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Rabbit Anti-AFF2 antibody

SL11703R

Product Name:	AFF2
Chinese Name:	脆性X综合征相关蛋白AFF2抗体
Alias:	FMR2; AF4/FMR2 family member 2; AF4/FMR2 family, member 2; AFF2; AFF2_HUMAN; FMR2; FMR2P; Fragile X E mental retardation syndrome protein; fragile X mental retardation 2; Fragile X mental retardation 2 protein; fragile X mental retardation gene associated with FRAXE; FRAXE; mild or borderline mental retardation; MRX2; OX19; Protein FMR-2; Protein Ox19.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Dog,Pig,Cow,Horse,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	145kDa
Cellular localization:	The nucleus
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human AFF2:1-80/1311
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed:	PubMed
Product Detail:	FMR2 is a 1311 amino acid nuclear protein belonging to the AF4 family. Expressed in the brain, placenta and lung, FMR2 exists as two isoforms produced by alternative splicing. Defects in the gene that encodes FMR2 have been found to be a cause of

FRAXE, an X-linked form of mental retardation. Individuals expressing the FRAXE site also have more than two-hundred copies of a GCC repeat adjacent to CpG island, compared to six to thirty-five copies of the GCC repeat in a normal individual. It is believed that loss of FMR2 expression causes this GCC expansion of the FRAXE site.

Function:
RNA-binding protein. Might be involved in alternative splicing regulation through an interaction with G-quartet RNA structure.

Subcellular Location:
Nucleus speckle. When splicing is inhibited, accumlates in enlarged speckles.

Tissue Specificity:
Brain (most abundant in hippocampus and amygdala), placenta and lung.

DISEASE:
Defects in AFF2 are the cause of fragile X-E mental retardation syndrome (FRAXE) [MIM:309548]. FRAXE is an X-linked form of mental retardation. Loss of FMR2 expression is correlated with FRAXE CCG(N) expansion. Normal individuals have 6-35 copies of the repeat, whereas cytogenetically positive, developmentally delayed males have more than 200 copies and show methylation of the associated CPG island.

Similarity:
Belongs to the AF4 family.

SWISS:
P51816

Gene ID:
2334

Database links:

Entrez Gene: 2334 Human

Omim: 309548 Human

SwissProt: P51816 Human

Unigene: 496911 Human

Important Note:
This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

	脆性X综合症，又称马丁－贝尔综合症，是一种遗传疾病。该综合症可以导致一系列的特征性症状，包括生理、智力、情绪、以及行为上的异常。症状的轻重各有不同。该疾病伴随着X染色体上一个简单的三核苷酸基因序列（CGG）的扩增。这种扩增导致了一种称为FMR- 1的蛋白质无法在病人体内表达，而该蛋白质是神经的正常发育必不可少的。 根据CGG重复序列的长度，目前普遍认可将脆性X综合症分为四种类型：正常人（含有19－31个CGG重复序列），前突变者（含有55－200个CGG重复序列），全突变者（含有200个以上的CGG重复序列），过渡型，又称“灰色区域型”（含有40－60个重复）。脆性X综合征这是一种导致智力低下的遗传疾病，是导致人群中智力低下的第二大病因——仅次于21三体综合症。