Myotonia 9 Jackson; a new remutation in the *Clcn1* gene

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Mutation (allele) symbol: *Clcn1*$^{adr\text{-}mto9J}$

Mutation (allele) name: myotonia 9 Jackson

Gene symbol: *Clcn1*

Strain of origin: C57BL/6J

Current strain name: C57BL/6J-*Clcn1*$^{adr\text{-}mto9J}$/J

Stock #: 008253 (This allele is now only available as DNA from The Jackson Laboratory DNA Resource)

Phenotype categories: neurological

Abstract
We have identified a new remutation of the *Clcn1* gene by a direct test of allelism. The stiffened rear leg phenotype of the new *Clcn1*$^{adr\text{-}mto9J}$ remutation is very similar to that caused by the original *Clcn1*$^{adr\text{-}mto}$ mutation, however may be less severe in this new remutation.

Origin and Description
The *Clcn1*$^{adr\text{-}mto9J}$ remutation was found in 2005 by Stacey L. Dannenberg in a colony of ENU treated C57BL/6J mice at The Jackson Laboratory and was first recognized by the rear leg paralysis. Mice homozygous for this new remutation stiffen their rear legs when touched and this stiffening can be recognized by two weeks of age. Affected mice are smaller in size and thinner than littermate controls and exhibit hind end hair loss.

Genetic Analysis
This new mutant was shown to be an allele of myotonia (*Clcn1*$^{adr\text{-}mto}$) by mating a heterozygous female BALB/cByJ-*Clcn1*$^{adr\text{-}mto2J}$ mouse to a male mouse heterozygous for this new mutation. This mating produced 2 affected mice out of 25 born, proving allelism.

Pathology
Routine pathological screening showed that one homozygous male at 13 weeks, and one homozygous female and one male at seven weeks of age had no lesions. The eyes of three homozygous *Clcn1*$^{adr\text{-}mto9J}$ mutant mice and two littermate controls at 1 month of age were examined with an ophthalmoscope and were determined to be normal.

Electroretinogram (ERG) testing was also normal. Hearing as assessed by auditory brainstem response testing of three homozygous *Clcn1*$^{adr\text{-}mto9J}$ mutant mice and two littermate controls at 1 month of age showed normal hearing in all mice.

Discussion

This new remutation has been shown to have a very similar, but milder phenotype compared to that of the original myotonia mutants. *Clcn1*$^{adr\text{-}mto9J}$ will be available from The Jackson Laboratory DNA Resource. No embryos will be cryopreserved.

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