

2010 MSG Abstract

Inherited Neuropathies Consortium

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The Inherited Neuropathies Consortium (INC) is one of the new members to the Rare Disease Clinical Research Network. The INC began with six sites – Wayne State University, University of Rochester, University of Pennsylvania, Children’s Hospital of Philadelphia, University of Miami, and the National Hospital for Neurology and Neurosurgery in Queens Square, London. We have since added two additional sites – Children’s Hospital of Westmead, Australia, and C. Besta Neurological Institute in Milan, Italy. By the end of the year we expect to have five additional US sites online. We are involved in three major studies in addition to our website. The first study is of the natural history of Charcot Marie Tooth disease. We are particularly interested in CMT types 1B, 2A, 4A, and 4C. Longitudinal data is being collected on all patients who consent to the study at participating centers, regardless of genetic subtype. The second study is looking for genetic modifiers to CMT1A and new genetic forms of CMT. The third study is validating a pediatric scoring scale for kids with CMT. We have two trainees who have begun the first of their two years in our fellowship. Overall, we are recruiting well for all three protocols, and we expect to continue to recruit patients rapidly and hope to be able to accumulate enough data to support a clinical trial by the end of the grant cycle. Our relationship with the MSG and MSG investigators is greatly aiding our translational efforts.