

Failure of MBNL1-dependent post-natal splicing transitions in myotonic dystrophy. Authors: Lin X, Miller JW, Mankodi A, Kanadia RN, Yuan Y, Moxley RT, Swanson MS, Thornton CA.

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Research has suggested that certain proteins expressed in skeletal muscle, specifically CUG repeat binding protein 1 (CUG-BP1) and muscleblind like (MBNL), are involved in the defective splicing of select pre-mRNA in DM1 and DM2. However, information is limited on the degree and developmental regulation of CUG-BP1 and MBNL involvement. These authors explored the mechanisms of MBNL and CUG and CCUG expansion RNA in the obstruction of pre-mRNA splicing.

Defective splicing was observed during the post-natal period in transgenic mouse models with CUG repeat expansions. Results indicated that the MBNL1 protein was depleted from muscle nucleoplasm and distributed to the ribonuclear foci. MBNL1 deficiency inhibited normal post-natal splicing transitions. However, splicing failure was not correlated with MBNL2 protein deficiency.

The authors concluded that MBNL1 plays a significant role in modifying skeletal muscle during the post-natal interval by inhibiting splicing of pre-mRNA in DM. Additionally, the authors found their mouse models to be accurately representative of the mis-splicing observed in the skeletal muscle of DM1 and DM2 patients.

More results and conclusions can be found at:

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