

B6.C3-*Spta1*^{sph}/BrkJ

Stock No: 000450 | B6-spherocytosis

 Congenic

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jaundice.

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GENETIC OVERVIEW

Genetic Background

Generation

Spta1^{sph}

Alele Type

Gene Symbol

Gene Name

Spontaneous

Spta1

spectrin alpha, erythrocytic 1

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RESEARCH APPLICATIONS

Cardiovascular Research

Hematological Research

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Details

Detailed Description

Homozygotes have spherocytic hemolytic anemia and neonatal jaundice. Erythrocytes have diminished biconcavity, a smaller than normal diameter, increased density, an appearance of exovesiculation, and a short lifespan. At birth homozygotes are very pale then become yellow during the first few hours of life and die generally within a day of being born. Although it is rare for a spherocytosis homozygote to survive beyond one day of birth on the C57BL/6J or WB/Re (Stock No. 000454) congenic background, homozygotes on the (C57BL/6J x WB/Re)F1 background are much more robust and the majority survive to adulthood. Those that make it to wean age are expected to have a reasonable lifespan, sometimes living as long as a year. Thus, most studies of this mutation in the adult have been done on the (C57BL/6J x WB/Re)F1 background. These homozygotes display diminished hematocrit, decreased red blood cell count, very elevated red blood cell protoporphyrin levels, increased erythropoiesis, reticulocytosis, hyperbilirubinaemia, cardiomegaly, hepatomegaly, and severe splenomegaly predominantly due to increased red pulp. Large areas of splenic necrosis and fibrosis develop. Thrombi can be found in the atrioventricular valves or within the atria of adults and infarcts have been observed in the myocardium and in the cerebrum, hippocampus, and cerebellum. Iron accumulates in the liver and kidneys, blood urea nitrogen levels become high, and hydronephrosis and membranoproliferative glomerulonephritis are found. By 2 to 3 months of age the urine becomes green due to hemosiderin and ferritin laden renal tubular cells and the urine later becomes red. Kaysser et al. posited that the likely cause of death in adult homozygotes is renal failure.

Development

Selected References

Genetics

Spta1^{sph}

Disease/Phenotype

Disease Terms

Research Areas By Phenotype

[+ Mammalian Phenotype Terms by Genotype](#)

[+ References](#)

[- 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

Sanger sequencing: [Spna1-Alternate 1](#)

[Genotyping resources and troubleshooting](#)

Citation

When using the B6-spherocytosis mouse strain in a publication, please [cite the originating article\(s\)](#) and include JAX stock #000450 in your Materials and Methods section.

Animal Health Reports

[Facility Barrier Level Descriptions](#)

Production of mice from cryopreserved embryos or sperm occurs in a maximum barrier room, [G200](#)

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Recovery

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