

## C57BL/6J-*Nek1*<sup>kat-2J</sup>/J

Stock No: **002854** | kidney, anemia and testis 2 Jackson

 Spontaneous Mutation

Typically mice are recovered in 10-14 weeks. Contact Customer Service to place an order or for more information.

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These mice are a model for Autosomal Recessive Polycystic Kidney Disease, Autosomal Recessive (ARPKD).

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## GENETIC OVERVIEW

Genetic Background

Generation

*Nek1*<sup>kat-2J</sup>

Alele Type

Gene Symbol

Gene Name

Spontaneous

*Nek1*

NIMA (never in mitosis gene a)-related expressed kinase 1

VIEW GENETICS

## RESEARCH APPLICATIONS

Developmental Biology Research

Hematological Research

Sensorineural Research

Internal/Organ Research

Neurobiology Research

Reproductive Biology Research

VIEW ALL RESEARCH APPLICATIONS

## BASE PRICE

Starting at:

\$2,854.50 Domestic price Cryo Recovery

V I E W   P R I C E   L I S T

### Details

#### Detailed Description

The *Nek1* gene encodes a serine/threonine kinase involved in cell cycle and cilia regulation. Mutations in this gene have been associated with short rib-polydactyly syndrome type II and familial amyotrophic lateral sclerosis (ALS). The *Nek1*<sup>kat-2J</sup> mutation is a single-base (G) insertion at position 966 that causes a frameshift and premature stop. The phenotype associated with the *Nek1*<sup>kat-2J</sup> allele is more severe than that of the *Nek1*<sup>kat</sup> allele. Mice homozygous for *Nek1*<sup>kat-2J</sup> all die before one year of age, a third of those that survive weaning die suddenly before 100 days of age, and the reported median survival age is 211 days. Mice homozygous for *Nek1*<sup>kat</sup> also have a shortened life expectancy with high pre-weaning mortality and a reported median survival of 286 days, but 22% were found to survive beyond 1 year. Both mutants are runted and have blunted noses with olfactory bulbs that are approximately half the normal size and lack most of the glomerular and external granular layers. In the brain there is dilation of the lateral and third ventricles, cerebral aqueduct and fourth ventricle along with large, fluid-filled cysts in the choroid plexus. Hydrocephalus occurs. Normochromic normocytic anemia is found and is more pronounced in *Nek1*<sup>kat-2J</sup> homozygotes, often detectable before weaning and progressing with age. By 7 to 8 months of age uremia with abnormally shaped erythrocytes can also be detected in the peripheral blood. The hematocrit levels of *Nek1*<sup>kat</sup> homozygotes is lower than normal in the young, but a significant drop occurs after approximately 200 days of age then continues to decrease. Abdominal distention occurs as a result of extensive renal enlargement. At 5 days of age microscopic examination fails to detect any morphologic change in the kidneys of either mutant, but fluid-filled cysts and dilated proximal tubules and Bowman spaces are found as early as 1 month of age in *Nek1*<sup>kat-2J</sup> homozygotes. The bilateral renal cystic disease progresses involving all levels of the nephron by 3 months of age, and after 6 months of age this progression becomes comparatively more rapid in the females. Disease progression includes growth of cysts and an increase in the number of cysts. *Nek1*<sup>kat</sup> homozygotes have similar renal cystic disease but the onset is slower relative to that in *Nek1*<sup>kat-2J</sup> homozygotes. (For morphologic details see Vogler et al., 1999.) Testicular hypoplasia with decreased spermatogenesis has been described for *Nek1*<sup>kat-2J</sup> homozygotes, and male sterility is a characteristic of both *Nek1* mutants. No histological abnormalities have been described in the ovaries and homozygous females can reproduce but litters are less frequent than from wildtype or heterozygous females. Focal portal bile duct proliferation and dilation have been found in the older *Nek1*<sup>kat-2J</sup> homozygotes. (Janaswami et al., 1997; Vogler et al., 1999.)

#### Development

#### Control Suggestions

### Genetics

#### *Nek1*<sup>kat-2J</sup>

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## – Disease/Phenotype

+ [Disease Terms](#)

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+ [Research Areas By Phenotype](#)

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+ [Mammalian Phenotype Terms by Genotype](#)

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+ [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

End Point Analysis: [Nek1 Endpoint](#)

[Genotyping resources and troubleshooting](#)

### Breeding Considerations

When maintaining a live colony, heterozygous mice may be bred to wildtype siblings or to C57BL/6J inbred mice (Stock No. [000664](#)). Homozygous males are infertile. Homozygotes have a shortened life expectancy.

[Additional Breeding and Husbandry Support](#)

### Citation

When using the kidney, anemia and testis 2 Jackson mouse strain in a publication, please [cite the originating article\(s\)](#) and include JAX stock #002854 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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## – Pricing & Availability



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Cryo  
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## DomesticInternational

Pricing effective for USA, Canada and Mexico shipping destinations

### CRYORECOVERY - DOMESTIC PRICING

SERVICE/PRODUCT	DESCRIPTION	PRICE
<a href="#">Cryo Recovery</a>	Heterozygous or Wild-type for Nek1<kat-2J>	\$2,854.50

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