

Curriculum Vitae et Studiorum– Francesca Boaretto

PERSONAL DATA

BORN Venezia, 26 August 1974
NATIONALITY Italian
PHONE Office +39-0498215503
EMAIL: francesca.boaretto@iov.veneto.it
MARITAL STATUS: Married, one daughter
POSITION PhD Biologist, Genetic Counselor at the Familial Cancer Clinic and Oncoendocrinology (Veneto Institute of Oncology, Padova)

EDUCATION

2008 PhD in Genetics and Molecular Biology of the Development. Dissertation thesis on: “Identification and characterization of inherited Spastic Paraplegia genes”. Supervisor Prof. ML Mostacciolo. University of Padova, Italy.
2004 Specialist in Medical Genetics. Dissertation thesis on: “Molecular characterization of Schizophrenia affected families originating from Chioggia (Italy)”. Supervisor Prof. A. Amoroso. 150/150 *cum laude*, University of Trieste, Italy.
2001 National licensing examination for biologist. 136/150, University of Padova, Italy.
2000-2001 Post-lauream training in Human Genetics Laboratory at the University of Padova, Italy. Supervisor Prof. ML Mostacciolo.
1999 Doctorate degree in Biological Sciences. Dissertation thesis on: Linkage analysis in a family diagnosed with a complicated form of Spastic Paraplegia. Supervisor Prof. ML Mostacciolo. 104/110, University of Padova, Italy.

GRANTS, CONTRACTS AND BURSARIES

2016-up to now Genetic Counselor activities at the Familial Cancer Clinic and Oncoendocrinology (Veneto Institute of Oncology, Padova). Head: Dr. S. Zovato.
2006-2015 Genetic Counselor activities first at the clinic of Endocrinology (Hospital of Padova) and then at the SSD Hereditary Tumors Unit (Veneto Institute of Oncology, Padova). Head: Prof. G. Opocher.
2016 One year grant for young scientists from the University of Padova (Grant Research Fellowship) on the project: Identification of

- genetic determinants of inherited diseases by NGS. Supervisor Dr. G Vazza.
- 2015 One year grant for young scientists from the University of Padova (Grant Research Fellowship) on the project: Use of NGS systems for molecular diagnosis of hereditary diseases characterized by high genetic heterogeneity such as motor neuropathies (HMSN , HMN) and primary ciliary dyskinesia (PCD). Supervisor Dr. G. Vazza.
- 2013 Two years grant for young scientists from the University of Padova (Senior Research Fellowship) on the project: A novel methodological approach for systematic molecular diagnosis of patients with axonal-mixed CMTs/dHMNs and identification of novel disease causing genes. Supervisor Prof. ML Mostacciuolo.
- 2012 Two years grant for young scientists from the University of Padova (Junior Research Fellowship) on the project: Analisi dei meccanismi patogenetici responsabili di patologie lisosomiali attraverso l'impiego di zebrafish biosensori. Supervisor Dr. E. Moro.
- 2011 Winner of Public selection, University of Padova. Project: Systematic molecular diagnosis of patients with CMTs, SLA, SMA, dHMNs, distal dystrophy and FSP identification of novel disease causing genes. Supervisor Prof. ML Mostacciuolo.
- 2009-2010 Winner of Public selection for 12 months contract with the IRCSS– IOV, Padova working on molecular and genetics aspects of hereditary endocrine and neuroendocrine tumors: i) genotype-phenotype correlation, molecular diagnosis and characterization of mutations causing MEN1, MEN2, PHEO/PGL, VHL, PPNAD and Carney Complex diseases; ii) Evaluation of the penetrance and expressivity of the pheochromocytoma/paraganglioma syndrome in a geographic isolate, ii) Molecular mechanisms involved in the pathogenesis of the multiple endocrine neoplasia type 1; iii) genetic analysis and molecular study in patients affected by Hereditary Cushing's Syndrome iv) genetic analysis of hyperparathyroidism (*HRPT2*, *CASR* genes). Supervisor Prof. G Opocher.
- 2005 One year grant for young scientists from the University of Padova on the project "Fine mapping in Schizophrenia and Bipolar Disorder affected families originating from Chioggia (Italy). Supervisor Prof. ML Mostacciuolo.
- 2001–2004 Winner of Public selection contract and reserch fellowship, University of Padova. Project: i) 3'UTR functional study of SPG14 complicated form gene. ii) linkage analysis and identification of genes causative of hereditary neuromuscular

disorders (FSP, CMT and dHMN) at Human Genetics Laboratory, Department of Biology at the University of Padova. Supervisor Prof. ML Mostacciolo.

RESEARCH INTERESTS

- PRE-DOCTORAL Linkage analysis and haplotype reconstruction with specific programs and graphic tools. Detection of microsatellite marker alleles with fluorescent labeling and capillary electrophoresis and polyacrilamide gel electrophoresis. Different techniques such as sanger sequencing and chromatogram interpretation, DNA extraction from peripheral blood, purification and precipitation of nucleic acids, restriction fragment length polymorphism (RFLP).
- DOCTORAL Identification of genes mutated in X-linked and autosomal recessive forms of Hereditary Spastic Paraplegia by linkage analysis and by different mutations detections technics (Sanger sequencing, DHPL, resolution melting, SSCP, RFLP, Real Time PCR, MLPA) and study of the nucleotide variants role in phenotype determination (molecular cloning and cellular studies).
- POST-DOCTORAL Identification and characterization of new genes mutated in different hereditary disorders (CMT, HSP, dHMN, SLA, SMA distal dystrophy). Molecular diagnosis by Sanger sequencing of known genes of different hereditary disorders (CMT, HSP, dHMN, SMA SLA, distal dystrophy). Improving approaches to detect mutations by using NGS ION platform applied to Genetics of PCD (Primary Ciliary Dyskinesia) and Axonal CMT/dHMNs. Particular attention to Genetics of PTC (papillary thyroid cancer) by using more approaches based on linkage analysis and exome sequencing.