Frataxin levels and pancreatic iron levels are reduced in these knock-in/knockout mice. However, unlike Friedreich's Ataxia patients, no detectable change in GAA repeat size is observed. This mutant mouse may be useful in studies of Friedreich's Ataxia.

Donating Investigator
IMR Colony, The Jackson Laboratory

<table>
<thead>
<tr>
<th>GENETIC OVERVIEW</th>
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<th>Fxn&lt;sup&gt;tm1Mkn&lt;/sup&gt;</th>
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<td><strong>Gene Symbol</strong></td>
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RESEARCH APPLICATIONS

Neurobiology Research
These frataxin knock-in/knockout (KIKO) mice harbor one allele of the frataxin \((\text{GAA})_{230}\) expansion mutation \((Fxn^{tm1Pand})\) on one chromosome, and one allele of the frataxin exon 4-deleted mutation \((Fxn^{tm1Mkn})\) on the homologous chromosome. KIKO mice are viable and fertile. Analysis of frataxin levels in tissues from KIKO mice demonstrate a reduction of frataxin to 25-36% of wildtype controls. KIKO animals up to 1 year of age perform equivalent to wildtype controls on rotarod test. Total iron concentrations were similar in all tested tissues of KIKO and wildtype mice except in pancreas: KIKO mice demonstrate lower iron levels in pancreatic tissues. No iron deposits and only mild collagen staining around the vessels of the heart were observed in both year old KIKO mice and wildtype controls. In contrast to Friedreich's Ataxia patients, no detectable change in GAA repeat size was found over six studied generations. Moreover, no evidence of somatic cell instability was noted as GAA repeat expansion size was the same in all analyzed tissues. However, characterization of KIKO mice performed at The Jackson Laboratory revealed that starting at 6 months of age, these animals exhibit an abnormal "weaving" gait when subjected to a forced treadmill walk. This phenotype occurs with increasing penetrance as the mice age. This mutant mouse may be useful in studies of Friedreich's Ataxia.

Importation of this model was supported by the Friedreich's Ataxia Research Alliance (FARA).
Disease/Phenotype

Disease Terms

Research Areas By Phenotype

Mammalian Phenotype Terms by Genotype

References

Technical Support

Genotyping Protocols
Standard PCR: Fxnalternate3
Standard PCR: Fxnalternate1
Genotyping resources and troubleshooting

Breeding Considerations

When maintaining the live KIKO colony, animals harboring one allele of the frataxin (GAA)230 expansion mutation (Fxntm1Pand) on one chromosome, and one allele of the frataxin exon 4-deleted mutation (Fxntm1Mkn) on the homologous chromosome may be bred together.

Additional Breeding and Husbandry Support

Citation

When using the KIKO mouse strain in a publication, please cite the originating article(s) and include JAX stock #014162 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.

Cryo Recovery

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

Domestic

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<th>SERVICE/PRODUCT</th>
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Related Products and Services

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Terms Of Use

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Email: TechTran@jax.org

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All Related Strains