129S6-Scn5a$^{tm(R222Q)Rdn}$/Vu

VANDERBILT UNIVERSITY labnodes

Allele #1 human cDNA for SCN5A replaces exon 2 of mouse Scn5a. Insertion at this location disrupts expression of mouse ortholog. This mouse is heterozygous for Scn5a R222Q. Recombinase mediated cassette exchange was used to insert human SCN5A between loxP sites. Expression of human SCN5A is dependent on native mouse promoters for SCN5A.

Keywords: R222Q | Scn5a | Rdn | RMCE

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Mouse Information

Common Name	RQH-SCN5A-SV129
VCMR ID	YI
Date Cryopreserved	2018-04-10
Method of Cryopreservation	Sperm
Trial IVF % Fertilization	31.00%

Genetic Alteration

Mutation #1: Targeted Mutagenesis

Allele	Name: sodium channel, voltage-gated, type V, alpha; targeted mutation, Dan M Roden Symbol: Scn5a$^{tm(R222Q))Rdn}$
Zygosity at cryopreservation	Heterozygous
PCR Genotyping Protocol	Genotyping_Protocol_YI.pdf
Citations	**Publication** SCN5A variant R222Q generated abnormal changes in cardiac sodium current and action potentials in murine myocytes and Purkinje cells. (2019) *Heart Rhythm* **16**: 1676-1685 (Added 6/14/2019) PMID: 31125670

Background Strain Information

Strain Type	Congenic Strain

Chimera/Founder Genetic Background	129S6/SvEvTac
Cryopreservation Strain Background (VCMR)	129S6/SvEvTac
Viability and Fertility Data	Homozygous R222Q pups are viable, but are usually smaller than normal and do not live past 1 month of age. Most die before they are weaned. Many are probably eaten by the mother.