

Bckdk-KO

Nomenclature	C57BL/6Smoc- <i>Bckdk</i> ^{em1Smoc}
Cat. NO.	NM-KO-2101591
Strain State	Sperm cryopreservation

Gene Summary

Gene Symbol Bckdk	Synonyms	AI327402
	NCBI ID	12041
	MGI ID	1276121
	Ensembl ID	ENSMUSG00000030802
	Human Ortholog	BCKDK

Model Description

exon 9-12 of Bckdk gene was deleted to generate Bckdk knockout mice.

*Literature published using this strain should indicate: Bckdk-KO mice (Cat. NO. NM-KO-2101591) were purchased from Shanghai Model Organisms Center, Inc..

Disease Connection

Autism Spectrum Disorder	Phenotype(s)	MGI:5829468
	Reference(s)	Tarlungeanu DC, Deliu E, Dotter CP, Kara M, Janiesch PC, Scalise M, Galluccio M, Tesulov M, Morelli E, Sonmez FM, Bilguvar K, Ohgaki R, Kanai Y, Johansen A, Esharif S, Ben-Omran T, Topcu M, Schlessinger A, Indiveri C, Duncan KE, Caglayan AO, Gunel M, Gleeson JG, Novarino G, Impaired Amino Acid Transport at the Blood Brain Barrier Is a Cause of Autism Spectrum Disorder. Cell. 2016 Dec 01;167(6):1481-1494.e18

Branched-Chain Keto Acid Dehydrogenase Kinase Deficiency	Phenotype(s)	MGI:3699323
	Reference(s)	Novarino G, El-Fishawy P, Kayserili H, Meguid NA, Scott EM, Schroth J, Silhavy JL, Kara M, Khalil RO, Ben-Omran T, Ercan-Sencicek AG, Hashish AF, Sanders SJ, Gupta AR, Hashem HS, Matern D, Gabriel S, Sweetman L, Rahimi Y, Harris RA, State MW, Gleeson JG, Mutations in BCKD-kinase lead to a potentially treatable form of autism with epilepsy. Science. 2012 Oct 19;338(6105):394-7

Validation Data

No data