

# Nefl-N98S

系統名	C57BL/6Smoc- <i>Nefl</i> <sup>em1(N98S)Smoc</sup>
SMOC番号	NM-KI-200251
維持形態	Embryo cryopreservation

## 遺伝子の概要

Gene Symbol Nefl	Synonyms	Nfl, NF-L, NF68, CMT2E, AI847934
	NCBI ID	<a href="#">18039</a>
	MGI ID	<a href="#">97313</a>
	Ensembl ID	<a href="#">ENSMUSG00000022055</a>
	Human Ortholog	NEFL

## 説明

These mice carry a p.N98S mutation of Nefl gene.

**応用分野:** Peroneal muscular atrophy

\*Literature published using this strain should indicate: Nefl-N98S mice (Cat. NO. NM-KI-200251) were purchased from Shanghai Model Organisms Center, Inc..

## 病気の予測

Charcot-Marie-Tooth Disease Type 2E	表現型	<a href="#">MGI:5751579</a>
	参考文献	Adebola AA, et al., Neurofilament light polypeptide gene N98S mutation in mice leads to neurofilament network abnormalities and a Charcot-Marie-Tooth Type 2E phenotype. Hum Mol Genet. 2015 Apr 15;24(8):2163-74

## 表現型データ

No data