A Neurological Mutation on Chromosome 14 named shimmy 2 Jackson

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Mutation (allele) symbol: *shmy*^{2J}

Mutation (allele) name: shimmy 2 Jackson

Strain of origin: B10.D1-*H2*^{q}/SgJ

Current strain name: B10.D1-H2*q*/SgJ-*shmy*^{2J}/GrsrJ

Stock #005133 (jaxmice.jax.org)

Phenotype categories: neurological

Abstract
A neurological mutation exhibiting a shaky/wobbly gait has been identified and mapped to Chromosome 14 in the same region as the mutation shimmy (*shmy*). A complementation test for allelism between this new mutant and the shimmy mice produced affected animals proving that they are allelic.

Origin and Description
Mice carrying the spontaneous recessive *shmy*^{2J} mutation were found by Marianne Urquhart in October 2001, in a production colony of B10.D1-*H2*^{q}/SgJ mice at The Jackson Laboratory and were brought to The Mouse Mutant Resource (MMR). Homozygous mutants are easily recognized by 5 weeks of age by their shaking and wobbly gait. The mice appear to vibrate and this vibration can be felt when the mice are held by their tails.

Genetic Analysis
Using the standard mapping protocols of The Mouse Mutant Resource, an intercross between B10.D1-*H2*^{q}/SgJ-*shmy*^{2J}/J and CAST/Ei F1 hybrids was set up and generated 42 affected F2 animals for linkage analysis. The *shmy*^{2J} mutation maps to mouse Chromosome 14, distal to *D14Mit252* (at 6.0 cM) and proximal to *D14Mit45* (at 12.5cM). The previously described mutation shimmy (*shmy*) has been mapped to position 6.5 cM. A direct test for allelism was set up by mating a mouse homozygous for this new mutation with a mouse homozygous for the original *shmy* mutation. From this mating 4 progeny out of 7 born were affected proving that the new mutation is an allele of shimmy.

Pathology
A pathological screen of one *shmy2J/shmy2J* mutant and one control littermate at 3 months of age revealed no gross lesions. Similarly, the pathological studies of the

original *shmy* showed a very few dystrophic axons in the lateral nucleus of the cerebellum in mutants, but did not reveal the cause of the neurological phenotype (Lane, et al.1994).

Hearing of four homozygotes and two heterozygotes as assessed by ABR testing was determined to be normal.

Discussion

The similarity in neurological phenotype, chromosomal location, and the results of the test for allelism confirm that *shmy*^{2J} is a remutation to shimmy (*shmy*). Likewise, the chromosomal location and similarity of the locomoter phenotypes of *shmy* snd shmy^{2J}, with the BK Channel (BK-/-) knockout mice (Sausbier, et al.2004), make the *Kcnma1* gene a good candidate for the *shmy* mutation.

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References

Lane PW, Cook SA, Bronson RT, Johnson KR, and Davisson MT. Shimmy (*shmy*), A New Mutation On Chromosome 14 Of The Mouse. Mouse Genome 1994, 92(4), 686-687.

Mouse Genome Database (MGD) Mouse Genome Informatics Project, The Jackson Laboratory, Bar Harbor, Maine. World Wide Web (www.informatics.jax.org)

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