

# Bckdk-KO

系統名	C57BL/6Smoc- <i>Bckdk</i> <sup>em15moc</sup>
SMOC番号	NM-KO-2101591
維持形態	Sperm cryopreservation

## 遺伝子の概要

Gene Symbol Bckdk	Synonyms	AI327402
	NCBI ID	<a href="#">12041</a>
	MGI ID	<a href="#">1276121</a>
	Ensembl ID	<a href="#">ENSMUSG00000030802</a>
	Human Ortholog	BCKDK

## 説明

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..

## 病気の予測

Autism Spectrum Disorder	表現型	<a href="#">MGI:5829468</a>
	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

<b>Branched-Chain Keto Acid Dehydrogenase Kinase Deficiency</b>	<b>表現型</b>	<a href="#">MGI:3699323</a>
	<b>参考文献</b>	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

### 表現型データ

No data