Psoriasis-like skin disease severity1-like; a new mutation affecting skin and hair

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Mutation (allele) symbol: *psds1l*

Mutation (allele) name: psoriasis-like skin disease severity1-like

Strain of origin: BALB/cJ

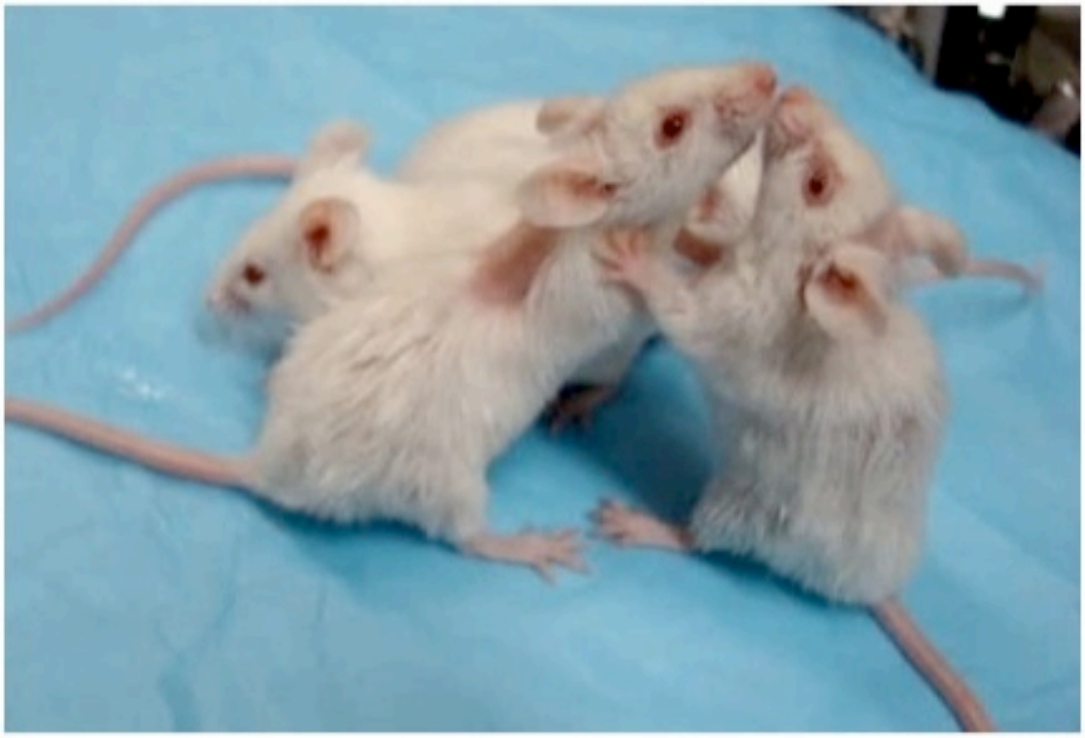
Current strain name: BALB/cJ-*psds1l*/GrsrJ

Stock #013715 (jaxmice.jax.org)

Phenotype categories: Skin and hair

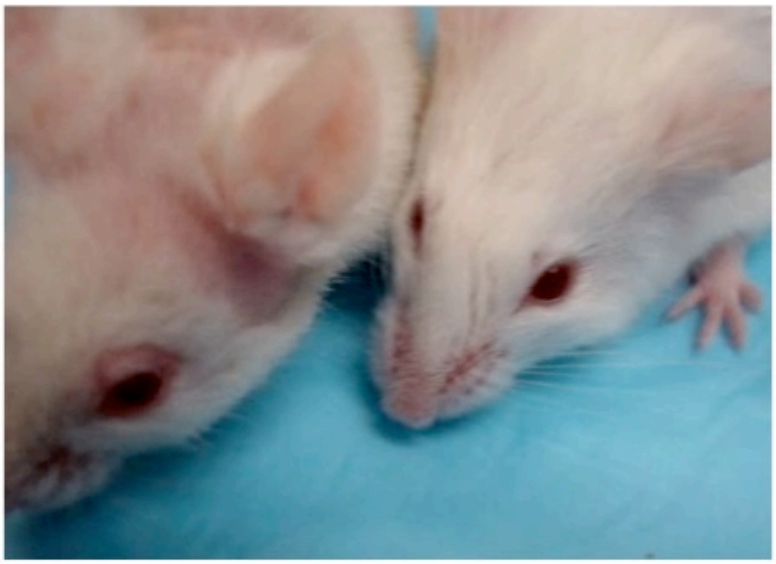
Origin and Description

The psoriasis-like skin disease severity1-like (*psds1l*) mutation arose spontaneously in a colony of BALB/cJ inbred strain mice and was discovered by Scott MacKenzie at The Jackson Laboratory. Mice homozygous for the *psds1l* mutation have a dirty white coat color and sparse hair; they can be recognized when their first coat of hair comes in.



Two mice affected by the *psds1l* mutation are shown in the foreground with an unaffected littermate control in the background. Note the lack of hair around the eyes of the affected mice and lack of hair on the back of the affected mouse on the left. All mice are 8 weeks of age.

Homozygotes often show a crusty growth around their eyes; usually there is no hair around the eyes. Compared with heterozygous or wild-type littermates, *psds1l* homozygotes have much less hair than their littermates and they frequently have loss of hair around the cervical area. Homozygotes are viable and fertile.



A *psds1l* homozygote at 8 weeks of age is shown on the left compared
To an unaffected littermate control shown on the right. Note the lack of hair on the top of the head and around the eye on the *psds1l* mutant.

Genetic Analysis

A *psds1l* homozygote was mated to an unaffected wild type A/J mouse. The normal F1 progeny from this mating were intercrossed and generated 54 affected mice for linkage analysis. Using standard SNP protocols, linkage analysis for the *psds1l* recessive mutation was done in The Fine Mapping Laboratory at The Jackson Laboratory.

The *psds1l* locus maps to Chromosome 4 and has been positioned between *D4Mit327* (NCBI 37 Position 86.0Mb) and *D4Mit176* (NCBI 37 Position 101.1Mb). There is no recombination with *D4Mit26* (NCBI 37 Position 88.7Mb) in 42 meioses tested. Based on phenotype and map position similarities *psds1* (NCBI 37 Position 86.0Mb) was thought to be a good candidate. Mice carrying the *psds1* mutation were not available to perform a direct test for allelism. As it was not possible to do the mating to confirm allelism this new mutation was named psoriasis-like skin disease severity1-like.

Pathology

A routine pathological screen of a homozygous *psds1l* mouse and a littermate control mouse at 20 weeks of age showed dermatitis in the homozygote with both the homozygote and control mouse displaying mild muscular dystrophy.

Auditory brain stem response testing of one *psds1l* homozygote and one control showed normal hearing.

The eyes of one *psds1l* homozygote at 19 weeks of age were examined by electroretinograph and showed abnormal cones and rods. One *psds1l* homozygote had normal eyes at age of 5 weeks as observed with an ophthalmoscope.

Acknowledgements
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