abcam

Product datasheet

# Recombinant Human FMRP protein ab114231

画像数 1

## 製品の詳細

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| **製品名** | Recombinant Human FMRP protein |
| **発現系** | Wheat germ |
| **アクセッション番号** | Q06787 |
| **タンパク質長** | Protein fragment |
| **Animal free** | No |
| **由来** | Recombinant |
| **生物種** | Human |
| **配列** | ATKDTFHKIKLDVPEDLRQMCAKEAAHKDFKKAVGAFSVTYD PENYQLVI LSINEVTSKRAHMLIDMHFRSLRTKLSLIMRNEEASKQLESS RQLASRFH |
| **予測される分子量** | 37 kDa including tags |
| **領域** | 121 to 220 |

## 特性

Our **Abpromise guarantee** covers the use of **ab114231** in the following tested applications.

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

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| **アプリケーション** | ELISA<br>SDS-PAGE<br>Western blot |
| **製品の状態** | Liquid |
| **備考** | Protein concentration is above or equal to 0.05 mg/ml |

## 前処理および保存

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| **保存方法および安定性** | Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles.<br>pH: 8.00<br>Constituents: 0.3% Glutathione, 0.79% Tris HCl |

## 関連情報

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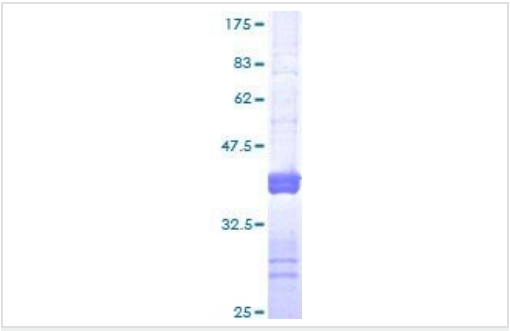
| **機能** | Translation repressor. Component of the CYFIP1-EIF4E-FMR1 complex which binds to the mRNA cap and mediates translational repression. In the CYFIP1-EIF4E-FMR1 complex this subunit mediates translation repression (By similarity). RNA-binding protein that plays a role in intracellular RNA transport 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). |
| **組織特異性** | Highest levels found in neurons, brain, testis, placenta and lymphocytes. Also expressed in epithelial tissues and at very low levels in glial cells. |
| **関連疾患** | 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.<br>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.<br>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. |
| **配列類似性** | Belongs to the FMR1 family.<br>Contains 2 KH domains. |
| **翻訳後修飾** | Phosphorylated on several serine residues. |
| **細胞内局在** | Cytoplasm. Nucleus > nucleolus. |

## 画像




SDS-PAGE - Recombinant Human FMRP protein (ab114231)

12.5% SDS-PAGE showing ab114231 at approximately 36.63kDa stained with Coomassie Blue.

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