

Recombinant Human FMR1 Mutant (I304N) Protein, Myc/DDK-tagged

Cat. No. FMR1-058H Lot. No. (See product label)

SPECIFICATION

Product Overview	Purified mutant recombinant protein of Human fragile X mental retardation 1 (FMR1), transcript variant ISO1, mutation at(I304N) with a Myc/DDK tag was expressed in HEK293
Species	Human
Source	HEK293
Description	The protein encoded by this gene binds RNA and is associated with polysomes. The encoded protein may be involved in mRNA trafficking from the nucleus to the cytoplasm. A trinucleotide repeat (CGG) in the 5' UTR is normally found at 6-53 copies, but an expansion to 55-230 repeats is the cause of fragile X syndrome. Expansion of the trinucleotide repeat may also cause one form of premature ovarian failure (POF1). Multiple alternatively spliced transcript variants that encode different protein isoforms and which are located in different cellular locations have been described for this gene.
Molecular Mass	71 kDa
Purity	> 80% as determined by SDS-PAGE and Coomassie blue staining
Stability	Stable for at least 12 months from receipt of products under proper storage and handling conditions. Avoid repeated freeze-thaw cycles.

 Tel: 1-631-559-9269 1-516-512-3133

 Email: info@creative-biomart.com  Fax: 1-631-938-8127

 45-1 Ramsey Road, Shirley, NY 11967, USA

Storage	Store at -80 centigrade after receiving vials.
Concentration	> 50 µg/mL as determined by microplate Bradford method
Storage Buffer	25 mM Tris.HCl, pH 7.3, 100 mM glycine, 10% glycerol

GENE INFORMATION

Gene Name	FMR1 FMRP translational regulator 1 [Homo sapiens (human)]
Official Symbol	FMR1
Synonyms	FMR1; FMRP translational regulator 1; POF; FMRP; POF1; FRAXA; synaptic functional regulator FMR1; fragile X mental retardation 1; fragile X mental retardation protein 1; truncated FMRP
Gene ID	2332
mRNA Refseq	NM_002024
Protein Refseq	NP_002015
MIM	309550
UniProt ID	Q06787

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