

Caracul 17 Jackson, a remutation in keratin 71

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Mutation (allele) symbol: *Krt71*^{Ca-17J}

Mutation (allele) name: caracul 17 Jackson

Gene symbol: *Krt71*

Strain of origin: C57BL/6J

Current strain name: C57BL/6J-*Krt71*^{Ca-17J}/GrsrJ

Phenotype categories: skin and hair

Origin and Description

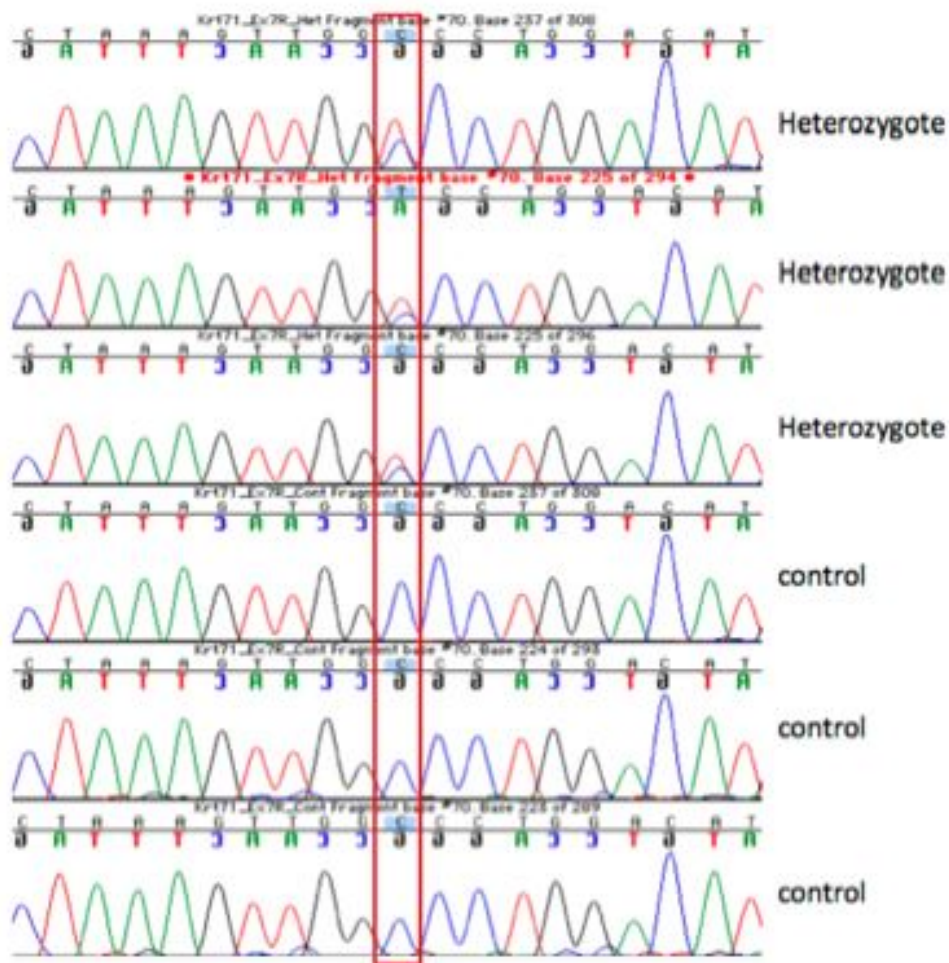
This new dominant mutation was induced by ENU mutagenesis of C57BL/6J mice at The Jackson Laboratory and was discovered by Leslie Haynes. Mice carrying the caracul 17 Jackson mutation can be recognized when a first coat of abnormally wavy hair comes in at about 7 to 8 days of age. Mutants are easily recognizable at 3 weeks of age by their very wavy coat and curved vibrissae. The mutant coat becomes less wavy with age but remains ruffled throughout adulthood. Both female and male heterozygotes are viable and fertile.

Genetic Analysis

Based on similar phenotype, *Krt71* was considered a potential candidate gene. A *Krt71* PCR product from genomic DNA of this mutant was produced for sequencing. Primers were generated that produce a 358 base pair *Krt71* product from wildtype controls: primer exon 7 Left (ACTGAATGTGAGGGGTTTGC) and primer exon 7 Right (TCTGCCTGCTGAGCTGC). Sequence analysis of mutant DNA identified a single nucleotide transition at position 101567032 on Chromosome 15 in *Krt71*. This mutation is predicted to change amino acid 425 from alanine to valine.

Mutant	Control
TTTGCTGCA GATAATGCC ATGCCTGCTT CTGTTTTCTA C L Q I M P M P A S V F . ACTTCTTTGC ATTTAGGCTT CTAACCTAGA GACAGCCATC L L C I . A S N L E T A I GCTGATGCCG AGCAGCGAGG TGACAGTGCC CTCAAGGATG A D A E Q R G D S A L K D A CCCGGGCCAA GCTGGATGAG CTGGAGGGTG CCTGCACCA R A K L D E L E G A L H Q GGCCAAGGAG GAGCTGGCCA GGATGCTGCG TGAATATCAG A K E E L A R M L R E Y Q GAGCTCATGA GCCTAAAGTT GCTCTGGAC ATGGAGATCG E L M S L K L V L D M E I A CCACCTACCG CAAACTTCTG GAGAGCGAGG AGTGCAGGTG T Y R K L L E S E E C R C TGCAGAGTGT GC A E C	TTTGCTGCA GATAATGCC ATGCCTGCTT CTGTTTTCTA C L Q I M P M P A S V F . ACTTCTTTGC ATTTAGGCTT CTAACCTAGA GACAGCCATC L L C I . A S N L E T A I GCTGATGCCG AGCAGCGAGG TGACAGTGCC CTCAAGGATG A D A E Q R G D S A L K D A CCCGGGCCAA GCTGGATGAG CTGGAGGGTG CCTGCACCA R A K L D E L E G A L H Q GGCCAAGGAG GAGCTGGCCA GGATGCTGCG TGAATATCAG A K E E L A R M L R E Y Q GAGCTCATGA GCCTAAAGTT GCCTGGAC ATGGAGATCG E L M S L K L A L D M E I A CCACCTACCG CAAACTTCTG GAGAGCGAGG AGTGCAGGTG T Y R K L L E S E E C R C TGCAGAGTGT A E

A portion of the protein coding region of *Krt71*. The control DNA sequence and its amino acid translation are shown on the right, and the *Krt71*^{Ca-17J} mutant DNA sequence and its translation on the left. A single nucleotide transition is enclosed by a blue box in the mutant sequence and a green box in the control sequence. The mutation is predicted to change amino acid 425 from alanine to valine. This change is indicated by a red box in the control and the mutant sequence.



Comparison of DNA sequence chromatograms of three heterozygous *Krt71*^{Ca-17} mutants and three controls.

Acknowledgements

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