

Recombinant Human FMRP protein ab132093

1 Image

Description	
Product name	Recombinant Human FMRP protein
Expression system	Wheat germ
Accession	<u>Q8IXW7</u>
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	MEELVVEVRGSNGAFYKAFVKDVHEDSITVAFENNWQPD RQIPFHDVRFPPVGYNKDINESDEVEVYSRANEKEPCCWWLAKVRMIKG EFYVIEYAACD ATYNEVTIERLRSVNPNKPATKDTFHKIKLDVPEDLRQMC AKEAAHKDF KKAVGAFSVTYDPENYQLVILSINEVTSKRAHMLIDMHFRS LRTKLSLIM RNEEASKQLESSRQLASRFHEQFVREDLMGLAIGTHGANI QQARKVPGV TAIDLDEDTCTFHIYGEDQDAVKKARSFLEFAEDVIQVPRN LVGLKI
Predicted molecular weight	58 kDa including tags
Amino acids	1 to 297
Tags	GST tag N-Terminus

Specifications	
Our Abpromise guarantee covers the use of ab132093 in the following tested applications.	
The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.	
Applications	Western blot SDS-PAGE ELISA
Form	Liquid

Preparation and Storage

Stability and Storage

Shipped on dry ice. Upon delivery aliquot and store at -80°C. Avoid freeze / thaw cycles.

pH: 8.00

Constituents: 0.31% Glutathione, 0.79% Tris HCl

General Info

Function

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).

Tissue specificity

Highest levels found in neurons, brain, testis, placenta and lymphocytes. Also expressed in epithelial tissues and at very low levels in glial cells.

Involvement in disease

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.

Sequence similarities

Belongs to the FMR1 family.

Contains 2 KH domains.

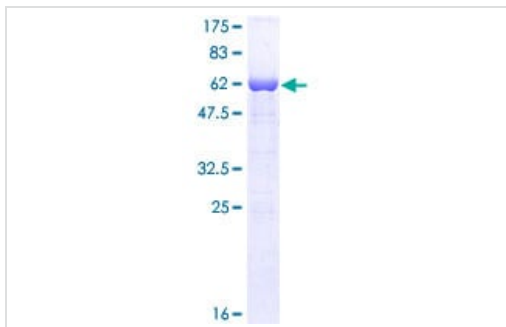
Post-translational modifications

Phosphorylated on several serine residues.

Cellular localization

Cytoplasm. Nucleus > nucleolus.

Images



12.5% SDS-PAGE analysis of ab132093 stained with Coomassie Blue.

SDS-PAGE - Recombinant Human FMRP protein
(ab132093)

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