

### WEEKLY GI RESEARCH WEBINAR

# "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.

<u>References:</u> (1) Morriss GR, Rajapakshe K, Huang S, Coarfa C, Cooper TA. 2018. Mechanisms of skeletal muscle wasting in a mouse model for myotonic dystrophy type 1. Hum Mol Genet 27:2789–2804 (2) Pang PD, Alsina KM, Cao S, Koushik AB, Wehrens XHT, Cooper TA. 2018. CRISPR -Mediated Expression of the Fetal Scn5a Isoform in Adult Mice Causes Conduction Defects and Arrhythmias. J Am Heart Assoc 7:e010393. (3) Brinegar AE, Cooper TA. 2016. Roles for RNA-binding proteins in development and disease. Brain Research 1647:1-8.

## MAY 6 • 4:00 PM

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