A new spontaneous mutation to pink-eyed dilution, *Oca2*$^{p\text{-}18J}$

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Mutation (allele) symbol: *Oca2*$^{p\text{-}18J}$

Mutation (allele) name: pink-eyed dilution 18 Jackson

Gene symbol: *Oca2*

Strain of origin: B10.RIII-*H2*^{r} *H2-T18*^{b}/(7INS)SnJ

Current strain name: B10.Cg-*H2*^{r} *H2-T18*^{b} *Oca2*$^{p\text{-}18J}$/GrsrJ

Stock #14100

Phenotype categories: Pigmentation

Abstract
A spontaneous recessive mutation that causes affected mice to have a diluted coat color and reduced eye pigmentation has been characterized and mapped to Chromosome 7. A direct test for allelism confirmed that this new mutation is an allele of *Oca2*^{p} and it has been named pink-eyed dilution 18 Jackson.

Origin and Description
The *Oca2*$^{p\text{-}18J}$ mutation was discovered by Bonnie Williams in a production colony of B10.RIII-*H2-T18*^{b}/(7INS)SnJ mice at the Jackson Laboratory. Mice homozygous for this spontaneous mutation are recognized by their diluted coat, yellowish-brown color, and red eye pigmentation. Mutants have red eyes at an early age, and when the mutants get older their eyes become darker. Both homozygous and heterozygous mice breed well and live a normal lifespan.

Genetic Analysis
Using our standard mapping procedures, a homozygous *Oca2*$^{p\text{-}18J}$ mouse was mated to a CAST/EiJ mouse. This mating produced unaffected F1 progeny proving the new mutation to be recessively inherited. The unaffected F1 hybrids were intercrossed, and affected F2 mice were generated for linkage analysis. The *Oca2*$^{p\text{-}18J}$ mutation was mapped to Chromosome 7 between *D7Mit230* (NCBIm34 position 56.7 Mb) and *D7Mit277* (NCBIm34 position 75.0 Mb).

Based on the phenotype and map position similarities of this new mutation to the previously described *Oca2*^{p} mutation, a direct test for allelism was set up by mating mice homozygous and heterozygous for this new mutation with *Oca2*^{p} homozygotes. This mating produced 28 progeny and 21 of the progeny displayed the diluted coat color with red eye phenotype, proving this new mutation allelic to *Oca2*^{p}.

Pathology
A routine pathological screen of a homozygote at 4 weeks of age revealed mild hydrocephalus. This is likely incidental. Everything else appeared normal.

Hearing as assessed by auditory brainstem response (ABR) testing of two homozygous mutants at 5 weeks of age revealed no hearing loss.

The eyes of two homozygous mutants at 5 weeks of age were tested with an ophthalmoscope; the mutants were determined to be normal.

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Addendum
In 2011 this mutation was found to be a C to A transversion resulting in a change from alanine to aspartic acid at amino acid 649.