Medical Student Research Program

Title: Genetics of Muscular Dystrophy

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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 independent 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-Goehringer 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.
4. Reddy HM, Hamed SA, Lek M, Mitsuhashi S, Estrella E, Jones MD, Mahoney LJ, Duncan AR, Cho KA, MacArthur DG, Kunkel LM, Kang PB. A homozygous nonsense mutation in *SGCA* is a common cause of LGMD in Assiut, Egypt. *Muscle & Nerve* 2016;54:690-695.
5. Reddy HM, Cho KA, Lek M, Estrella E, Valkanas E, Jones MD, Mitsuhashi S, Darras BT, Amato AA, Lidov HGW, Brownstein CA, Margulies DM, Yu TW, Salih MA, Kunkel LM, MacArthur DG, Kang PB. The sensitivity of exome sequencing in identifying causative mutations for LGMD in the United States. *Journal of Human Genetics*, in press.