

Bad hair day (*Bhrd*), a semidominant X-linked mutation affecting skin and hair.

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Mutation (allele) symbol: *Bhrd*

Mutation (allele) name: Bad hair day

Strain of origin: B6.V-*Lep*^{ob}/J

Current strain name: B6(V)-*Bhrd*/GrsrJ

Stock #008129 (jaxmice.jax.org)

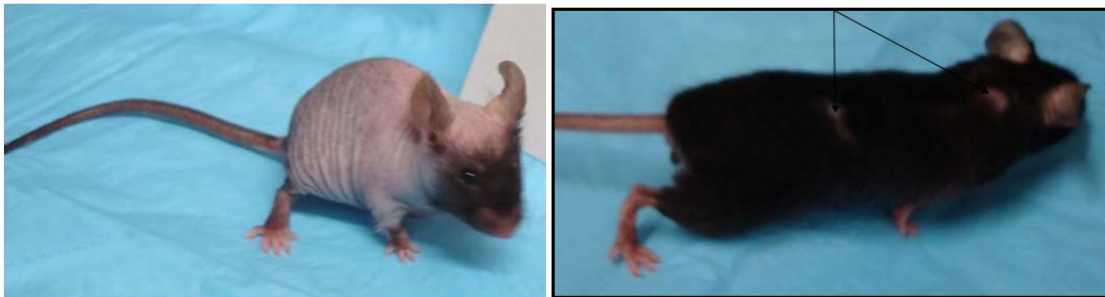
Phenotype categories: Skin and hair

Abstract

A new spontaneous, semi-dominant mutation has been identified and named bad hair day (*Bhrd*). This new mutation has been mapped to Chromosome X in the same region as the previously described patchy fur (*Paf*) mutation. A direct test for allelism was not performed because the original patchy fur mutation is only available as cryo-preserved embryos.

Origin and Description

The bad hair day mutation arose spontaneously at the Jackson Laboratory in a colony of B6.V-*Lep*^{ob}/J mice in 2004 and was discovered by Leanne Belcher. Female mice affected by the *Bhrd* mutation (*Bhrd*/+) have patches of fur missing in the coat, and in affected males (*Bhrd*/Y) the phenotype is more severe and the animals appear almost bald. This mutation is linked to Chromosome X, identified by mating male *Bhrd*/Y mutant mice with C57BL/6J females. This mating produced only affected females (*Bhrd*/+). Matings of normal looking males to (*Bhrd*/+) females, produced males with the almost bald phenotype and females with the patchy fur. Homozygous mutant females are almost as bald as hemizygous male mutant mice. Both female and male mutant *Bhrd* mice live normal life spans and breed normally.



Hemizygous male at 7 weeks of age on left and heterozygous female at 20 weeks of age on right with arrows pointing to bare patches in the coat.

Genetic Analysis

Using The Mouse Mutant Resource standard mapping protocols, female mice carrying the *Bhrd* mutation were mated to unaffected CAST/EiJ male mice. The affected female F1 mice generated by this first cross were then backcrossed to unaffected male mice from the *Bhrd* colony. This cross generated 25 affected mice of which 21 were used for linkage analysis. This mutation was mapped distal to DXMit10 (4.7% recombination) (NCBI 36 position 146.6 Mb), and is non-recombinant with DXMit156 (NCBI 36 position 158.6 Mb), DXMit160 (NCBI 36 position 162.5 Mb), DXMit29 (NCBI 36 position 163.2 Mb), and DXMit30 (NCBI 36 position 163.7Mb). The original *Paf* mutation has not been mapped to the NCBI 36 assembly, however the centimorgan position listed for *Paf* in the Mouse Genome Informatics database is 73.3 cM. The same centimorgan position of 73.3 is listed for our non-recombinant markers: DXMit160, DXMit29, and DXMit30. A direct test for allelism was not performed because the original patchy fur mutation is only available as cryo-preserved embryos.

Pathology

Our standard pathological screen of a (*Bhrd*/+) mutant female mouse at 26 weeks of age and an almost bald male (*Bhrd*/Y) mouse at 18 weeks of age revealed no gross abnormalities.

Hair samples taken from a bald (*Bhrd*/Y) male were viewed microscopically and were short and thin, with no normal hairs present. Hair samples from a patchy (*Bhrd*/+) female were normal. Hair in both mice had more pigment than normal.

Hearing assessed by auditory brain stem¹ (ABR) testing on one female (*Bhrd*/+) mouse at 20 weeks of age was normal. ABR testing on two bald male (*Bhrd*/Y) mice showed high frequency hearing loss at 8 weeks age.

The eyes of a (*Bhrd*/+) female at 8 weeks of age, and a bald (*Bhrd*/Y) male at 8 weeks of age were examined with an ophthalmoscope and an electroretinogram (ERG) test was performed, and both showed normal results.

Acknowledgements

We thank Leanne Belcher for discovery of the mutant, Rod Bronson and Coleen Marden for pathological screening, Chantal Longo-Guess for hearing assessment, Norm Hawes and Ron Hurd for the eye examinations.

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.