Ruby-eye 7 Jackson (*ru-7J*); a new remutation in the *Hps6* gene

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Mutation (allele) symbol: *Hps6*$^{ru\text{-}7J}$

Mutation (allele) name: ruby-eye 7 Jackson

Gene symbol: *Hps6*

Strain of origin: B6.129S4-Cd86^{tm1Shr}/J

Current strain name: B6(129S4)-*Hps6*$^{ru\text{-}7J}$/GrsrJ

Stock #005559 (jaxmice.jax.org)

Phenotype categories: coat color

Origin and Description

A new spontaneous recessive remutation of ruby-eye (*Hps6*^{ru}) has been identified. Mice carrying this new remutation were discovered in a colony of B6.129S4-*Cd86*^{tm1Shr}/J (Stock #003609) at the Jackson Laboratory. Mice homozygous for the new remutation were crossed to C57BL/6J mice to eliminate the *Cd86* targeted mutation. Mutants are recognized by a grey coat color and ruby colored eyes. Like the previously described ruby-eye mutation, mice homozygous for *Hps6*$^{ru\text{-}7J}$ also have severe loss of pigment in the retina and in the ear skin. The B6(129S4)-*Hps6*$^{ru\text{-}7J}$/J colony is maintained by homozygous and heterozygous sibling matings.

A mouse homozygous for the *Hps6*$^{ru\text{-}7J}$ mutation at four months of age. Note the light coat color, the ruby colored eyes and lack of pigment in ear skin.

Genetic Analysis
Using standard mapping protocols, this new remutation was mapped to Chromosome 19 between markers *D19Mit11* (NCBI 36 position 42.4 Mb), and *D19Mit58* (NCBI 36 position 50 Mb), and is non-recombinant with *D19Mit17* (NCBI 36 position 45.6 Mb).

Based on map position and phenotype similarities to the previously described ruby-eye (*ru*) mutation, a complementation test was performed. A mouse homozygous for the ruby-eye mutation (B6.Cg-*Hps6*^{ru}/JLlp) was bred to a mouse heterozygous for this new mutation. This mating produced 1 litter of 5 progeny of which 3 displayed the ruby-eye phenotype, proving this new remutation is an allele of ruby-eye (*Hps6*^{ru}).

Pathology
A routine pathological screen of two mutants and two controls at eight and thirteen weeks of age and revealed no gross abnormalities in any of the mice.

Hearing as assessed by auditory-evoked brainstem response testing of three homozygous mutants and one control at four weeks of age revealed no hearing loss.

The eyes of one homozygous mutant were examined with an ophthalmoscope and had severe pigment loss in the peripheral retina. Electroretinogram (ERG) testing was normal.

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