Splotch-like 2; a new semidominant spotting mutation located on Chromosome 1

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Mutation (allele) symbol: *Splchl2*

Mutation (allele) name: Splotch-like 2

Gene symbol: *Splchl2*

Strain of origin: C57BL/6J

Current strain name: C57BL/6J-*Splchl2*/GrsrJ

Stock #:012853

Phenotype categories: color spotting and tail

Abstract
A new semidominant spotting mutation named splotch-like 2 has been mapped to Chromosome 1. Mice heterozygous for this mutation display predominantly white belly spots of varying sizes, but may also have various spotting on the feet and tail and may have a curly tail. The curly tail may disappear with age. Homozygotes (*Splchl2/ Splchl2*) may be dying *in utero* as no live homozygotes have been produced. Sibling (+/+) mice have a normal phenotype.

Origin and Description
The Splotch-like 2 mutation was discovered by Peter Keeney in a production colony of C57BL/6J mice at The Jackson Laboratory and was recognized by its belly spot and curled tail (see photos on the MGI allele detail page). It is maintained by mating a mouse heterozygous for the splotch-like 2 mutation with a wild type sibling. Both of these genotypes have a normal lifespan.

Genetic Analysis
Using our standard mapping protocols a backcross to *Mus Castaneus* produced 40 affected mice that were used to map the Splotch-like 2 mutation to Chromosome 1. The Splotch-like 2 mutation maps between *D1Mit77* (NCBI 37 position 73.7 Mb) and *D1Mit488* (NCBI 37 position 91.9 Mb) and is non-recombinant with *D1Mit7* (NCBI 37 position 74.9 Mb), *D1Mit46* (NCBI 37 position 75.5 Mb), *D1Mit79* (NCBI 37 position 76.6 Mb), *D1Mit216* (NCBI 37 position 79.8 Mb), and *D1Mit44* (NCBI 37 position 82.5 Mb). Based on phenotype and map location the Splotch-like 2 mutation is likely an allele of the *Pax3* gene. A direct test for allelism was not performed as both mutations are semidominant.

Pathology
A pathological screen of one heterozygote male and two wild type females showed no somatic lesions. Hearing as accessed by auditory brain stem testing (ABR) on one heterozygote and two normal mice showed normal hearing. The eyes of four heterozygotes and four wild type mice were examined with an ophthalmoscope and were determined to be normal.

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