A remutation to bouncy on the C57BL/6J background, bouncy 8 Jackson

Son Yong Karst, Melissa L. Berry, David E. Bergstrom, and Leah Rae Donahue

Source of Support: This research was supported by grants RR01183 to the Mouse Mutant Resource (Leah Rae Donahue, PI) and Cancer Core Grant CA34196.

Mutation (allele) symbol: bc^{8J}

Mutation (allele) name: bouncy 8 Jackson

Gene symbol: *bc*

Strain of origin: C57BL/6J

Current strain name: C57BL/6J-bc^{8J}/GrsrJ

Stock #014106 (jaxmice.jax.org)

Phenotype categories: neurological

Origin and Description

A new recessive neurological mutation arose spontaneously in the C57BL/6J mouse strain at The Jackson Laboratory, where it was identified by Emma Tigno in 2009. Homozygous mutants exhibit a moderate tremor and impaired movement gait. Mutants are noticeable as early as 2-3 weeks of age and are smaller than littermates. Mutant mice live a normal life span and breed fairly well, although male homozygotes are not as productive as female homozygotes.

Genetic Analysis

Using our standard mapping procedure, a homozygote was mated with CAST/EiJ producing only non-affected F1 progeny and proving that this mutation is recessive. Intercrossed F1 progeny generated affected F2 animals for linkage analysis. The genetic map position for this new mutation was determined in the Fine Mapping Laboratory at The Jackson laboratory using their single nucleotide polymorphic (SNP) genotyping protocol. This mutation mapped to Chromosome 18 with a SNP-defined critical interval extending from NCBI 37 position 24079024 bp to NCBI 37 position 63144592 bp and concordant with a single SNP at NCBI 37 position 39065568 bp. This map position and phenotype indicated bouncy (*bc*) as a candidate gene and a direct allele test was set up by mating this new mutation with bc^{6J}. This mating produced 12 progeny of which 1 expressed the mutant phenotype, proving this new mutation to be a remutation to bouncy, and this mutation was then named bouncy 8 Jackson (bc^{8J}).

Pathology

A routine pathological screen of two bc^{8J} homozygotes at 19 weeks of age, showed mild hydrocephalus in one, which is common in C57BL/6J mice and is not a significant phenotype of this mutation. Hearing as assessed by auditory-evoked brainstem response

testing (ABR) of one mutant at 4 months of age was normal, consistent with other mutants of this gene.

Acknowledgments
The authors thank Emma Tigno for discovery of the mutant, Roderick T. Bronson and Coleen Kane for pathological screening, Chantal Longo-Guess for ABR testing.