Recessive spotting-like 2 Jackson: a new remutation on mouse Chromosome 5

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Mutation (allele) symbol: $rslk^{2J}$

Mutation (allele) name: recessive spotting-like 2 Jackson

Gene symbol: rslk

Strain of origin: C57BL/6J

Current strain name: C57BL/6J-rslk^{2J}/GrsrJ

Stock #006107 (jaxmice.jax.org)

Phenotype categories: coat color

Abstract

A new spontaneous recessive remutation that causes affected mice to have a diluted gray coat color and white spotting has been characterized and mapped to Chromosome 5. A direct test for allelism confirmed that this new mutation is an allele of recessive spotting-like (*rslk*) and it has been named recessive spotting-like 2 Jackson.

Origin and Description

Mice carrying the $rslk^{2J}$ remutation were discovered by Tina Morse in a production colony of C57BL/6J mice at the Jackson Laboratory. Mice homozygous for this spontaneous and recessive remutation are recognized by their diluted gray coat color and white spotting that occurs randomly on both dorsal and ventral sides of the mice.



Figure 1: Three mice carrying the $rslk^{2J}$ mutation in the background and a littermate control on the lower right of photo. All one month of age.

Homozygous mutants live a normal lifespan and both sexes breed. The colony is maintained by homozygous x heterozygous matings. The phenotype of the original recessive spotting (*rs*) mutation is strain background dependant, whereby on some strain backgrounds the subtle spotting in heterozygotes is not observed. Subtle belly spotting has been observed in C57BL/6J-*rslk*^{2J}/J heterozygotes.



Figure 2: A three-month-old $rslk^{2J}$ homozygote on the left and a littermate control on the right

Genetic Analysis

Using the standard mapping procedures of The Mouse Mutant Resource, a homozygous male $rslk^{2J}$ mouse was mated to a CAST/Ei female mouse. The heterozygous F1 mice produced from this mating were then intercrossed and produced 43 affected F2 progeny of which 21 were used for linkage analysis. The new $rslk^{2J}$ remutation was mapped to Chromosome 5, 7 cM distal to D5Mit255 (NCBIm34 position 53.7 Mb) and 2.3 cM proximal to D5Mit114 (NCBIm34 position 80.4 Mb) and is non-recombinant with D5Mit235 (NCBIm34 position 74.0 Mb).

Based on the phenotype and map position similarities of this new mutation to the previously described recessive spotting-like (*rslk*) mutation, a direct test for allelism was set up by mating a female mouse homozygous for this new mutation with a male homozygous B6(SJL) $Ptprc^{a} Pepc^{b}$ -*rslk*/J mouse (Stock #005574) and also mating a female homozygous B6(SJL) $Ptprc^{a} Pepc^{b}$ -*rslk*/J mouse to a male mouse homozygous for this new mutation. From these two matings 17 progeny were produced in 3 litters and all displayed the diluted coat color and white spotting proving allelism. The original recessive spotting (*rs*) mutant is cryopreserved and was not available for the direct test for allelism.

Pathology

A routine pathological screen of 2 homozygous mutant mice and 2 controls at 21-25 weeks of age revealed no gross lesions.

Hearing as assessed by auditory brainstem response testing of two homozygous mutants and 2 controls at 3 months of age revealed no hearing loss.

The eyes of 2 homozygous mutant mice at 4.5 months of age were tested with an ophthalmoscope and were determined to be normal.

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