

Resource Summary Report

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B6;129S4-Gba1^{tm1Rlp}/Mmnc

RRID:MMRRC_000117-UNC

Type: Organism

Proper Citation

RRID:MMRRC_000117-UNC

Organism Information

URL: https://www.mmrrc.org/catalog/sds.php?mmrrc_id=117

Proper Citation: RRID:MMRRC_000117-UNC

Description: Mus musculus with name B6;129S4-Gba1^{tm1Rlp}/Mmnc from MMRRC.

Species: Mus musculus

Notes: Research areas: Immunology and Inflammation; Mutation Type: Targeted Mutation ; Collection:

Phenotype: enlarged liver [MP:0000599]| reddish skin [MP:0001190]| wrinkled skin [MP:0001211]| hyperkeratosis [MP:0001242]| decreased body weight [MP:0001262]| postnatal lethality [MP:0002082]| increased liver weight [MP:0002981]| increased spleen weight [MP:0004952]| postnatal lethality [MP:0011085]

Affected Gene: Gba1

Catalog Number: 000117-UNC

Background: Targeted Mutation

Database: Mutant Mouse Resource and Research Center (MMRRC)

Database Abbreviation: MMRRC

Source References: [PMID:11994410](#), [PMID:9482915](#)

Alternate IDs: MMRRC_117-UNC, MMRRC_000117, MMRRC_117

Organism Name: B6;129S4-*Gba1*^{tm1Rlp}/Mmnc

Record Creation Time: 20230308T054750+0000

Record Last Update: 20250419T222355+0000

Ratings and Alerts

No rating or validation information has been found for B6;129S4-*Gba1*^{tm1Rlp}/Mmnc.

No alerts have been found for B6;129S4-*Gba1*^{tm1Rlp}/Mmnc.

Data and Source Information

Source: [Integrated Animals](#)

Source Database: Mutant Mouse Resource and Research Center (MMRRC)

Usage and Citation Metrics

We found 7 mentions in open access literature.

Listed below are recent publications. The full list is available at [FDI Lab - SciCrunch.org](#).

Nam Y, et al. (2024) DJ-1 protects cell death from a mitochondrial oxidative stress due to GBA1 deficiency. *Genes & genomics*, 46(5), 519.

Mahoney-Crane CL, et al. (2023) Neuronopathic GBA1L444P Mutation Accelerates Glucosylsphingosine Levels and Formation of Hippocampal Alpha-Synuclein Inclusions. *The Journal of neuroscience : the official journal of the Society for Neuroscience*, 43(3), 501.

Kuo SH, et al. (2022) Mutant glucocerebrosidase impairs α -synuclein degradation by blockade of chaperone-mediated autophagy. *Science advances*, 8(6), eabm6393.

Li H, et al. (2019) Mitochondrial dysfunction and mitophagy defect triggered by heterozygous GBA mutations. *Autophagy*, 15(1), 113.

Yun SP, et al. (2018) α -Synuclein accumulation and GBA deficiency due to L444P GBA mutation contributes to MPTP-induced parkinsonism. *Molecular neurodegeneration*, 13(1), 1.

Migdalska-Richards A, et al. (2017) The L444P *Gba1* mutation enhances alpha-synuclein induced loss of nigral dopaminergic neurons in mice. *Brain : a journal of neurology*, 140(10), 2706.

Fishbein I, et al. (2014) Augmentation of phenotype in a transgenic Parkinson mouse heterozygous for a Gaucher mutation. *Brain : a journal of neurology*, 137(Pt 12), 3235.