Myotonic Dystrophy FOUNDATION

Methods – Myotonic Dystrophy Animal Models & Tools

Animal models play a key role in basic, translational and clinical research. The following tables highlight and summarize available animal models and tools for myotonic dystrophy research. Literature links connect to the original publication.

This table contains descriptions of **research tools and methods** import for DM research. This table was last updated and reviewed in June 2024.

To find additional animal models or learn more about each respective system, please examine and follow the associated literature links and references within each table.

To find additional information and resources focused on myotonic dystrophy, visit the Myotonic Dystrophy Foundation website at: www.myotonic.org.

Methods - Myotonic Dystrophy Animal Models & Tools

Methods	Description	References
MBNL Concentration	Measurement of dose dependent splicing events and severity of DM	https://www.ncbi.nlm.nih.gov/pubmed/27681373
Splicing Biomarker	Alternative splicing changes in skeletal muscle may serve as biomarkers of disease severity and therapeutic response in myotonic dystrophy	https://www.ncbi.nlm.nih.gov/pubmed/23929620 Wang ET, Treacy D, Eichinger K, Struck A, Estabrook J, Olafson H, Wang TT, Bhatt K, Westbrook T, Sedehizadeh S, Ward A, Day J, Brook D, Berglund JA, Cooper T, Housman D, Thornton C, Burge C. Transcriptome alterations in myotonic dystrophy skeletal muscle and heart. Hum Mol Genet. 2019 Apr 15;28(8):1312-1321. doi: 10.1093/hmg/ddy432.
Non-invasive Splicing Biomarker	A panel of alternative splicing events assessed in urine	https://www.ncbi.nlm.nih.gov/pubmed/30254196
MLPA and ddPCR Assays to quantify toxic RNA	Two quantitative methods, Multiplex Ligation-Dependent Probe Amplification and droplet digital PCR, for studying the mutant DMPK transcript (DMPKexpRNA) and the aberrant alternative splicing in DM1 and DM2 human tissues and cells	https://www.nature.com/articles/s41598-018-24156-x www.nature.com/articles/s41467-023-37619-1#:~:text=In%20myotonic%20dystrophy%20type%201,frequency%20of%20oxidative%20muscle%20fibers.
Splicing assessment in DM1 patients	ddPCR based spcing assessment	https://insight.jci.org/articles/view/163856/pdf
Splicing assessment in DM1 patients	characterization of DM1 and DM2 patient-derived fibroblasts for use in small molecule screens and therapeutic studies using splicing assays	https://pubmed.ncbi.nlm.nih.gov/35479399/
Splicing assessment in DM1 patients	RNAseq	https://academic.oup.com/hmg/article/32/9/1413/6759135