PERSONAL INFORMATION

FEDERICO ZARA

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Male, Napoli, 20/09/1968, Italian

Enterprise	University	EPR
☐ Management Level	☒ Full professor	☐ Research Director and 1st level Technologist / First Researcher and 2nd level Technologist
☐ Mid-Management Level	☐ Associate Professor	☐ Level III Researcher and Technologist
☐ Employee / worker level	☐ Researcher and Technologist of IV, V, VI and VII level / Technical collaborator	☐ Researcher and Technologist of IV, V, VI and VII level / Technical collaborator

WORK EXPERIENCE

From 2020 to date

Full Professor of Medical Genetics at University of Genoa, Head of Laboratory Department and Medical Genetics Unit, IRCCS G. Gaslini Institute

- Coordination of research and diagnostic testing activity

From 2021 to date

Director, Department of Laboratory and Services, IRCCS G. Gaslini Institute

- Budgeting and strategic planning of activities related to Laboratories (Genetics, Clinical and Anatomic Pathology Laboratories), Research Labs and Services (Radiology, Neuroradiology, Transfusion Center)

From 2023 to date

Scientific ViceDirector, IRCCS G. Gaslini Institute

- Coordination Experimental Research activities

EDUCATION AND TRAINING

From 1992 to 1996

MS Degree in Biological Sciences

110/110 cum laude

University of Genova

- Molecular Cytogenetics

From 1993 to 1997

Specialization in Medical Genetics

50/50 cum laude

University of Genova

- Genetics of Neurological Disorders, Molecular Cytogenetics

PERSONAL SKILLS

Mother tongue(s) Italian

Other language(s) English

Job-related skills Supervision of Research activities, Grant writing, Genetic Testing, Management of funds and personnel, Teaching

Project Management skills

Experience in coordination of Multicenter Research Projects in Life Sciences at national (Ministry of Health, Ministry of Reaserch, National Fundations) and international level (EU, international Fundations)

ADDITIONAL INFORMATION

Projects

International Consortia:

- Partner of the European project "Epicure"
- Partner of the European project "EpiPGX"
- Partner of the ILAE International Consortium on Epilepsy Genetics
- Partner of the European project "Desire"
- Partner of the “BrainStorm” Consortium

Funding from:

Telethon Foundation, Mariani Foundation, Italian Institute of Health,Italian Ministry of Health, Italian Ministry of Research, Compagnia di San Paolo, European Community, European Leukodystrophy Foundation, Jerome Lejeune Foundation

Active grants:

- Unveiling the hidden side of NEUrodevelopmental DIsorder Genetics (NEUDIG): a multidisciplinary pathway to new molecular diagnoses by integrating genomic, transcriptomic, and functional analyses – Italian Ministry of Research
- Human induced pluripotent stem cells from Down Syndrome: modelling synaptic dysfunction and exploring therapeutic approach – Lejeune Foundation
- Genome-editing regulation of alternative splicing provides new therapeutic opportunities for episodic ataxia type II – Telethon Foundation
- Implementing clinical exome sequencing into the diagnostic workflow of epileptic encephalopathies and exploiting its potential for personalized medicine – Italian Ministry of Health

Publication Track record

470 publications, over 20.000 citations, h-index of 70 (Scopus)

Publications

Selection of 10 relevant papers

- A Case-Control Survey Using Microarray-Based Comparative Genomic Hybridization. Arch Neurol. 2012; 69:322-30.
- Nava et al. De novo mutations in HCN1 cause early infantile epileptic encephalopathy. Nat Genet. 2014;46:640-5.
- Vanni et al. Impairment of ceramide synthesis causes a novel progressive myoclonus epilepsy. Ann Neurol. 2014;76:206-12.
- Niturad et al. Rare GABRA3 variants are associated with epileptic seizures, encephalopathy and dysmorphic features. Brain. 2017; 140:2879-2894.
- Fruscione et al. PRRT2 controls neuronal excitability by negatively modulating Na+ channel 1.2/1.6 activity. Brain. 2018; 141:1000-1016.
- Brainstorm Consortium. Analysis of shared heritability in common disorders of the brain. Science 2018; 360(6395).
- May et al. Rare coding variants in genes encoding GABA(A) receptors in genetic generalised epilepsies: an exome-based case-control study. Lancet Neurol. 2018; 17:699-708.
- Epi25 Collaborative. Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. Am J Hum Genet. 2019 Aug 1;105(2):267-282.
- Corbett et al. Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nat Commun. 2019; 10:4920.
- Lammertse et al. Homozygous STXBP1 variant causes encephalopathy and gain-of-function in synaptic transmission. Brain. 2020; 143:441-451.