

A New Hair Mutation Named Ruffled Maps to Chromosome 13

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Mutation (allele) symbol: *rul*

Mutation (allele) name: ruffled

Strain of origin: RB156Bnr/EiJ

Current strain name: RB156Bnr/Ei-*rul*/J

Stock #005362 (jaxmice.jax.org)

Phenotype categories: skin and hair

Abstract

We have identified a new recessive mouse mutation that causes abnormal ruffled looking hair that can be classified at about 10 days of age. The mutation was mapped using an intercross with CAST/Ei and was found to be on Chromosome 13 at the 18 cM position.



A 6-month-old mutant homozygous for the ruffled mutation is shown at the top of the photo compared to a control littermate below.

Origin and Description

This mutation arose in the RB156/Bnr/EiJ strain in the Mouse Mutant Resource at The Jackson Laboratory in 1989 and was discovered by Belinda Harris. The phenotype of this mutation can be identified at about 10 days of age by a ruffled looking coat, which is

different than the smooth coat of control littermates. The hair of mutant mice is sparse and has some curling. The ruffled looking coat is maintained throughout the animal's lifespan, unlike the caracul and caracul-like mutants who lose some of their curly coat with age. Mice homozygous for the ruffled mutation have a normal lifespan and both sexes breed normally.

Genetic Analysis

This mutation was first identified as a recessive mutation by crossing an RB156Bnr/EiJ-*rul*/J mutant to an inbred C57BL/6JEi. In this cross, no mutants were produced in the F1 generation, but mutants were produced in the F2 intercross generation. Using the standard mapping procedures of The Mouse Mutant Resource, an intercross with CAST/Ei was set up and generated 24 affected progeny of which 22 were used for linkage analysis. In order to avoid errors by misclassification, only animals with the most severe phenotype were used in the linkage analysis. The mutation maps on mouse Chromosome 13 between *D13Mit60* (NCBI33 position 35.3 Mb) and *D13Mit19* (NCBI33 position 43.2 Mb) and is non-recombinant with *D13Mit18* (NCBI33 position 35.6 Mb). Direct tests for allelism were set up by crossing a RB156Bnr/EiJ-*rul*/J mutant with hair mutants that have been mapped to Chromosome 13: furless (*fs*), juvenile alopecia (*jal*), and juvenile depilation (*jd*). The results showed 0 affected progeny out of 46 born in tests with *fs*, 0 affected progeny out of 17 born in tests with *jal*, and 0 affected progeny out of 32 born in tests with *jd* proving that all three of these Chr mutations are not allelic with ruffled. Before the map position of *rul*/J was known, direct tests for allelism were set up by crossing a RB156Bnr/EiJ-*rul*/J mutant with each of the following known mutants: lanceolate hair (*lah*) on Chr 18, fuzzy (*fz*) on Chr 1, waved 1 (*wal*) on Chr 6, waved 2 (*wa2*) on Chr 11, and frizzy (*fr*) on Chr 7. All were found not to be allelic.

Pathology

A routine pathological screen done on two male mutants showed one with no lesions and the other animal had a severe cataract and mild hydrocephalus. Results of auditory brainstem response¹ tests showed that homozygous mutant mice (N=12) and littermate heterozygous controls (N=6) have normal hearing up to ten weeks of age. At older ages, some hearing loss is observed in both mutant and control mice, likely an effect of the strain background. Eyes were examined with an ophthalmoscope and showed cataracts in both mutants and controls, which is characteristic of the RB156Bnr/Ei strain background.

Discussion

A previously unknown hair mutation causing a sparse and ruffled coat has been identified and mapped to Chr 13 at the 18 cM position. The cause of this new mutation has not been determined.

Acknowledgements

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References

Mouse Genome Database (MGD) Mouse Genome Informatics Project, The Jackson Laboratory, Bar Harbor, Maine. World Wide Web (2005) (www.informatics.jax.org)

MGSC27.33c.1. Mouse Genome Sequencing Consortium
(www.ensembl.org/Mus_musculus/)

¹ **ABR thresholds** in mice are determined using a semi-automated computer system (Intelligent Hearing Systems, Miami, Florida). Subdermal needle electrodes are inserted at the vertex and ventrolaterally to both ears of anesthetized mice. Specific auditory stimuli from 10-100 dB SPL are delivered binaurally through plastic tubes from high frequency transducers. ABR thresholds are obtained, in an acoustic chamber, for clicks and for 8, 16, and 32 kHz pure-tone pips.