泛酸激酶 2 抗体

产品货号： mlR8338

英文名称： PANK2

中文名称： 泛酸激酶 2 抗体

别　名： C20orf48; HARP; hPANK2; HSS; MGC15053; NBIA1; PANK2; PANK2_HUMAN; Pantothenate kinase 2 (Hallervorden Spatz syndrome); Pantothenate kinase 2; Pantothenic acid kinase 2; PKAN; RP23 387C21.4.

研究领域： 肿瘤　细胞生物　免疫学　神经生物学　信号转导

抗体来源： Rabbit

克隆类型： Polyclonal

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

产品应用： WB=1:500-2000 ELISA=1:500-1000 IHC-P=1:400-800 IHC-F=1:400-800 IF=1:50-200 （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： 57kDa

细胞定位： 细胞浆

性　　状： Lyophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human PANK2:401-500/570

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： Defects in PANK2 are the cause of neurodegeneration with brain iron accumulation type 1 (NBIA1); also known as pantothenate kinase-associated neurodegeneration (PKAN) or Hallervorden-Spatz

syndrome (HSS). It is an autosomal recessive neurodegenerative disorder associated with iron accumulation in the brain, primarily in the basal ganglia. Clinical manifestations include progressive muscle spasticity, hyperreflexia, muscle rigidity, dystonia, dysarthria, and intellectual deterioration which progresses to severe dementia over several years. It is clinically classified into classic, atypical, and intermediate phenotypes. Classic forms present with onset in the first decade, rapid progression, loss of independent ambulation within 15 years. Atypical forms have onset in the second decade, slow progression, maintenance of independent ambulation up to 40 years later. Intermediate forms manifest onset in the first decade with slow progression or onset in the second decade with rapid progression. Patients with early onset tend to also develop pigmentary retinopathy, whereas those with later onset tend to also have speech disorders and psychiatric features. All patients have the 'eye of the tiger' sign on **brain MRI.**

Defects in PANK2 are the cause of hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration (HARP). HARP is a rare syndrome with many clinical similarities to NBIA1.

Function:

May be the master regulator of the CoA biosynthesis (By similarity).

Subcellular Location:

Isoform 1: Mitochondrion.

Isoform 2: Cytoplasm (Potential).

Tissue Specificity:

Ubiquitous.

DISEASE:

Defects in PANK2 are the cause of neurodegeneration with brain iron accumulation type 1 (NBIA1) [MIM:234200]; also known as pantothenate kinase-associated neurodegeneration (PKAN) or Hallervorden-Spatz syndrome (HSS). It is an autosomal recessive neurodegenerative disorder associated with iron accumulation in the brain, primarily in the basal ganglia. Clinical manifestations include progressive muscle spasticity, hyperreflexia, muscle rigidity, dystonia, dysarthria, and intellectual deterioration which progresses to severe

dementia over several years. It is clinically classified into classic, atypical, and intermediate phenotypes. Classic forms present with onset in the first decade, rapid progression, loss of independent ambulation within 15 years. Atypical forms have onset in the second decade, slow progression, maintenance of independent ambulation up to 40 years later. Intermediate forms manifest onset in the first decade with slow progression or onset in the second decade with rapid progression. Patients with early onset tend to also develop pigmentary retinopathy, whereas those with later onset tend to also have speech disorders and psychiatric features. All patients have the 'eye of the tiger' sign on brain MRI.

Defects in PANK2 are the cause of hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration (HARP) [MIM:607236]. HARP is a rare syndrome with many clinical similarities to NBIA1.

Similarity:

Belongs to the type II pantothenate kinase family.

SWISS:

Q9BZ23

Gene ID:

80025

Important Note:

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