Small swaying lethal; a new spontaneous neurological mutation on Chromosome 6

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Mutation (allele) symbol: *ssl*

Mutation (allele) name: small swaying lethal

Strain of origin: BALB/cJ

Current strain name: BALB/cJ-*ssl*/GrsrJ

Stock #006019 (jaxmice.jax.org)

Phenotype categories: neurological

Abstract
A new spontaneous recessive mutation has been identified and mapped to Chromosome 6. Mice homozygous for the *ssl* mutation are smaller than their littermates and lean and fall over on their sides.

Origin and Description
Mice carrying the small swaying lethal (*ssl*) mutation were discovered by Amy Fickett in a production colony of BALB/cJ at the Jackson Laboratory in May of 2001. The mutants are recognized at 12-15 days of age. As the affected mice age, their neurological phenotype becomes more pronounced, and they do not survive past four to five weeks of age. Ovarian transplants of homozygous females have not been successful. The BALB/cJ-*ssl*/J colony is maintained by progeny testing.

Genetic Analysis
Using the standard mapping protocols of The Mouse Mutant Resource, a linkage cross was set up by mating a female heterozygous for the *ssl* mutation to a CAST/EiJ male mouse. The mutation carrying offspring of this mating were then intercrossed and produced 50 affected F2 mice that were used for linkage analysis. Using standard PCR protocols a genome scan confirmed linkage of the *ssl* mutation on Chromosome 6. The mutation maps between *D6Mit38* (NCBI 36 position 100.0 Mb). and *D6Mit55* (NCBI 36 position 112.4 Mb) and is non-recombinant with *D6Mit107* (NCBI 36 position 113.5 Mb) and *D6Mit11*(NCBI 36 position 114.2 Mb).

Based on phenotype and map position similarities, a direct test for allelism was set up with deaf waddler by mating a C3H/HeJ-*Atp2b2*^{dfw}/J with a mouse carrying the new *ssl* mutation. This mating produced no affected progeny out of 27 born, so it is unlikely that the new *ssl* mutation is an allele of deaf waddler.

Pathology
A pathological screen of 3 homozygous mutants at three weeks of age revealed atrophy

of the thymus in one mutant and no gross lesions in the other two animals.

Hearing as assessed by auditory brainstem response testing of one homozygous mutant at three weeks of age revealed no hearing loss, providing further evidence that *ssl* is not allelic with deaf waddler.

The eyes of a 19-day-old female mouse appeared normal when examined with an ophthalmoscope, but ERG testing of the same animal showed normal rods and poor cones.

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