

Hospital 12 de Octubre (Madrid, Spain)



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Biosketch:

I am Associated Consultant in Clinical Biochemistry, working at Biochemistry and Clinical and Genetics Departments of the '12 de Octubre' University Hospital, Madrid (Spain). I am a member of AEGH (Spanish Association for Human Genetics), and SEQC-ML (Spanish Society of Clinical Chemistry, and Laboratory Medicine). We received a Master degree in Pharmaceutical Sciences in 1998, a Clinical Biochemistry specialization degree in 1993, and got a PhD in Biochemistry and Molecular Biology in 2001. I received an MBA in Biotechnology in 2007.

My research lines are focused on the study of the biochemical and molecular genetic basis of the mitochondrial OXPHOS diseases, metabolic myopathies and exercise intolerance disorders (particularly, McArdle disease). I am in charge of the Mitochondrial and Neuromuscular Diseases group at the '12 de Octubre' Hospital Health Research Institute (i+12), a multidisciplinary group consisted of clinicians, biochemists, geneticists and molecular biologists.

I have participated as co-author in 201 indexed publications (Web of Science-Thompson), with an h-index of 39 (total cites 4749; mean cites per article 24.2). For more details, you can visit ORCID number 0000-0003-4741-772X.

I have supervised 7 doctoral theses and tutored several PhD students, as well as several post-doc researchers. Our research group belongs to the Spanish Network, CIBER for Rare Diseases (ISCIII). Our group belongs to Spanish National Reference Units (CSURs) of Metabolic Hereditary Disorders, and Neuromuscular disorders as well as to the European Reference Network Metab-ERN.