Title of Program: Pediatric Neuromuscular Disorders Research

Dept/Center/Lab: Department of Pediatrics

Principal Mentor: Susan Sparks, MD, PhD

Summary Description:
Dr. Susan Sparks is a board certified Pediatrician with additional board certifications in Clinical Genetics and Clinical Biochemical Genetics. She joined the Department of Pediatrics in the Division of Clinical Genetics at Levine Children's Hospital in March 2009. In addition to her clinical expertise in genetics and inborn errors of metabolism, she has a particular interest in neuromuscular disorders with a biochemical basis.

Dr. Sparks has joined the Neuromuscular and Muscular Dystrophy Research team at Carolinas Medical Center, the McColl-Lockwood Laboratory for Muscular Dystrophy Research, and is PI of the Pediatric Muscular Dystrophy Laboratory at CMC's Cannon Research Center. Due to its success as a highly respected research team in the international effort to advance neuromuscular research, this team recently was accepted as a member of the Cooperative International Neuromuscular Research Group (CINRG), an international network of medical and scientific investigators from 22 academic and research centers focused on finding a cure and advancing the standard of care for patients with DMD and related disorders. CINRG was founded in 1999 as the clinical research arm of the Duchenne Muscular Dystrophy Research Center and the Research Center for Genetic Medicine at the Children's National Medical Center in Washington, DC. Dr. Sparks is the principal investigator of the CMC CINRG site.

In addition to leading the CINRG site at Carolinas Medical Center, she has several clinical research projects on-going. These include:

1. Pediatric neuromuscular data registry
2. Evaluation of Limb-girdle muscular dystrophy
3. Lysosomal storage disease registries
4. Clinical and observational research studies in muscular dystrophy

She also has established a translational research laboratory, the Pediatric Muscular Dystrophy Laboratory, in Cannon Research Laboratories to improve the diagnostic evaluation of unknown muscular dystrophies, including congenital and limb-girdle muscular dystrophy. Her projects include:

1. Improved diagnostics for Limb-Girdle muscular dystrophy
2. Diagnosis and Etiology of Congenital Muscular Dystrophy
3. Glycosylation defects in muscular dystrophy

Dr. Sparks has opportunities for full-time and summer student projects in both clinical research and translational/laboratory research. Please feel free to contact Dr. Sparks (phone: 704-381-6810; email Dr. Sparks), if you are interested in hearing more about these opportunities.

Expectations and Role of Student: The successful student will be expected to bring enthusiasm, inquisitiveness, hard work, and passion. The student will be expected to learn well at

least one aspect of an ongoing project in the clinic or laboratory, to frame and refine an important hypothesis; and to design and carry out experiments designed to confirm or refute the key hypothesis. The student will present oral and written summaries of research and will be required to prepare and present an abstract and paper summarizing findings. Preferred students: Have an interest in clinical research and direct patient interaction.