

**TITLE:**

Genetics of Muscular Dystrophy

**FACULTY MENTOR NAME, EMAIL PHONE NUMBER**

Peter B. Kang, MD

pbkang@ufl.edu

352-273-8922

**FACULTY MENTOR DEPARTMENT**

Pediatrics

**RESEARCH PROJECT DESCRIPTION** (brief overview of background, hypothesis, methods, role of medical student, funding and relevant publications)

A number of causative genes have been linked to various forms of muscular dystrophy, yet many such patients remain without a genetic diagnosis. Some families have limited access to genetic testing, but others harbor mutations in novel genes. Our laboratory's core project involves gene discovery for muscular dystrophy and is funded by the National Institutes of Health. Genetic analyses will be supplemented by studies of human tissue and relevant disease models. The laboratory has extended the examination of one novel gene, *MEGF10*, into a separate spinoff project. A medical student could participate in either project, or potentially a new one if it complements the overall goals of the laboratory.

1. Boyden SE, Mahoney LJ, Kawahara G, Myers JA, Mitsuhashi S, Estrella EA, Duncan AR, Dey F, DeChene ET, Blasko-Goehring JM, Bönnemann CG, Darras BT, Mendell JR, Lidov HGW, Nishino I, Beggs AH, Kunkel LM, Kang PB. Mutations in the satellite cell gene *MEGF10* cause a recessive congenital myopathy with minicores. *Neurogenetics* 2012;13:115-124.
2. Mitsuhashi S, Mitsuhashi H, Alexander MS, Sugimoto H, Kang PB. Cysteine mutations cause defective tyrosine phosphorylation in *MEGF10* myopathy. *FEBS Letters* 2013;587:2952-2957.
3. Draper I, Mahoney LJ, Mitsuhashi S, Pacak CA, Salomon RN, Kang PB. Silencing of *drpr* leads to muscle and brain degeneration in adult *Drosophila*. *American Journal of Pathology* 2014;184:2653-2661.