"Myotonic dystrophy: What is the basis for prevalent GI dysfunction?"

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Myotonic dystrophy type 1 is caused by a CTG repeat expansion with multisystemic effects including a variety of GI symptoms. The molecular mechanisms of the disease are known to involve a toxic gain of function of the CUG repeat RNA from the expanded allele. GI disturbances are common among patient reported symptoms but the mechanisms remain to be identified. Our group has used mouse models to study heart and skeletal muscle disease mechanisms and have initiated studies to define the molecular mechanisms causing GI symptoms.