PERSONAL INFORMATION

Barbara Castellotti

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WORK EXPERIENCE

Dates (from 2019-today)

Italian NHS Reseacher

IRCCS Carlo Besta Neurological Institute Foundation

UO Medical Genetics and Neurogenetics

Analysis of gene panels by NGS for the study of patients with childhood epileptic encephalopathy and/or neuronal migration defects.
Evaluation by (trios-analysis) "exome-sequencing" (WES) of clinically instrumental and cytogenetically selected patients for identification of possible new disease genes related to infantile epileptic encephalopathy forms

Dates (from 2002-2019)

Contract Researcher

IRCCS Carlo Besta Neurological Institute Foundation
Study of the molecular basis of motor neuron disease: analysis of disease and/or susceptibility genes in Amyotrophic Latera

Dates (from 1994-2001)

Junior and Senior Scholarship

IRCCS Carlo Besta Neurological Institute Foundation
Study of neurogenetic disorders by triplet expansion.
Genotype-phenotype correlation

EDUCATION AND TRAINING

dates (2018-2022)

Medical Genetics Specialization

University of Milan

Study rare diseases, genetic counseling and return of NGS reports.
Genetic-molecular and cytogenetic techniques.
In-depth study of human genetics

1993 Membership in the Professional Register of Biologists (Mat. No. AA_041283)

1992 Bachelor's degree in Biological Sciences
University of Milan

1986 Scientific high school graduation
Liceo G. Gandini Lodi

ACHIEVEMENTS AND AWARD

Grants

RF-2019-12370491 Ministry of Health: Genetic-functional stratification as a guide for personalized treatment in epileptic and developmental encephalopathies caused by mutations in potassium channels" CUP: J45F21000050001 (RF221).
Coordinator U.O2 Investigator for Clinical recruitment, Gene panel analysis, Therapeutic intervention

TECHNICAL SKILLS

CELL CULTURES of peripheral and marrow blood, amniocytes, chorionic villi, fibroblasts, abortive tissue, immortalization of lymphocyte lines by EBV

CYTOGENETIC ANALYSIS: reconstruction of karyotypes from peripheral and bone marrow blood from amniocytes and chorionic villi

MOLECULAR GENETICS

- DNA extraction from lymphocytes (separated by gradient) and tissues by salt precipitation and phenol-chloroform purification
- RNA extraction from lymphocytes and from biopsy and autopsy tissues
- preparation of cDNA
- molecular analysis of DNA by PCR, RFLPs, Southern blot and Northern blot, SSCP (single strand conformational polymorphisms)
- direct nucleotide sequence analysis (automated sequencing) or after cloning into vectors
- synthesis of oligonucleotides
- mutation search by DHPLC
- quantitative PCR (real time PCR)
- MLPA (Multiplex Ligation-dependent Probe Amplification).
- HRM (High Resolution Melting PCR)
- Next Generation Sequencing (NGS): TruSeq Custom Amplicon Illumina, Nextera Illumina gene panels. Agilent SureSelect Rapid Capture Exome Sequencing (Whole Exome Sequencing) using Illumina employed Next Seq 550.