

Product datasheet

Recombinant Human MeCP2 protein (His tag) ab196070

[1 Image](#)

Description

Product name	Recombinant Human MeCP2 protein (His tag)	
Purity	> 95 % SDS-PAGE.	
Endotoxin level	< 1.000 Eu/μg	
Expression system	HEK 293 cells	
Accession	<u>P51608</u>	
Protein length	Full length protein	
Animal free	No	
Nature	Recombinant	
Species	Human	
Sequence	MVAGMLGLREEKSEDQDLQGLKDKPLKFKKVKKDKKEE KEGKHEPVQPSA HHSAPAEAGKAETSEGSGSAPAVPEASASPQRRSIIR DRGPMYDDPTL PEGWTRKLRKSRGSRGKYDVYLINPQGKAFRSKVELIA YFEKVGDTSL DPNDFDFTVTGRGSPSRREQPPKKPKSPKAPGTGRGR GRPKGSGTTRPK AATSEGVQVKRVLEKSPGKLLVKMPFQTSPGGKAEGGG ATTSTQVMVIKR PGRKRKAEADPQAIPKKRGRKPGSVVAAAAAAEAKKKAV KESSIRSVQETV LPIKKRKTRETVSIEVKEVVKPLLVSTLGEKSGKGLKTCKS PGRKSKESS PKGRSSSASSPPKKEHHHHHHHSESPKAPVLLPPLPPP PPEPESSEDPT SPPEPQDLSSSVCKEEKMPRGGSLSDGCPKEPAKTQP AVATAATAAEKY KHRGEGERKDMSSSMRPNREEPVDSRTPVTERVSVDH HHHHH	
Predicted molecular weight	53 kDa including tags	
Amino acids	1 to 486	

Tags His tag C-Terminus

Specifications

Our **Abpromise guarantee** covers the use of **ab196070** in the following tested applications.

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

Applications	HPLC SDS-PAGE
Form	Lyophilized

Preparation and Storage

Stability and Storage Shipped at 4°C. The lyophilized protein is stable for a few weeks at room temperature. Upon delivery aliquot. Store at -20°C. Avoid freeze / thaw cycle.

pH: 8.00

Constituents: 0.87% Sodium chloride, 99% Tris

Reconstitution Always centrifuge tubes before opening. Do not mix by vortex or pipetting. It is not recommended to reconstitute to a concentration less than 100 µg/ml. Dissolve the lyophilized protein in 1X PBS. Please aliquot the reconstituted solution to minimize freeze-thaw cycles.

General Info

Function Chromosomal protein that binds to methylated DNA. It can bind specifically to a single methyl-CpG pair. It is not influenced by sequences flanking the methyl-CpGs. Mediates transcriptional repression through interaction with histone deacetylase and the corepressor SIN3A.

Tissue specificity Present in all adult somatic tissues tested.

Involvement in disease Defects in MECP2 may be a cause of Angelman syndrome (AS) [MIM:105830]; also known as happy puppet syndrome. AS is a neurodevelopmental disorder characterized by severe mental retardation, absent speech, ataxia, sociable affect and dysmorphic facial features. AS and Rett syndrome have overlapping clinical features.

Defects in MECP2 are the cause of mental retardation syndromic X-linked type 13 (MRXS13) [MIM:300055]. Mental retardation is a mental disorder characterized by significantly sub-average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRXS13 patients manifest mental retardation associated with other variable features such as spasticity, episodes of manic depressive psychosis, increased tone and macroorchidism.

Defects in MECP2 are the cause of Rett syndrome (RTT) [MIM:312750]. RTT is an X-linked dominant disease, it is a progressive neurologic developmental disorder and one of the most common causes of mental retardation in females. Patients appear to develop normally until 6 to 18 months of age, then gradually lose speech and purposeful hand movements and develop microcephaly, seizures, autism, ataxia, intermittent hyperventilation, and stereotypic hand movements. After initial regression, the condition stabilizes and patients usually survive into adulthood.

Defects in MECP2 may be the cause of susceptibility autism X-linked type 3 (AUTSX3) [MIM:300496]. AUTSX3 is a pervasive developmental disorder (PDD), prototypically characterized by impairments in reciprocal social interaction and communication, restricted and

stereotyped patterns of interests and activities, and the presence of developmental abnormalities by 3 years of age.

Defects in MECP2 are the cause of encephalopathy neonatal severe due to MECP2 mutations (ENS-MECP2) [MIM:300673]. Note=The MECP2 gene is mutated in Rett syndrome, a severe neurodevelopmental disorder that almost always occurs in females. Although it was first thought that MECP2 mutations causing Rett syndrome were lethal in males, later reports identified a severe neonatal encephalopathy in surviving male sibs of patients with Rett syndrome. Additional reports have confirmed a severe phenotype in males with Rett syndrome-associated MECP2 mutations.

Defects in MECP2 are the cause of mental retardation syndromic X-linked Lubs type (MRXSL) [MIM:300260]. Mental retardation is characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRXSL patients manifest mental retardation associated with variable features. They include swallowing dysfunction and gastroesophageal reflux with secondary recurrent respiratory infections, hypotonia, mild myopathy and characteristic facies such as downslanting palpebral fissures, hypertelorism and a short nose with a low nasal bridge. Note=Increased dosage of MECP2 due to gene duplication appears to be responsible for the mental retardation phenotype.

Sequence similarities

Contains 2 A.T hook DNA-binding domains.

Contains 1 MBD (methyl-CpG-binding) domain.

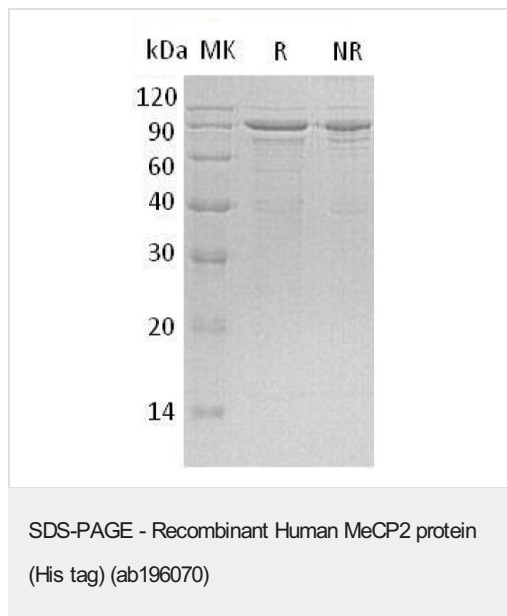
Post-translational modifications

Phosphorylated on Ser-423 in brain upon synaptic activity, which attenuates its repressor activity and seems to regulate dendritic growth and spine maturation.

Cellular localization

Nucleus. Colocalized with methyl-CpG in the genome.

Images



SDS-PAGE - Recombinant Human MeCP2 protein (His tag) (ab196070).

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