

Scrambler 5 Jackson, a new spontaneous mutation in the gene disabled 1

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Mutation (allele) symbol: *Dabl*^{scm-5J}

Mutation (allele) name: scrambler 5 Jackson

Gene symbol: *Dabl*

Strain of origin: BKS.Cg-*Dock7*^m +/+ *Lepr*^{db}/J

Current strain name: BKS(Cg)-*Dabl*^{scm-5J}/GrsrJ

Stock #016903 (jaxmice.jax.org)

Phenotype categories: neurological

Origin and Description:

Mice that were thin, weak, and had an ataxic gait were found in the BKS.Cg-*Dock7*^m +/+ *Lepr*^{db}/J strain at The Jackson Laboratory by Lindsay Keaney. These mutant mice are severely unstable in their gait and are unable to right themselves at about two weeks of age. Nearly all die by three weeks of age and none have survived beyond four weeks of age. This colony is maintained by ovarian transplantation. Transplanted recipient females are crossed with C57BLKS/J mice and then heterozygous offspring are intercrossed. This mutation has been backcrossed 4 generation to C57BLKS/J and the misty and diabetes mutations have been bred out of this mutant subline.

Genetic Analysis:

A transplant host with ovaries from a mutant transplant donor was mated with an FVB/NJ male. The F1 hybrid mice from this mating all had a normal looking phenotype, proving that this mutation has recessive inheritance. These F1 hybrid mice were intercrossed and generated affected and unaffected F2 mice for linkage analysis. Using standard SNP protocols, linkage analysis for this mutation was completed in the Fine Mapping Laboratory at The Jackson Laboratory. This mutation mapped to Chromosome 4, between position 85,096,243 bp and position 119,566,789 bp (MGSCv37).

Based on phenotypic similarities, a direct test for allelism was performed by mating this new mutation to a scrambler 4 Jackson (*Dabl*^{scm-4J}) mouse. This mating produced 11 progeny, of which 5 pups had the scrambler mutant phenotype proving this new mutation to be an allele of *Dabl*.

Pathology:

A routine pathological examination of two homozygous males at 3.5 weeks of age showed that the brains of these mice had the scrambled arrangement of the cerebral cortex, hippocampus and cerebellum that is found in other *Dabl* mutants.

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