甲基化 CpG 结合蛋白 2 抗体

产品货号： mIR22098

英文名称： MeCP2

中文名称： 甲基化 CpG 结合蛋白 2 抗体

别　　名： AUTSX 3; AUTSX3; Mbd 5; MECP-2; MeCP 2 protein; Methyl CpG binding protein 2 (Rett syndrome); Methyl CpG binding protein 2; MRX 16; MRX 79; MRX16; MRX79; MRXS 13; MRXS13; MRXSL; PPMX; RTS; RTT; WBP 10; WBP10. MECP2_HUMAN

研究领域： 细胞生物　免疫学　发育生物学　染色质和核信号　神经生物学　转录调节因子

抗体来源： Rabbit

克隆类型： Polyclonal

交叉反应　： Human, Mouse, Rat, Dog, Pig, Cow, Horse, Sheep,

产品应用： IHC-P=1:400-800 （石蜡切片需做抗原修复）
not yet tested in other applications.
optimal dilutions/concentrations should be determined by the end user.

分 子 量： 52kDa

细胞定位： 细胞核

性　　状： Lyophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human MeCP2:421-486/486

亚　　型： IgG

纯化方法： affinity purified by Protein A

储 存 液： 0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.

保存条件： 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

产品介绍： DNA methylation is the major modification of eukaryotic genomes and plays an essential role in mammalian development. Human proteins MECP2, MBD1, MBD2, MBD3 and MBD4 comprise a family of nuclear proteins related by the presence in each of a methyl CpG binding domain (MBD). Each of these proteins, with the exception of MBD3, is capable of binding specifically to methylated DNA. MECP2, MBD1 and MBD2 can also repress transcription from methylated gene promoters. In contrast to other MBD family members, MECP2 is X-linked and subject to X inactivation. MECP2 is dispensible in stem cells, but is essential for embryonic development. MECP2 gene mutations are the cause of some cases of Rett syndrome, a progressive neurologic developmental disorder and one of the most common causes of mental retardation in females.

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.

Subunit:

Interacts with FNBP3 (By similarity). Interacts with CDKL5

Subcellular Location:

Present in all adult somatic tissues tested.

Tissue Specificity:

Present in all adult somatic tissues tested.

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

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 adaptative 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 adaptative 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.

Similarity:

Contains 2 A.T hook DNA-binding domains.

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

SWISS:

P51608

Gene ID:

4204

Important Note:

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

MECP2 蛋白是一种丰富的染色质结合蛋白,属于 DNA 结合蛋白大家族中一员,是一种转录抑制因子,能选择性地与甲基化的 CpG(甲基胞嘧啶)结合,特别是与含单个甲基化的 CpG 结合。MECP2 的突变可以导致持续性

神经系统退化（neurodegenerative disorder）Rett 综合症。这个 x-染色体相关综合症（X-linked syndrome）主要在女性身上体现， 他突变也可以导致男性的智力障碍，Rett 综合征是一种严重影响儿童精神运动发育的神经遗传病。

产品图片