单跨膜蛋白 FOXRED1 抗体

产品货号： mlR13209

英文名称： FOXRED1

中文名称： 单跨膜蛋白 FOXRED1 抗体

别　　名： FAD dependent oxidoreductase domain containing 1; FAD dependent oxidoreductase domain containing protein 1; FAD-dependent oxidoreductase domain-containing protein 1; FOXRED 1; FOXRED1; FP634; FXRD1_HUMAN; H17.

研究领域： 肿瘤　细胞生物　神经生物学　信号转导　新陈代谢

抗体来源： Rabbit

克隆类型： Polyclonal

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

产品应用： WB=1:500-2000 ELISA=1:500-1000 IHC-P=1:400-800 IHC-F=1:400-800 ICC=1:100-500 IF=1:100-500（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： 54kDa

细胞定位： 细胞膜

性　　状： Lyophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human FOXRED1:251-350/486

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 ℃ 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 ℃.

PubMed： PubMed

产品介绍 ： FOXRED1 is a 486 amino acid single-pass membrane protein. Utilizing FAD as a cofactor, FOXRED1 may act as a chaperone protein essential for the function of mitochondrial complex I. Mutations to FOXRED1 may result in mitochondrial complex I deficiency (MT-C1D), which results in a wide range of clinical maladies from lethal neonatal disease to adult onset neurodegenerative disorders. Common phenotypes of MT-C1D include cardiomyopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. FOXRED1 exists as three alternatively spliced isoforms and is encoded by a gene mapping to human chromosome 11q24.2. With approximately 135 million base pairs and 1,400 genes, chromosome 11 makes up around 4% of human genomic DNA and is considered a gene and disease association dense chromosome.

Subcellular Location:

Membrane; Single-pass membrane protein (Potential).

DISEASE:

Defects in FOXRED1 are a cause of mitochondrial complex I deficiency (MT-C1D) [MIM:252010]. A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease.

SWISS:

Q96CU9

Gene ID:

55572

Important Note:

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

Leigh 综合征的发生率占新生儿的 1/40,000.具有不同的基因类型,但临床具有共性特点,一般发病在 1 岁或以后,表现为肌张力减退,发作性呕吐,共济失调,舞蹈徐动症和过度通气,脑病表现为丧失语言发育能力,运动异常表现为痉挛性运动和异常呼吸节律,出现脑干或基底节损害体征和听力丧失,小脑损害导致共济失调,眼震和张力失常.眼科症状表现为视力丧失和眼肌麻痹.出现亚临床的周围神经病,出现神经传导速度减慢 45%.临床体征可以在感染或糖尿病后出现.病程进展出现运动或智能减退.常在发病后 2 年内死亡.

产品图片