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For Immediate Release:

Researchers Identify Novel Gene Connected to Rare Muscle Disease

Bethesda, Md. — Researchers from the Department of Anesthesiology, Uniformed Services University of the Health Sciences, USU, along with research teams from the National Institutes of Health and from Australia, the Netherlands and Spain, have identified a novel gene on chromosome 15q that, when altered, causes nemaline myopathy with cores, a rare inherited muscle disorder. The gene encodes a member of the BTB/Kelch family of proteins.

Nemaline myopathy is a heterogeneous group of neuromuscular disorder characterized by the presence of thread- or rod-like (nemaline) bodies and occasionally core lesions in the skeletal myofibers. It is the most common non-dystrophic congenital myopathy that affects mainly infants and children. The presence of nemaline bodies in affected muscle is the tell-tale sign of this myopathy.

The research team used a variety of gene-hunting techniques in making their findings, including gene mapping, positional cloning, mutation screening, and high-throughput copy number analysis in the screening a cohort of patients with a history of the disorder. The patients, from Australia, the Netherlands, and Spain, who were found to have autosomal dominant mutation (only requires a single mutation to show itself) in the KBTBD13 protein, showed poor exercise tolerance, characteristic slowness of movements, gait abnormality, and the development of slowly progressive muscle weakness of the neck and proximal limb muscles beginning in childhood. “This particular protein can be added to the list of more than 60 proteins in BTB/Kelch family. However, only 3 of them have been associated with human disorders,” said Nyamkhishig Sambughin, Ph.D., USU, lead author on the study. “Although we do not know the function of KBTBD13 protein yet, it is important because it appears to be the first time there has been an association between this protein mutation and congenital muscle disease. The discovery of KBTBD13 is the first step toward understanding disease mechanism and the development of treatment for this devastating disease that affect mostly children ”

The findings appear in the December 10, 2010 issue of the *American Journal of Human Genetics*. An electronic version was made available on the publication’s web site on November 24, 2010.

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