

C57BL/6J-Pax3<sup>Sp</sup> /J

Stock No: 002469

 Spontaneous Mutation

Please contact Technical Support for more information

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## GENETIC OVERVIEW

Genetic Background

Generation

*Pax3<sup>Sp</sup>*

**Allele Type**

Spontaneous

**Gene Symbol**

*Pax3*

**Gene Name**

paired box 3

VIEW GENETICS

## RESEARCH APPLICATIONS

Mouse/Human Gene Homologs

Dermatology Research

Developmental Biology Research

Neurobiology Research

VIEW ALL RESEARCH APPLICATIONS

 Details

 Detailed Description

Mice homozygous for the splotch spontaneous mutation ( $Pax3^{Sp}$ ) die at E13 due to neural tube defects. Malformations of homozygous mutant embryos include rachischisis in the lumbosacral region and frequently in the region of the hindbrain. Heterozygous splotch mice show white spotting on the belly and occasionally on the back, feet, and tail. Splotch is a point mutation within intron 3 of the paired homeobox 3 ( $Pax3$ ) gene on mouse Chromosome 1. The mutation interferes with normal splicing of intron 3 and leads to at least 4 aberrantly spliced mRNAs with exon 4 deleted. The  $Pax3^{Sp}$  mutation also impairs homodimerization of the protein, a function associated with the octapeptide-encoding central segment of the gene. There are multiple alleles at this locus including splotch-delayed ( $Pax3^{Sp-d}$ , Stock No. 000565), which is similar to splotch but displays caudal rachischisis only.

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## Genetics

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+  $Pax3^{Sp}$

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## Disease/Phenotype

+ Disease Terms

+ Research Areas By Phenotype

+ Mammalian Phenotype Terms by Genotype

+ References

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## Technical Support

C O N T A C T   T E C H N I C A L   S U P P O R T

### Genotyping Protocols

[Genotyping resources and troubleshooting](#)

### Appearance

black with white spotting

Related Genotype:  $a/a Pax3^{Sp} /+$

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Q U E S T I O N S   A B O U T   T E R M S   O F   U S E

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- By Gene
- By Collection

All Related Strains



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