abcam

Product datasheet

Human USH1C peptide ab45568

概述

产品名称	人USH1C肽

描述

性质	Synthetic

氨基酸序列

种属	Human
序列	DRKVAREFRHKVD-C
氨基酸	2 to 14

技术指标

Our Abpromise guarantee covers the use of **ab45568** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

应用	Blocking - Blocking peptide for Anti-USH1C antibody (ab19045)
形式	Liquid

制备和贮存

稳定性和存储	Shipped at 4°C. Upon delivery aliquot and store at -20°C. Avoid freeze / thaw cycles.

常规信息

功能	May be involved in protein-protein interaction.
组织特异性	Expressed in small intestine, colon, kidney, eye and weakly in pancreas. Expressed also in vestibule of the inner ear.
疾病相关	Defects in USH1C are the cause of Usher syndrome type 1C (USH1C) [MIM:276904]; also known as Usher syndrome type I Acadian variety. USH is a genetically heterogeneous condition characterized by the association of retinitis pigmentosa and sensorineural deafness. Age at onset and differences in auditory and vestibular function distinguish Usher syndrome type 1 (USH1), Usher syndrome type 2 (USH2) and Usher syndrome type 3 (USH3). USH1 is characterized by profound congenital sensorineural deafness, absent vestibular function and

	prepubertal onset of progressive retinitis pigmentosa leading to blindness. Defects in USH1C are the cause of deafness autosomal recessive type 18 (DFNB18) [MIM:602092]. DFNB18 is a form of sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information.
序列相似性	Contains 3 PDZ (DHR) domains.
结构域	The PDZ domain 1 mediates interactions with USH1G/SANS and SLC4A7.

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