ICORD
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Rare Diseases & Orphan Drugs

Petra Kaufmann

Petra Kaufmann is the director of both the Office of Rare Diseases Research and the Division of Clinical Innovation. Her work includes overseeing NCATS' Rare Diseases Clinical Research Network, Genetic and Rare Diseases Information Center, and Clinical and Translational Science Awards Program as well as the NIH/NCATS Global Rare Diseases Patient Registry Data Repository/GRDR® program. Kaufmann focuses on engaging a broad range of stakeholders to accelerate translation from discovery to health benefits through use of innovative methods and tools in translational research and training.

Before joining NCATS, Kaufmann was the director of the Office of Clinical Research at the National Institute of Neurological Disorders and Stroke (NINDS), where she worked with investigators to plan and execute a large portfolio of clinical research studies and trials in neurological disorders, including many in rare diseases. She established NeuroNEXT, a trial network for Phase II trials using a central institutional review board, streamlined contracting, active patient participation in all project phases, and a scientific and legal framework for partnership with industry. Kaufmann also promoted data sharing, working with multiple stakeholders from the academic, patient organization and industry sectors to develop data standards for more than 10 neurological diseases.

A native of Germany, Kaufmann earned her M.D. from the University of Bonn and her M.Sc. in biostatistics from Columbia University's Mailman School of Public Health. She completed an internship in medicine at St. Luke's/Roosevelt (now part of Mt. Sinai) in New York City, training in neurology and clinical neurophysiology at Columbia University, and a postdoctoral fellowship in the molecular biology of mitochondrial diseases at Columbia's H. Houston Merritt Clinical Research Center for Muscular Dystrophy and Related Diseases. Before joining NINDS, Kaufmann was a tenured associate professor of neurology at Columbia, where she worked as a researcher and clinician in the neuromuscular division, the electromyography laboratories and the pediatric neuromuscular clinic.

She has served on scientific advisory committees for many rare disease organizations and is a member of the American Academy of Neurology Science Committee, the International Rare Disease Research Consortium Interdisciplinary Scientific Committee and the Clinical Trial Transformation Initiative Steering Committee.

Kaufmann is board certified in neurology, neuromuscular medicine and electro diagnostic medicine. Kaufmann's research focus is on the clinical investigation of rare diseases, such as spinal muscular atrophy, amyotrophic lateral sclerosis and mitochondrial diseases. She currently sees patients in the Muscular Dystrophy Association Clinic at Children's National Medical Center.