These knock-in mice carry a H67D mutation in the mouse Hfe (hemochromatosis) gene that is homologous to a human H63D mutation found at higher frequencies in neurodegenerative diseases. Brain concentrations of iron aren’t significantly modified in these mice, but proteins that manage the iron are.

Donating Investigator
James R Connor, The Pennsylvania State University
The *Hfe* (hemochromotosis) gene expresses a protein commonly associated with iron homeostasis. Increasing evidence suggests a potential role in neurodegenerative diseases.

These knock-in mice carry a H67D mutation in the mouse *Hfe* gene that is homologous to a human H63D mutation found at higher frequencies in neurodegenerative diseases. At 6 and 12 months of age, mutant mice have higher body weight than wildtype mice. Homozygotes have altered brain and hepatic iron homeostasis, and increased oxidative stress and gliosis in the brain.

Brain concentrations of iron aren't significantly modified in these mice, but proteins that manage the iron are. Six-month-old H67D mice have increased levels of HFE and H-ferritin (*FTH1*) expression. At 12 months, H67D mice have increased H- and L-ferritin (*FTL1*) but decreased transferrin (*TRF*) expression, suggesting increased iron storage and decreased iron mobilization.

When combined with a G93A mutant SOD1 (superoxide dismutase 1, soluble) mouse model for amyotrophic lateral sclerosis (ALS) (see Stock No. 002726), mice show a shorter survival time and an accelerated disease progression.
Genotyping Protocols
End Point Analysis: Hfe-EP
Genotyping resources and troubleshooting

Breeding Considerations
Heterozygotes and homozygotes are viable and fertile.

Additional Breeding and Husbandry Support

Citation
When using the B6;129X1-Hfe<tm1.1Jrco> /J mouse strain in a publication, please cite the originating article(s) and include JAX stock #023025 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

Pricing & Availability

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

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<tr>
<th>SERVICE/PRODUCT</th>
<th>DESCRIPTION</th>
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<tr>
<td>Cryo Recovery</td>
<td>Heterozygous for Hfe&lt;tm1.1(H67D)Jrco&gt;</td>
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