Ruby-eye 8 Jackson; a remutation in the *Hps6* gene

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Mutation (allele) symbol: *Hps6*$^{ru\text{-}8J}$

Mutation (allele) name: ruby-eye 8 Jackson

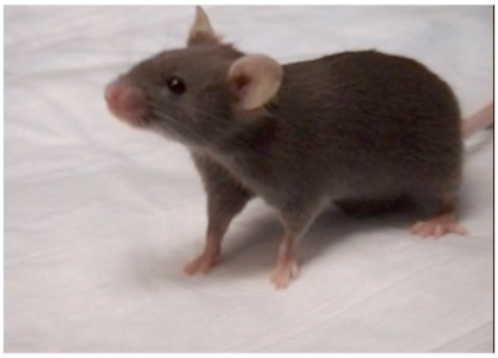
Gene symbol: *Hps6*

Strain of origin: B6.CB17-*Prkdc*^{scid}/SzJ

Current strain name: B6(CB17)-*Hps6*$^{ru\text{-}8J}$/J

Stock #: 009368

Phenotype categories: coat color



An *Hps6*$^{ru\text{-}8J}$ homozygote. Note the gray coat color and ruby colored eyes

Origin and Description

The new recessively inherited ruby-eye 8 Jackson (*Hps6*$^{ru\text{-}8J}$) mutation arose spontaneously and was found by Joyce Preble in a production colony of B6.CB17-*Prkdc*^{scid}/SzJ mice at the Jackson Laboratory. Like previously described ruby-eye mutants, mice homozygous for the ruby-eye 8 Jackson mutation have a gray coat color and ruby colored eyes. Homozygous mutant mice can be recognized when their first coat of hair comes in at about 10 days of age. Both homozygous and heterozygous mice breed well and live a normal lifespan.

Platelet storage pool deficiency in mouse pigment mutations are associated with seven distinct genetic loci. Ruby-eye in a homozygous condition greatly reduces the number of melanocytes in retina, ear skin, harderian gland and nictitans. (Markert and Silvers1956).

The ruby-eye mutation causes a reduced number of projections of retinal ganglion cells to the ipsilateral lateral geniculate nucleus (LaVail et al. 1978). Ruby-eye mice have a platelet storage pool deficiency, characterized by prolonged a bleeding time, normal platelet number, and low platelet dense granule numbers and a dense granule serotonin content (Novak et al.1984).

Genetic Analysis

This new mutation has recessive inheritance as shown by the results of backcrossing homozygous B6(CB17)-*Hps6*$^{ru\text{-}8J}$/J mice to unaffected C57BL/6J mice. This mating produced only unaffected N1 progeny proving that the new mutation has recessive inheritance.

A direct test for allelism was performed. A mouse homozygous for the original *ru*

mutation was mated to a *ru-8J* heterozygote mouse and this mating produced 17 progeny of which 5 pups were born with the *ru* phenotype.

A B6(129S4)-*Hps6*$^{ru\text{-}7J}$/J homozygous mouse was mated to a mouse heterozygous for this new *ru-8J* mutation. This mating produced 29 progeny, of which 26 were affected with the *ru-7J* phenotype, proving allelism.

The *Hps6* gene is located on Chromosome 19 at NCBI build 37 position 46077998-46080643 bp. Mapping and sequencing data has not been generated for the *ru-8J* mutation.

References

Markert CL and Silvers WK. The effects of Genotype and Cell Environment on Melanoblast Differentiation in the House Mouse. Genetics 41 (3) 429-50 May 1956.

LaVail JH; Nixon RA; Sidman RL. Genetic control of retinal ganglion cell projections. J Comp Neurol 182 (3)399-421 1978 Dec 1

Novak EK; Hui SW; Swank RT. Platelet storage pool deficiency in mouse pigment mutations associated with seven distinct genetic loci. Blood 63(3)Pages: 536-44 1984 Mar

Acknowledgements

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