abcam

Product datasheet

Human FMRP (phospho S499) peptide ab48658

描述

产**品名称** 人FMRP (phospho S499)多肽

纯度 > 70 % HPLC.

70 - 90% by HPLC

无动物成分 No

性质 Synthetic

种属 Human

技术指标

Our Abpromise guarantee covers the use of ab48658 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

形式 Liquid

补充说明 - First try to dissolve a small amount of peptide in either water or buffer. The more charged

residues on a peptide, the more soluble it is in aqueous solutions.

- If the peptide doesn't dissolve try an organic solvent e.g. DMSO, then dilute using water or

buffer.

- Consider that any solvent used must be compatible with your assay. If a peptide does not

dissolve and you need to recover it, lyophilise to remove the solvent.

- Gentle warming and sonication can effectively aid peptide solubilisation. If the solution is

cloudy or has gelled the peptide may be in suspension rather than solubilised.

- Peptides containing cysteine are easily oxidised, so should be prepared in solution just prior

to use.

制备和贮存

稳定性和存储 Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw

cycles.

Information available upon request.

常规信息

1

功能 Translation repressor. Component of the CYFIP1-EIF4E-FMR1 complex which binds to the

mRNA cap and mediates translational repression. In the CYFIP1-EIF4E-FMR1 complex this subunit mediates translation repression (By similarity). RNA-binding protein that plays a role in intracellular RNA transport and in the regulation of translation of target mRNAs. Associated with polysomes. May play a role in the transport of mRNA from the nucleus to the cytoplasm. Binds strongly to poly(G), binds moderately to poly(U) but shows very little binding to poly(A) or poly(C).

组织特异性 Highest levels found in neurons, brain, testis, placenta and lymphocytes. Also expressed in

epithelial tissues and at very low levels in glial cells.

疾病相关 Defects in FMR1 are the cause of fragile X syndrome (FRAX) [MIM:300624]. Fragile X syndrome

is a common genetic disease (has a prevalence of one in every 2000 children) which is characterized by moderate to severe mental retardation, macroorchidism (enlargement of the testicles), large ears, prominent jaw, and high-pitched, jocular speech. The defect in most fragile X syndrome patients results from an amplification of a CGG repeat region which is directly in front

of the coding region.

Defects in FMR1 are the cause of fragile X tremor/ataxia syndrome (FXTAS) [MIM:300623]. In FXTAS, the expanded repeats range in size from 55 to 200 repeats and are referred to as 'premutations'. Full repeat expansions with greater than 200 repeats results in fragile X mental retardation syndrome [MIM:300624]. Carriers of the premutation typically do not show the full fragile X syndrome phenotype, but comprise a subgroup that may have some physical features of fragile X syndrome or mild cognitive and emotional problems.

Defects in FMR1 are the cause of premature ovarian failure syndrome type 1 (POF1)

[MIM:311360]. An ovarian disorder defined as the cessation of ovarian function under the age of 40 years. It is characterized by oligomenorrhea or amenorrhea, in the presence of elevated levels

of serum gonadotropins and low estradiol.

序列相似性 Belongs to the FMR1 family.

Contains 2 KH domains.

翻译后修饰 Phosphorylated on several serine residues.

细胞定位 Cytoplasm. Nucleus > nucleolus.

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