

Louis Ptáček, MD

Dr. Ptáček's laboratory focuses on genetic diseases of muscle, heart, and brain, as well as hereditary variation of human sleep behavior. His group has cloned genes causing many disease and behavioral phenotypes in humans. In addition, he and collaborators probe normal function of the encoded proteins in nervous system and pathophysiology of mutant proteins in human diseases using cellular electrophysiology, biochemistry, cell biology, and animal modeling. One focus of his work is episodic neurological diseases. He pioneered the field of channelopathies that began with the mapping and cloning of a number of genes causing periodic paralysis and nondystrophic myotonias. He and collaborators identified and characterized the first human families with a Mendelian circadian rhythm variant. His group and that of Ying-Hui Fu have gone from the clinical and physiologic characterization of this phenotype to the mapping and cloning of causative genes, biochemical study of the encoded proteins, and generation of animal models.