Bckdk-KO

品系全名	C57BL/6Smoc-*Bckdk*^{em1Smoc}
目录号	NM-KO-2101591
品系状态	精子冻存

基因信息

基因名 **Bckdk**	**基因曾用名**	AI327402
	NCBI ID	12041
	MGI ID	1276121
	Ensembl ID	ENSMUSG00000030802
	人类同源基因	BCKDK
	人类同源基因关联疾病	支链酮酸脱氢酶激酶缺乏症、智力低下

品系描述

通过敲除Bckdk基因exon 9-12，建立Bckdk基因敲除小鼠模型。
*使用本品系发表的文献需注明: Bckdk-KO mice (Cat. NO. NM-KO-2101591) were purchased from Shanghai Model Organisms Center, Inc..

疾病预测

支链酮酸脱氢酶激酶缺乏症 **Branched-Chain Keto Acid Dehydrogenase Kinase Deficiency**	**近似模型的表型**	MGI:3699323
	参考文献	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

自闭症谱系障碍 **Autism Spectrum Disorder**	**近似模型的表型**	MGI:5829468
	参考文献	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

验证数据

暂无数据