

Rhino 10 Jackson: a new mutation in the hairless gene

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Mutation (allele) symbol: Hr^{rh-10J}

Mutation (allele) name: rhino 10 Jackson

Gene symbol: Hr

Strain of origin C57BL/6J

Current strain name: C57BL/6J- Hr^{rh-10J} /GrsrJ

Stock #021500 (jaxmice.jax.org)

Phenotype categories: skin and hair

Origin and Description:

We have identified a new hairless mutation in the progeny of an ENU mutagenized C57BL/6J mouse. This hairless mutation mimics the original rhino (Hr^{rh}) mutant phenotype with its wrinkled skin, small size, long toenails, and hairless appearance as it ages. It was discovered by Leslie Haynes at The Jackson Laboratory in a mutagenized colony of C57BL/6J mice. The skin appears thick and wrinkly, and the mutant is easily identifiable by one week of age by its skin and subsequently by its lack of hair by three weeks of age. Homozygotes have some hair on their legs, tail and face by 3 weeks of age but lose all of their hair by 8 weeks of age, with only their straight whiskers left. This colony is maintained by mating a homozygous female or male to C57BL/6J, then subsequently sibling intercrossing the obligate heterozygous offspring. Homozygotes do not usually breed well, but are usually fertile if mated as soon as they reach adulthood. Homozygotes live a normal lifespan. This colony is maintained in a standard animal room and on standard rodent diet. The average litter size for heterozygous matings is 6.95 pups per litter and with 7 surviving mutants out of 132 pups born from heterozygous intercrosses, far fewer than the expected 25% affected progeny are observed.

Genetic Analysis:

This mutant was proven recessive by outcrossing to C57BL/6J and obtaining no mutants in the F1 hybrid population, then intercrossing unaffected F1 hybrids and obtaining mutant and wild type mice in the F2 segregating population. Using standard SNP protocols, linkage analysis for this mutation was completed in the Fine Mapping Laboratory at The Jackson Laboratory. This mutation mapped to Chromosome 14 between positions 66,780,687 and 79,390,536 (NCBI37/mm9), an interval that includes the gene hairless (Hr). A complementation test using female heterozygotes of the new mutation and a male rhino Jackson homozygous from the strain RHJ/LeJ produced 3

affected progeny and 5 normal progeny, thus confirming this a mutation in *Hr*.

Pathology:

Two females at 9 weeks and 11 weeks of age and one male at 11 weeks of age had hair follicles that were very dilated or cystic (pilary cysts), which is typically seen in the hairless/rhino mice. No other lesions were observed in a normal pathology screen. An ophthalmic check of one female and one male homozygote showed normal retinas, white corneas and possible eye infections in both.

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