

Full professor of Medical Genetics at the Department of "Precision Medicine" of the "University "Luigi Vanvitelli" of Naples, Italy " and Associate Investigator of the Telethon Institute of Genetics and Medicine (TIGEM). Born in Naples July 28, 1960, graduated in Medicine. University Researcher from 1992 to 2000, associate professor from 2000 to 2006 and full professor of general pathology from 2006 to 2010. In 1982-1990 he was at the Institute of General Pathology and Oncology, as a student and then with a fellowship of the Italian cancer research association (AIRC), aimed to the study of the mechanism of action of the estrogen receptor. From 1989 to 1994, he was at the International Institute of Genetics and Biophysics (IIGB), CNR, Naples with Edoardo Boncinelli (developmental biology, identification of transcription factors that regulate embryogenesis and the formation of brain). Since 1992, his research team is involved in the study of muscular dystrophies. He published >180 articles in peer reviewed journals (h=47). Among the most significant results, the identification of delta-sarcoglycan and mutations that cause limb-girdle muscular dystrophy (LGMD2F), the identification of the gene that causes the cardiomyopathy of the BIO14.6 hamster, a leading experimental model. In addition, he identified the causes of other Mendelian disorders, such as FG syndrome 4, LGMD1F, etc. He directs the laboratory of Medical Genetics. He led research projects on the gene therapy of delta-sarcoglycanopathy and on the identification and classification of novel causes of genetic myopathies using next generation sequencing. He developed specific strategies for detecting mutations in neuromuscular disorders, lysosomal storage disorders, neurofibromatosis, kidney disorders, etc. He is coordinator of the Tigem-University Next Generation Sequencing facilities, and co-coordinator of the Telethon Undiagnosed Program that provides diagnosis of new and unrecognized genetic disorders based on NGS.