Caracul-like recessive (*calre*): a new curly coat mutation mapping near the *Krt2-6g* locus on Chromosome 15

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Mutation (allele) symbol: *calre*

Mutation (allele) name: caracul-like recessive

Gene symbol: *calre* (probable allele of *Krt2-6g*)

Strain of origin: BKS.Cg-*m*+/+*Lepr*^{db}/J

Current strain name: BKS(Cg)-*calre*/GrsrJ

Stock #005736

Phenotype categories: Hair

Abstract
A spontaneous recessive curly coat mutation has been identified and mapped to Chromosome 15 in the same position as the previously described mutations caracul (*Krt2-6g*^{Ca}) and caracul-like (*Cal*). A direct test for allelism was not set up between mice carrying the *Ca* or *Cal* mutations and mice carrying this new mutation, because both *Ca* and *Cal* are dominant alleles.

Figure 1. Two 2-week-old calre/calre mice with curly coats and a littermate control with a normal smooth coat (upper right).

Origin and Description
The caracul-like recessive mutation was discovered by Leslie Haynes in 2004, in a production colony of BKS.Cg-*m*+/+*Lepr*^{db}/J mice at the Jackson Laboratory. The curly

coat of this spontaneous homozygous mutant mouse is recognized at 10-12 days of age when hair is fully covering the body. At 2 weeks of age the coat of homozygous mutants looks very curly, and the mutants also have curly vibrissae (see images on the allele detail page). After several weeks, the curly phenotype is reduced; the hair becomes fuzzy in appearance, and the vibrissae straighten out and appear normal. Some mutant mice have no whiskers at wean age. Also, in older homozygous and heterozygous mice more whiskers are lost, and homozygous mutants lose more hair as they age. Homozygous mutant mice live normal life spans and breed normally. The BKS.Cg-*m*+/+*Lepr*^{db}/J-*calre*/J colony is maintained by mating homozygous x heterozygous or heterozygous x heterozygous mice. Descriptions of four dominant caracul-like mutants have also been characterized by the Mouse Mutant Resource: *Cal4*, *Cal5*, *Cal6*, and *Cal7*. This new *calre* mutation differs from the previously described *Cal* mutations in that it has a recessive mode of inheritance.

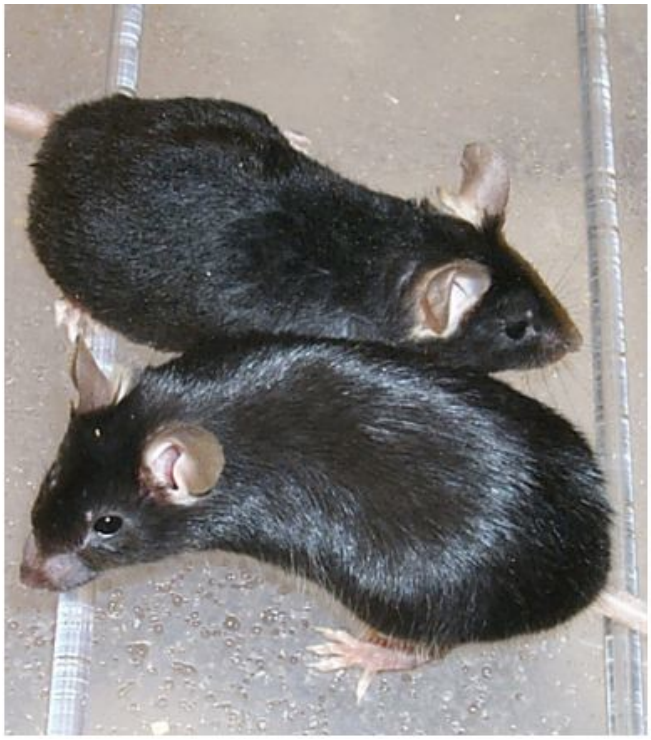

Figure 2. A *calre*/*calre* mouse (upper) compared to a normal littermate control (lower), both at 4 weeks of age. At this age the coat of the mutant is losing its curl.

Genetic Analysis

This mutation has recessive inheritance as shown by mating a homozygous BKS.Cg-*m*+/+*Lepr*^{db}/J-*calre*/J female mouse to an unrelated male +/+ CAST/Ei mouse. This mating produced all unaffected progeny proving the new mutation to be recessive. The unaffected F1 hybrids produced in this cross were then intercrossed, and 50 affected F2 animals were produced for linkage analysis. This new mutation was mapped to Chromosome 15, distal to *D15Mit14* (1 recombinant/40 meioses tested) and non-recombinant with *D15Mit16*. The marker *D15Mit14* is at the 99.7 Mb NCBIm34 position, *D15Mit16* is at the 103Mb NCBIm34 position, and the original caracul mutation is positioned at the 101.7Mb NCBIm34 position.

Pathology

A pathological screen of two homozygous caracul-like recessive mutant mice revealed no gross lesions. Plucked hair samples taken from mutant mice showed that there is a normal distribution of hair types, but zigzag hairs have abnormal bends and kinks. Skin sections were histologically normal.

Hair from a *calre* homozygous mutant showing bending and kinks (see arrow) 10X mag.

Hearing as assessed by auditory-evoked brainstem response testing of two homozygous mutants and 2 controls at 3 months of age revealed moderate hearing loss in both homozygous mutant and control mice. A homozygous mutant tested at 7 months and another at 12 months of age were both deaf. This hearing loss is characteristic of the BKS.Cg-*m*+/+*Lepr*^{db}/J background strain and not the result of the new mutation. The eyes of 2 homozygous mutant mice were tested with an ophthalmoscope and were determined to be normal.

Discussion

Based on the similarity of the hair phenotype with caracul mutants (*Krt2-6g*^{Ca}) and the position on Chromosome 15, it is likely that caracul-like recessive is a remutation of the *Krt2-6J* gene.

Acknowledgements

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References

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