Caracul 18 Jackson

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Mutation (allele) symbol: *Krt71*$^{Ca\text{-}18J}$
Mutation (allele) name: Caracul 18 Jackson
Gene symbol: *Krt71*
Strain of origin: NOD/ShiLtJ
Current strain name: NOD/ShiLtJ-*Krt71*$^{Ca\text{-}18J}$/GrsrJ
Stock #022312 (jaxmice.jax.org)
Phenotype categories: skin and hair

Origin and Description:
A spontaneous mutation was identified in the NOD/ShiLtJ inbred strain that causes a wavy coat, noticeable by two weeks of age, and a slight bend or wave at the ends of the whiskers. Both sexes are viable and are good breeders. These mutants live a normal lifespan when maintained in a standard mouse room. This mutant line has been maintained by brother/sister mating a mutant of either sex with a wild-type sibling.

Genetic Analysis:
An outcross of a mutant to 129S1/SvImJ produced 16 affected offspring out of 24 total progeny, proving that this is a dominant mutation. A backcross of these affected F1 hybrid mice to NOD/ShitJ mice produced affected and unaffected mice in the N2 progeny. These N2 mice were used for mapping. SNP analysis localized this mutation to Chromosome 15. None of the 25 heterozygotes showed recombination with rs3023429 on Chromosome 15 position 101,810,823 bp (GRCm38). Exome sequencing detected a heterozygous G to T transversion at Chromosome 15 position 101,736,583 (GRCm38) that is predicted to result in a C431Y missense mutation in Keratin 71 (*Krt71*). This mutation was confirmed to be present in four affected heterozygous animals and absent in four unaffected controls using PCR amplification with primers Krt71_1F GGCAGGTTGTAATGGCTGAG and Krt71_1R GTGACAGTGCCCTCAAGGAT and Sanger sequencing. This mutation has been designated caracul 18 Jackson, *Krt71*$^{Ca\text{-}18J}$.

Pathology:
A routine pathological screen showed two mutant females at 8 and 10 weeks of age and two mutant males at 10 weeks of age were normal except for indications of insulitis, typical of the NOD/ShiLtJ background. Eye examinations were performed on two heterozygotes and two controls at three months of age. All had normal eyes.

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