PATRIZIA MALASPINA_CV

EDUCATION

1980 Degree in Biological Sciences (cum laude), University "La Sapienza", Rome.

1992 PhD in Medical Genetics, University "La Sapienza" Rome.

1993 Degree of Specialization in Medical Genetics (cum laude), University "La Sapienza" Rome.

POSITIONS

1983-1985 Research fellow, Department of Biology and Genetics, University of Naples, Italy

1985-1989 PhD student, Department of Biology, University “Tor Vergata”, Rome, Italy

1987 Honorary research assistant, Galton Lab. (UCL, London, U.K.).

1991-92 Research fellow, MRC Human Biochemical Genetic Unit (UCL, London, U.K.)

1994-2001 Assistant Professor of Genetics, Department of Biology, University “Tor Vergata”, Rome, Italy.

2002 to present Associate Professor of Genetics, Department of Biology, University “Tor Vergata”, Rome, Italy.

HONORS

2001 CNR Short term mobility fellowship with the project “Molecular characterization of 4-idroxybutyric aciduria patients” at Dept. of Molecular and Medical Genetics, OHSU, Portland, OR, USA.

1991-1992 Bursary issued by EEC in the “Human Genome Analysis Program”

FUNDINGS

1996 Telethon project "Characterization of the genomic structure of the human SSADH gene"

1998-2000 Telethon project "Characterization of the genomic structure of the human SSADH gene"

2000-2002 Ministero degli Affari Esteri “Human biodiversity of Russsian population” Russian-Italian collaboration).

2001 Programma CNR – Agenzia 2000 “Individuazione delle linee evolutive del cromosoma Y umano nelle popolazioni dell’Italia meridionale”.

2002-2006 Ministero Affari Esteri Una banca del DNA rappresentativa della biodiversità genetica umana nella popolazione russa” “Human biodiversity of Russsian population” Russian-Italian collaboration).

2010-2013 Dipartimento Amministrazione Penitenziaria (DAP): attività sperimentali e di ricerca per la realizzazione di un laboratorio di tipizzazione genetica.

2020-2022 Progetto Regione Lazio POR FESR Lazio 2014-2020: "Sviluppo di un kit per la prevenzione all’osteoporosi basato su rischio genetico e stile di vita”.

Research field

In the last years, we addressed our research on the molecular characterization of genes relevant to GABA metabolism to identify variants that alter the risk of metabolic and neurodegenerative diseases with involvement of the GABAergic system. One of the objectives is the development of experimental protocols that allow the determination of fast and economic methods to identify molecular variants in groups of patients and controls. Furthermore, the expression of genes involved in the metabolism of GABA is studied through *in vitro* cell systems in response to endogenous or exogenous stresses that may alter this pathway.

Publications (last years)

1) MENDUTI G, BIAMINO E, VITTORINI R, VESCO S, PUCCINELLI MP, PORTA F, CAPO C, LEO S, CIMINELLI BM, IACOVELLI F, SPADA M, FALCONI M, **MALASPINA**, ROSSI L. (2018). Succinic semialdehyde dehydrogenase deficiency: The combination of a novel ALDH5A1 gene mutation and a missense SNP strongly affects SSADH enzyme activity and stability. *Mol Genet Metab*. 124:210-215.

2) MESSINA F, DI CORCIA T, RAGAZZO M, SANCHEZ MELLADO C, CONTINI I, **MALASPINA P**, CIMINELLI BM, RICKARDS O, JODICE C. (2018). Signs of continental ancestry in urban populations of Peru through autosomal STR loci and mitochondrial DNA typing. *PLoS One*. 13(7):e0200796.

3) BALZARINI M, ROVELLI V, PACI S, RIGOLDI M, SANNA G, PILLAI S, ASUNIS M, PARINI R, CIMINELLI BM, **MALASPINA P** (2019). Novel mutations in two unrelated Italian patients with SSADH deficiency. *Brain Metabolic Disease* 34:1515-1518.

4) MENDUTI G, VITALITI A, CAPO CR, LETTIERI-BARBATO D, AQUILANO K, **MALASPINA P**, ROSSI L (2020). SSADH variants increase susceptibility of U87 cells to mitochondrial pro-oxidant insult. *Int J Mol Sci* 21:4374. doi: 10.3390

5) CIMINELLI BM, MENDUTI G, BENUSSI L, GHIDONI R, BINETTI G, SQUITTI R, RONGIOLETTI M, NICA S, NOVELLETTO A, ROSSI L, **MALASPINA P**. (2020). Polymorphic Genetic Markers of the GABA Catabolism Pathway in Alzheimer's Disease. *J Alzheimer's Dis*. 77:301-311 doi: 10.3233/JAD-200429.