Associate Professor in Experimental Biology (BIO/13 group) - Università degli Studi di Milano

1999–2003 – Professor of the PhD Course “Neurology, Psychiatry and Neurogenetics” - Università degli Studi di Milano e Università di Genova associated.

1998 to date Professor in Molecular Genetics of the Post Lauream Specialization School in Medical Genetics - Università degli Studi di Milano

1995 – 2001 - University Researcher (E13X), Università degli Studi di Milano.

FELLOWSHIPS

1988-1992 PhD Fellow in Molecular and Cellular Biology - University of Milan

1993–1994 Post Doctoral Fellow - University of Milan

1994-1997 Post Lauream School in Medical Genetics Fellow - University of Milan

TECHNOLOGY TRANSFER**PATENTS**

1998 – Inventor with Dr. L. Corrado of “A simple method to generate Locus-Specific probes for FISH analysis”. Patent n°. 0001302260, Milano 24/09/98

2005 – Inventor of a “Scientific didactic Instrument – Simulation of in Situ Hybridization”. Patent n°. 0001365451, Milano 20/04/05

CREATION OF NEW COMPANIES

2012 - Partner of a technological district coordinated by the “Consorzio Italbiotec” - National cluster according to the DGR IX/2892/2011 IX/3084/2012 in the field Biotechnology – Green Chemistry.

2013 – Partner of a Temporary Network of Company providing the Istituto Nazionale dei Tumori (Milano) a certified DNA sequencing diagnostic service.

2015 – 2016 Partner of a Temporary Network of Company providing the Hospital of Crema a certified oncological FISH diagnostic service.

2016 – 2018 Coordinator of a Temporary Network of Company providing the Hospital of Crema the certified DNA sequencing and oncological FISH diagnostic services.

INSTITUTIONAL ACTIVITY – University of Milan

2017 to date Member of the School of Exercise and Sport Sciences programming board

2014 – 2020 President of the Joint Committee consisting of students and teachers of the MSc Medical Biotechnology and Molecular Medicine, MSc Medicine and BSc Medical Biotechnology

2012 - January 2018 Responsible of the Biology and Genetics section of the Department of Medical Biotechnology and Translational Medicine

2012-2015 Member of the Academic Disciplinary Board

2010 – 2012 Member of the Medical Faculty programming board

2008 – 2012 Director of the Department of Biology and Genetics for Medical

2006-2012 member of the Academic Scientific Research and Technological Transfer board for the Biological Area

2004 - 2012 member of the Academic Committee for the Evaluation of Teaching

YOUNG RESEARCHERS' FORMATION

Tutor of 8 PhD students, 5 post lauream Medical Genetics School students, and supervisor of 7 post-doctoral fellows

BIBLIOMETRIC INDICATORS

Scopus Author ID: 35552872000

Scopus Metrics (03-07-2023):

70 documents

1782 citations by 1521 Documents

22 h-index

SCIENTIFIC ACTIVITY

TECHNOLOGICAL EXPERTISE

The research activity of P. Riva Research group deals with the human molecular genetics by applying molecular genetic approaches to genome structure studies, DNA, RNA and protein analysis in both genetic diseases and cancer. The following skills were acquired and applied in different research fields:

- Cell culture techniques and Somatic cell genetics by means of conventional and high resolution FISH on metaphase, nuclei, stretched chromosomes and DNA fiber
- Molecular genetics: Sanger sequencing and NGS, DNA analysis by Southern blotting and Pulse Field Gel electrophoresis hybridization, PCR analysis, cloning of genomic and cDNA, mutation screening techniques (SSCP, Heteroduplex analysis, Protein truncation test), DHPLC, direct sequencing; RNA analysis by Northern blotting, dot blotting, RT-PCR and RealTime-PCR; protein analysis by Western blotting, immunoprecipitation.
- Recombinant DNA: cloning of genomic DNA, and cDNA in plasmidic vectors, cloning of cis regulatory elements in reporter gene vector, generation of FISH locus-specific probes by single

strand PCR (patented procedure), construction of YAC, PAC and BAC contiguos, cDNA selection applied to candidate-positional gene approach

- Functional genetics: functional studies of post-transcriptional regulatory elements by luciferase assays; transfection of siRNAs/miRNAs and their biological validation; down-regulation/overexpression of specific genes in danio rerio (zebrafish) animal model, by means of morpholino/expression-constructs injection.

Bioinformatic analysis of DNA, mRNA and protein.

MAIN RESEARCH FIELDS

GENETICS OF HUMAN DISEASES

- RASopathies: identification of new genetic and epigenetic mechanisms:

a) Noonan Syndrome: identification of new pathogenetic genes and variants of modifier genes

b) Neurofibromatosis type 1 (NF1): genotype-phenotype correlation in NF1, including spinal NF1 and NF1 microdeletion syndrome

- Genotype-phenotype correlation in neurofibromatosis type 2

CANCER GENETICS

- Genetic characterization of solid tumors: leiomyosarcoma and chordoma

- Identification of specific genetic lesions and gene expression profiles targeting dermal and/or plexiform neurofibromas

- Identification of pathogenetic mutations in familial breast cancer, Lynch Syndrome and familial adenomatous polyposis

EDITORIAL ADVISORY BOARD

Member of the Editorial Panel of EC Neurology

Member of the Editorial Panel IJ Molecular Science

Member of the Editorial Panel IJ Pediatrics and Child Health

Member of the editorial Panel Frontiers in Genetics

AD HOC REVIEWER OF SEVERAL INTERNATIONAL SCIENTIFIC JOURNALS

CONCERNING Human Genetics, Cancer Genetics, Molecular Genetics

RESPONSIBLE OF THE FOLLOWING GRANTED PROJECTS

1997-1999 Telethon Grant, Project n. E780 "COMPARATIVE GENOTYPE-PHENOTYPE STUDY IN DELETED NF1 PATIENTS" Co-PIs Paola RIVA, Lidia LARIZZA (80.000.000 Lit)

2008 PRIN Grant 2007 - prot. 2007MWCEAL_003 "REGOLAZIONE POST-TRASCRIZIONALE DEL GENE CDK5R1 E POSSIBILI IMPLICAZIONI PATOGENETICHE NEL RITARDO MENTALE E IN MALATTIE NEURODEGENERATIVE" OU PI Paola RIVA (43.470 Eu), Coordinator Angelo NICOLIN

2010-2013 AIRC Grant (IG 10525) "STUDY OF CHORDOMA TUMORIGENESIS ADDRESSING PROGNOSTIC MARKERS AND PHARMACOLOGICAL TARGETS IDENTIFICATION" PI Paola RIVA (120.000 Eu)

2012 -2014 Italian Ministry of Health Grant, Project N. RF-INN-2008-1204836 –Rare Diseases "IMATINIB FOR THE TREATMENT OF PLEXIFORM NEUROFIBROMAS IN NF1 PATIENTS" – OU PI Paola RIVA, Coordinator Marica EOLI (250.000 Eu)

2014-2015 Istituto Nazionale dei Tumori, Milano - Research contract after competition "IDENTIFICATION OF MUTATIONS IN GENES INVOLVED IN FAMILIAL TUMOR OF BREAST AND COLON", PI Paola RIVA (770.120,00 Eu)

2018 -2021 Italian Ministry of Health Grant, Project N. RF-2016-02361293 – "UNSOLVED CHALLENGES IN NEUROFIBROMATOSIS TYPE 1: THE SEARCH FOR NOVEL CLINICAL AND MOLECULAR PREDICTORS FOR SPINAL NEUROFIBROMATOSIS DIAGNOSIS AND MANAGEMENT" OU PI Paola RIVA, Coordinator Marica EOLI (427.032,375 Eu)

2023 PRIN – 2022 prot. 2022BF89JZ – “EMERGING ROLE OF DIGENIC OR PSEUDO-DIGENIC INHERITANCE MODELS IN MENDELIAN DISEASES: IMPLICATION OF HYPOMORPHIC VARIANTS IN NOONAN SYNDROME’S PATHOGENETIC MECHANISMS AND IN RAS PATHWAY DEREGULATION”, Coordinator Paola RIVA (252.586 eu)

REVIEWER OF NATIONAL AND INTERNATIONAL PROJECTS

- 2002 Austrian Science Fund, US National Science Fund/Research Councils UK
- 2009 e 2010 Academic grants, Seconda Università di Napoli - Italy
- 2010 AIDS, Ministero della Salute - Italy
- 2013 FIRB, MIUR - Italy
- 2014 SIR, MIUR - Italy
- 2014 The Research Foundation - Flanders (FWO) – Belgium
- 2015 SIR, MIUR - Italy
- 2016 Cooperation, Israeli Ministry of Science Technology and Space, Italian Ministry of Foreign Affairs
- 2020 Fundació La Marató TV3 – Spain
- 2022 Research Council KU Leuven - Belgium

SCIENTIFIC SOCIETIES

- Member of the directory board of Associazione Italiana Biologia e Genetica
- European Society of Human Genetics
- Società Italiana di Genetica Umana
- Orphan Disease Center

CURRENT TEACHING ACTIVITY – UNIVERSITY OF MILAN

MSc Medical Biotechnology and Molecular Medicine – course of Genetic and Molecular basis of Diseases

MSc Medicine – course of Biology and Genetics

BSc Exercise, Sport and Health Sciences – course of Biology

BSc Medical Biotechnology – course of Genetics

DIAGNOSTIC ACTIVITY

Responsible of the molecular-genetic lab providing a certified service for the Italian National Health System

