

This week in techniques

Approach	Summary	Licensing status	Publication and contact information
Disease models			
Canine model for autosomal-recessive myotubular myopathy	Dogs with splicing mutations in <i>bridging integrator 1 (BIN1)</i> could be useful as models to help identify therapies for treating autosomal-recessive myotubular myopathy. Genomic analyses of three patients and five Great Danes with an inherited myopathy identified homozygous <i>BIN1</i> splicing mutations in skeletal muscle that were not observed in healthy subjects and dogs. Biopsies of muscle tissue from the affected dogs recapitulated multiple markers of disease seen in patient muscle tissue, such as nuclear internalization, fiber atrophy and membrane alterations. Next steps include establishing a population of Great Danes and smaller dog breeds harboring the <i>BIN1</i> mutation to help elucidate the pathogenesis of myotubular myopathy.	Unpatented; available for partnering	Böhm, J. <i>et al. PLoS Genet.</i> ; published online June 6, 2013; doi:10.1371/journal.pgen.1003430 Contact: Jocelyn Laporte, Institute of Genetics and Molecular and Cellular Biology, Illkirch, France e-mail: jocelyn@igbmc.fr
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