

Age of Discovery

NEUROMUSCULAR DISORDERS

Kathryn Swoboda, M.D., Director of the Pediatric Motor Disorders Research Program, is spearheading research on spinal muscular atrophy (SMA), a rare genetic disorder that causes motor nerve degeneration, resulting in muscle wasting and weakness. SMA, the leading genetic cause of infant death, is caused by a mutation in the SMN1 gene, which leads to abnormally low levels of a protein needed for the survival of motor nerves. Currently, there is no known cure for SMA and treatment is based on supportive care. However, gene-based therapies that focus on increasing the amount of SMN protein offer new hope for SMA patients. In collaboration with ISIS Pharmaceuticals, Dr. Swoboda is now conducting Phase II studies of a drug that targets the backup copy of the SMN1 gene and alters how it is processed in order to boost SMN protein levels. The University of Utah is one of only four sites nationwide chosen to participate in this study, and Dr. Swoboda's patients will make up one-third of the study population.

"SMA is a devastating condition, and there is an urgent need for treatments that can modify the course of the disease and improve quality of life for these young patients," says Dr. Swoboda. "We have one of the largest SMA databases, with over 350 patients, and the ultimate goal of our research is to find a 'rescue therapy' that not only normalizes the amount of SMN protein, but also allows damaged motor nerves to recover."

MOVEMENT DISORDERS

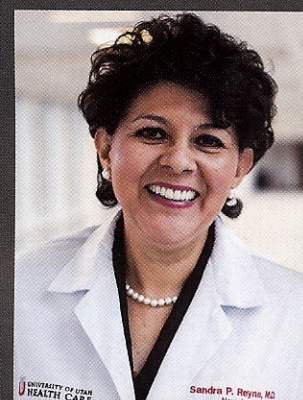
Daniel Scoles, Ph.D., Research Associate Professor of Neurology, is currently performing research on another rare genetic disorder known as spinocerebellar ataxia

2 (SCA2). SCA2 causes progressive degeneration of nerve cells in the cerebellum, the part of the brain that coordinates movement. SCA2 is caused by mutations in the ATXN2 gene, and there is no known therapy to delay or stop progression of the disease. In conjunction with the NIH Chemical Genomics Center, Dr. Scoles is screening chemical libraries for potential treatments for SCA2. Using a mouse model of SCA2, Dr. Scoles is also collaborating with ISIS Pharmaceuticals on screening potential drugs that can be injected directly into the brain to treat SCA2 by decreasing expression of the defective ATXN2 gene.

NEUROLOGY CLINICAL TRIALS UNIT

The Neurology Clinical Trials Unit (NCTU), under the direction of **Sandra Reyna, M.D.**, Assistant Professor of Neurology, has been instrumental in solidifying the University of Utah's reputation as a top-ranking center for clinical trials. The NCTU supports clinical and translational research in the neurosciences in every aspect of clinical trial management, from the very competitive process of site selection to contracting/negotiations, administration, study coordination/conduct, regulatory/compliance, and financing. With 26 active trials, the NCTU has the fourth largest clinical trial portfolio at the University of Utah.

"The NCTU is part of Dr. Pulst's vision of distinguishing the University of Utah as a leading academic research center," says Dr. Reyna. "Our track record in SMA, as well as our ability to recruit and retain clinical trial patients from around the world, is a testament to the personal effort and passion of members of our department who are willing to go above and beyond to provide outstanding patient care and research."



Sandra Reyna, M.D.,
Assistant Professor of Neurology



Daniel Scoles, Ph.D.,
Research Associate Professor
of Neurology