# SHORT CV Prof. Lidia Larizza



Affiliation:

IRCSS ISTITUTO AUXOLOGICO ITALIANO, Milano, Italy, www.auxologico.it

Phone. +39 02619113041 Fax +39 0 619112464 e-mail: l.larizza@auxologico.it

## Education

1968 – Pavia University – **Medical Doctor** degree cum laude

1973 - Stanford University CA - Research Fellow at Dept. of Genetics (Prof. L.L. Cavalli Sforza)

1982-1983- Heidelberg, Germany- Guest Scientist at DKFZ, Institute of Immunology and Genetics (Prof. V. Schirrmacher)

1990 – Stanford University CA- Visiting Scientist at Dept. of Genetics (Prof. L.L. Cavalli Sforza)

#### **Present**

Research Director of Laboratory of Medical Cytogenetics and Human Molecular Genetics, Istituto Auxologico Italiano, Milan (since 1993), honorary member of ECA (European Cytogeneticists Association), honorary member of SIGU (Società Italiana Genetica Umana) and member of SIGU "probiviri" committee, Management Committee Member of BMBS COST Action BM1208 "European Network for Human Congenital Imprinting Disorders", Member of the editorial board of "Cancer Letters" and other journals in the field of Human Genetics and Genomics. Italian partner of "CHROMISYN" Project ERA-NET-NEURON JTC 815-035. Member of American Society Human Genetics (ASHG), European Society Human Genetics (ESHG), Human Genome Organization (HUGO). Author of 241 publications in PubMed (<a href="http://www.ncbi.nlm.nih.gov/pubmed/?term=larizza+1">http://www.ncbi.nlm.nih.gov/pubmed/?term=larizza+1</a>) and 17 monographies.

## **Main Research Interests**

Rare disorders of chromatin regulators: Cohesinopathies: exploring clinical and genetic heterogeneity

Rubinstein-Taybi and related syndromes of the epigenetic machinery: generation of iPSC-derived neurons from patients with different level of intellectual disability to disclose syndrome- and patient-specific biomarkers for therapeutics

Chromosomal/genomic instability syndromes with Cancer Predisposition: Rothmund-Thomson and RECQL4 syndromes and Poikiloderma with Neutropenia. Zebrafish modelling of Pokiloderma with Neutropenia.

Imprinting Disorders affecting growth: Beckwith-Wiedemann, Silver Russell and Hemihyperplasia. Pathomechanisms of Multilocus Methylation Disturbances (MLID).

Neurodevelopmental Imprinting Disorders: Angelman and Prader-Willi syndromes. iPSC modelling of imprinting disorders associated with the chromosome 15q11-q13 region

 $\label{lem:condition} Genomic \ disorders: \ NAHR \ (Non \ Allelic \ Homologous \ Recombination) \ and \ NHEJ \ (Non \ Homologous \ End \ Joining) \ - \ mediated \ Microdeletions/Microduplication \ syndromes. \ NF1 \ microdeletion \ syndromes.$ 

The MARK4 gene: alternative splicing and role of the MARK4L isoform in gliomagenesis

The c-kit gene: loss of function mutations and Piebald Trait; gain of function mutations and Core Binding Factor Leukemia and Inherited Gastrointestinal stromal tumors

## **Previous Positions and experience**

Full Professor of MedicalGenetics, University of Milan (1986-2014), formerly Associate Professor of General Biology (1971-1986),
University of Milan

Founder and Director (1989-2001; 2004-2010) of the Post-Lauream School in Medical Genetics, University of Milan

Member of Delegation "People to People" of United Nations to China and Mongolia (October 1993)

President of AICM (Associazione Italiana di Citogenetica Medica) (1995-1997)

Chair of the Fourth European Cytogenetics Conference, Bologna 6-9 September 2003

Faculty member of European Advanced Postgraduate Course in Classical & Molecular Cytogenetics, Montpellier/Nimes, France (1998-2010)

Chairperson of COST ACTION B19 on "Molecular Cytogenetics of Solid tumors" http://www.costb19.net (2000-2006).

President of European Cytogeneticists Association (ECA) (2006-2009)

President of Società Italiana di Genetica Umana (SIGU) (2009-2011)

Member of the scientific Committee of 8th, 9th, 19th, 11th and 12th European Workshops on Cytogenetics & Molecular Cytogenetics of Human Solid Tumors, Barcelona 12-15 Sept 2002, Brno 16-19 Sept 2004, Montpellier 8-11 June 2006, Bilbao 6-9 Sept 2008, Nijmegen 3-6 June 2010.

Member of ESHG (European Society of Human Genetics) Programme Committee Conference, Milan 29 May-3 June 2014

Elected member of the Committee for Research and Technological Transfer and President of the Committee for Medical Sciences of the University of Milan: http://unimi.it/ (2003-2012)

Faculty Member of the PhD course "Neurology, Neurogenetics and Psychiatry" held jointly by the University of Genoa and the University of Milan (1998-2007)

Faculty Member of the PhD Course "Pathology and Experimental Neuropathology", University of Milan (2007-2014)

Member of the steering committee of the Doctorate School "Biomedical Clinical and Experimental Sciences", University of Milan (2007-2012)

Member of the Committee of the Dept of Medicine, Surgery & Dentistry (DMCO), University of Milan (2007-2012)

Member of the Committee of Dept of Health Sciences (DISS- formerly DMCO), University of Milan (2012-2014)

Chair of the "Scientific Committee" of DISS, University of Milan (2012-2014)

Member of scientific board (SAB) of Nina Foundation & Angelman Syndrome Alliance (<a href="http://www.ninafoundation.eu">http://www.ninafoundation.eu</a>) (2014-2016)

Member of Evaluation Committee "CE12 Genetics, Genomics, Gene expression, Regulator RNAs" ANR (Agence Recherche, France, <a href="http://www.agence-nationale-recherche.fr">http://www.agence-nationale-recherche.fr</a> ) (2015-2016)

Numerous other minor positions are omitted