Francesco Ramirez

Current Position: Dr. Amy and James Elster Professor of Molecular Biology (Connective Tissue Diseases) in the Department of Pharmacology and Systems Therapeutics at Mount Sinai School of Medicine

Education: D.Sc. In Genetics (1969) from University of Sciences in Palermo, Italy

Non-scientific Interests: World history and current affairs, music, arts, movies, food and wine—in other words, NYC

I was born and raised in Italy where I received my graduate education in genetics and developmental biology. I then did a postdoctoral fellowship in the laboratory of Dr. Arthur Bank at Columbia University working on the molecular characterization of thalassemias. That experience forged my interest in the study of birth defects as well as my decision to pursue an academic career in the States. As a faculty in the department headed by Dr. Darwin Prockop at Rutgers Medical School, I applied my expertise in gene cloning to the collagen system and together with my colleagues defined the underlying defects in osteogenesis imperfecta, Ehlers Danlos syndrome, and chondrodysplasias. Later I joined Mount Sinai School of Medicine, and through a collaboration with the late Dr. Hollister, I became involved in the identification of the genetic lesion in Marfan syndrome, a goal that had long eluded significant research effort. My laboratory subsequently developed fibrillin mutant mice that have been instrumental in delineating the molecular pathogenesis of Marfan syndrome and the role of extracellular microfibrils in controlling TGFβ and BMP bioavailability during organ formation and tissue remodeling. My primary research interest remains the study of extracellular matrix components in mammalian development and in congenital and acquired disorders of the connective tissue.

Harry C. Dietz

Current Position: Victor A. McKusick Professor of Medicine and Genetics, Institute of Genetic Medicine, in the Departments of Pediatrics, Medicine, and Molecular Biology & Genetics; and Investigator, Howard Hughes Medical Institute, at Johns Hopkins University School of Medicine

Education: M.D. (1984) from SUNY Upstate Medical University; B.S.E. in Biomedical Engineering (1980) from Duke University

Non-scientific Interests: History of science (particularly cosmology and theoretical physics), marine aquariums, cooking and wine

I was initially trained as a pediatrician and cardiologist. In the 1950s, Dr. Victor McKusick declared a dedication to the study and care of individuals with Marfan syndrome, resulting in a large patient population at Johns Hopkins. While caring for children with Marfan syndrome, I became frustrated with the use of management protocols that seemed to make very little difference in their lives. I made the decision to leave clinical practice in order to receive basic science research training in human genetics—with the prime objective of interrogating the pathogenesis of Marfan syndrome in order to achieve informed therapeutic strategies. Within two years, while working in the laboratory of Dr. Clair Francomano at Johns Hopkins, I demonstrated that mutations causing Marfan syndrome occur in the gene encoding the connective tissue protein fibrillin-1. My clinical experience dictated that a pathogenetic model that singularly invokes structural failure of the tissues could not reconcile many findings in Marfan syndrome. The insights that Marfan syndrome largely reflects a failure of fibrillin-1 to properly regulate TGFβ and that many manifestations could be attenuated or prevented by TGFβ antagonists in mice, and perhaps people, are largely attributable to the dedication and insights of truly remarkable trainees in my lab. Improvement in the length and quality of life in my patients continues to be my primary research motivation.

Read Drs. Ramirez and Dietz's article entitled: Extracellular Microfibrils in Vertebrate Development and Disease Processes ... http://www.jbc.org/cgi/content/full/284/22/14677