

BACKGROUND

The *FMR1* gene is involved in three different syndromes: the Fragile X syndrome, premature ovarian failure (POF) and the Fragile X-associated tremor/ataxia syndrome (FXTAS) at older age.¹ Fragile X syndrome is caused by an expanded CGG repeat above 200 units in the *FMR1* gene resulting in the absence of the *FMR1* mRNA and protein. The FMR1 protein is a multifunctional protein, and proposed to act as a regulator of mRNA transport and/or translation. FMR1 protein binds RNA and is associated with polysomes. It binds strongly to poly(G), binds moderately to poly(U) but shows very little binding to poly(A) or poly(C). It may be involved in mRNA trafficking from the nucleus to the cytoplasm.² A trinucleotide repeat (CGG) in the 5' UTR is normally found at 6-53 copies. It was shown that FMR1 plays a role in synaptic maturation and function. POF and FXTAS are found in individuals with an expanded repeat between 50 and 200 CGGs and are associated with increased *FMR1* mRNA levels. The presence of elevated *FMR1* mRNA in all patients suggests that these syndromes may represent a gain-of-function effect from the elevated message levels. The level of *FMR1* mRNA is in fragile balance and is therefore critical for normal functioning.³

References:

1. Coffee, B. et al: Nature Genetics 22:98-101, 1999
2. Ashley, C.T. et al: Science 262:563-6, 1993
3. Crawford, D.C. et al: Genet. In Med. 3:359-71, 2001

TECHNICAL INFORMATION

Source:

FMR1 antibody is a mouse monoclonal antibody raised against purified recombinant human FMR1 fragments expressed in *E. coli*.

Specificity and Sensitivity:

This antibody detects endogenous FMR1 proteins without cross-reactivity with other related proteins.

Storage Buffer: PBS and 30% glycerol

Storage:

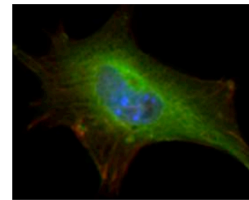
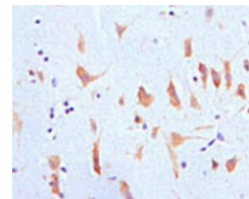
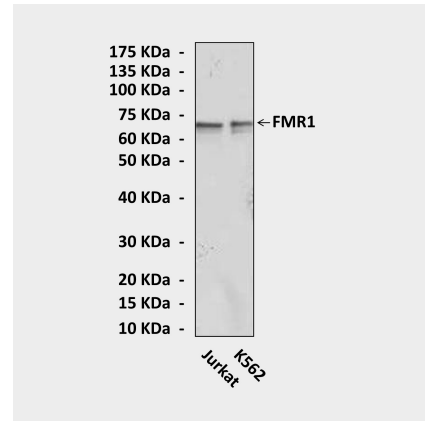
Store at -20°C for at least one year. Store at 4°C for frequent use. Avoid repeated freeze-thaw cycles.

APPLICATIONS

Application:	*Dilution:
WB	1:1000
IP	1:50
IHC	1:200
ICC	1:200
FACS	n/d

**Optimal dilutions must be determined by end user.*

QUALITY CONTROL DATA



Top: Western Blot detection of FMR1 proteins in Jurkat and K562 cell lysates using FMR1 Antibody.
Middle: This antibody stains paraffin-embedded human brain tissue in immunohistochemical analysis.
Bottom: It also stains NIH3T3 cells in confocal immunofluorescent testing (FMR1 Antibody: Green; actin filament: Red; and DRAQ5 DNA dye: Blue).

