



GBA D409V KI Mouse Model

GBA D409V KI mice have a knock in of the D409V mutation in the murine GBA gene at exon 10, which corresponds to the D409V mutation in the mature human GBA protein. The gene is driven by the murine GBA promoter. Mice additionally contain loxP sites flanking exons 6 to 8 of the GBA gene, so it is possible to knockout this gene segment.

Pathological characteristics of GBA D409V KI mice:

GBA D409V KI mice

- Severely decreased GCCase levels in the brain and liver at 4 months
- Severely increased glucosylsphingosine levels in the brain and liver at 4 months
- Severely increased α -synuclein levels at 12 months

GBA D409V/+ mice (12 months of age)

- Highly reduced GCCase levels in the hippocampus and cortex
- Increased activated microglia and astrocytosis levels in the hippocampus
- Decreased synaptophysin and vAChT levels in the hippocampus
- Increased CHAT levels in the hippocampus
- Neither α -synuclein aggregates nor pSer129 α -synuclein in the hippocampus
- Cognitive impairments in the Morris Water Maze and Y-Maze
- No motor deficits

Due to the association of the GBA1 gene to Parkinson's disease, the model is also valuable for crossbreeding studies with Parkinson's disease mice to model GBA-associated Parkinson's disease.

References:

Clarke et al., 2019 Oct;129:104502. doi:10.1016/j.neuint.2019.104502.
Burballa et al., 2019 Oct 16;11(514):eaau6870. doi: 10.1126/scitranslmed.aau6870.

Scantox
Discovery

Important note:

Representative data are shown throughout this document. However, biological variability might cause deviations from shown data.

Scantox Group, HQ
Hestehavevej 36A, Ejby
DK – 4623 Lille Skensved
clientservice@scantox.com
www.scantox.com
+45 5686 1500

© Scantox A/S
Scantox is a registered
trademark of Scantox A/S.

126-37-GaucherGBA-EN (04/2024)

