Pearl 15 Jackson, a remutation of the *Ap3b1* gene

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Source of Support: The research was supported by NIH/NCRR grant RR01183 to the Mouse Mutant Resources (M. T. Davisson, PI) and Cancer Center Core Grant CA34196.

Mutation (allele) symbol: $Ap3b1^{pe\text{-}15J}$

Mutation (allele) name: pearl 15 Jackson

Gene symbol: *Ap3b1*

Strain of origin: C3.Sw-$H2^{b}$/SnJ

Current strain name: C3(SW)-$H2^{b}$ $Ap3b1^{pe\text{-}15J}$/GrsrJ

Stock #: 005952 (available only as DNA from the Jackson Laboratory DNA Resource)

Phenotype categories: Coat color

Origin and Description

The $Ap3b1^{pe\text{-}15J}$ remutation was discovered by Dawn Martin in a production colony of C3.Sw-$H2^{b}$/SnJ mice (Stock #000438) in AX-12 at the Jackson Laboratory. Mice homozygous for this spontaneous, recessive mutation are recognizable by a diluted light gray coat color. This dilution also lightens the eyes, feet, ears and tail. Both homozygous males and females breed and live a normal life span. Descriptions of three other remutations of the *Ap3b1* gene, $Ap3b1^{pe\text{-}13J}$, $Ap3b1^{pe\text{-}14J}$ and $Ap3b1^{pe\text{-}16J}$ are also available on this web site.

Genetic Analysis

The $Ap3b1^{pe\text{-}15J}$ mutation was first identified as a recessive mutation by crossing a female C3H.Sw/SnJ mouse carrying this new mutation to an inbred C57BL/6J mouse. In this cross no mutants were produced in the F1 generation, but mutants were produced in the F2 intercross generation. Using our standard mapping procedures an intercross with CAST/Ei was set up and generated 42 affected progeny that were used for linkage analysis. This new mutation maps on mouse Chromosome 13 between *D13Mit191* (NCBIm34 position 70.2 MB) and *D13Mit147* (NCBIm34 position 94.3 Mb) and is non-recombinant with *D13Mit191* (NCBIm34 position 81.5 Mb), *D13Mit159* (NCBIm34 position 88.8 MB), and *D13Mit128* (NCBIm34 position 93.5 Mb).

Based on phenotype and map position similarity of this new mutation to the previously described $Ap3b1^{pe}$ (NCBIm34 position 90.5-90.7 Mb) mutation, a direct test for allelism was set up by mating a female mouse, homozygous for the $Ap3b1^{pe\text{-}11J}$ mutation, to a

male homozygous for this new mutation. This mating produced a litter with a total of 7 progeny of which all were affected, proving the new mutation to be an allele of the *Ap3b1*^{pe} gene.

Pathology

A routine pathological screen[1] done on one homozygous and one heterozygous mouse showed no lesions.

Acknowledgements

The authors wish to thank Dawn Martin for the discovery of the mutant and Coleen Marden for her excellent technical assistance

[1] **Standard Histology Protocol used in the Mouse Mutant Resource**

For fixation of tissues, mice were deeply anesthetized with tribromoethanol (avertin) until they no longer displayed a withdrawal reflex in the hind limbs and then perfused intracardially with Bouin's fixative following a flush of the vasculature with saline solution. After soaking in Bouin's for one week to demineralize bones, tissues were dissected. Six segments of spine with axial muscles and spinal cord in situ, representing cervical, thoracic and lumbar spinal segments, were dissected. The brain was removed and sliced into 6 cross sectional pieces at the levels of olfactory lobes, frontal cortex, striatum, thalamus, midbrain, rostral and caudal medulla with cerebellum. Midsagittal slices of hind leg through the knees were prepared. Slices of basal skull through the pituitary and inner ears were taken. Both eyes, salivary glands and submandibular lymph node, trachea plus thyroid and sometimes parathyroid were removed and cassetted. A longitudinal slice of skin from the back was removed. The thymus, slices of lung, and a longitudinal slice of heart were cassetted. Similarly slices of liver through gall bladder, kidney with adrenal attached, pancreas and spleen were prepared. The stomach was sliced longitudinally to include both squamous and glandular portions. Loops of small intestine from 3 levels and slices of large intestine and cecum were removed, as were slices of urinary bladder. The whole uterus, with ovaries attached, was taken. In males testes were sliced longitudinally. The accessory male organs including seminal vesicles, coagulating gland and prostate were removed en block. Altogether in most cases all tissue fit into a total of 10 cassettes. The cassettes were processed in an automatic tissue processor to dehydrate tissues which were then embedded in paraffin. Six micron sections were cut and stained with hematoxylin and eosin (H&E). Sections of brain and spinal cord in vertebral bones also were stained with luxol fast blue (LFB) for myelin and cresylecht violet (CV) for cellular detail.