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Rabbit Anti-SLC25A13 antibody

SL4038R

Product Name:	SLC25A13
Chinese Name:	Mitochondrion内钙结合天冬氨酸/谷氨酸载体蛋白抗体
Alias:	ARALAR2; Calcium binding mitochondrial carrier protein Aralar2; Citrin; CTLN2; Ctrn; Mitochondrial aspartate glutamate carrier 2; RGD1565889; Solute carrier family 25 (citrin) member 13; Solute carrier family 25 member 13 (citrin); Solute carrier family 25 member 13; AI785475; CMC2_HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Chicken,Dog,Pig,Cow,Horse,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800IF=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:	74kDa
Cellular localization:	cytoplasmicThe cell membraneMitochondrion
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human SLC25A13:351-450/675
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:	SLC25A13 is a member of the mitochondrial carrier family. It contains four EF-hand Ca(2+) binding motifs in the N-terminal domain, and localizes to mitochondria. It catalyzes the exchange of aspartate for glutamate and a proton across the inner mitochondrial membrane, and is stimulated by calcium on the external side of the inner

mitochondrial membrane. Mutations in the SLC25A13 gene result in citrullinemia, type II. Multiple transcript variants encoding different isoforms have been found for this gene.

Function:
Catalyzes the calcium-dependent exchange of cytoplasmic glutamate with mitochondrial aspartate across the mitochondrial inner membrane. May have a function in the urea cycle.

Subcellular Location:
Mitochondrion inner membrane; Multi-pass membrane protein.

Tissue Specificity:
High levels in liver and low levels in kidney, pancreas, placenta, heart and brain.

DISEASE:
Defects in SLC25A13 are the cause of citrullinemia type 2 (CTLN2) [MIM:603471]. Citrullinemia belongs to the urea cycle disorders. It is an autosomal recessive disease characterized primarily by elevated serum and urine citrulline levels. Ammonia intoxication is another manifestation. CTLN2 is characterized by neuropsychiatric symptoms including abnormal behaviors, loss of memory, seizures and coma. Death can result from brain edema. Onset is sudden and usually between the ages of 20 and 50 years.
Defects in SLC25A13 are the cause of neonatal intrahepatic cholestasis due to citrin deficiency (NICCD) [MIM:605814]. NICCD is a form of citrullinemia type 2 with neonatal onset. NICCD is characterized by suppression of the bile flow, hepatic fibrosis, low birth weight, growth retardation, hypoproteinemia, variable liver dysfunction. NICCD is generally not severe and symptoms disappear by one year of age with an appropriate diet. Years or even decades later, however, some individuals develop the characteristic features of citrullinemia type 2 with neuropsychiatric symptoms.

Similarity:
Belongs to the mitochondrial carrier family.
Contains 4 EF-hand domains.
Contains 3 Solcar repeats.

SWISS:
Q9UJS0

Gene ID:
10165

Database links:

Entrez Gene: 10165Human

Entrez Gene: 50799Mouse

Entrez Gene: 362322Rat

Omim: 603859Human

SwissProt: Q9UJS0Human

SwissProt: Q9QXX4Mouse

Unigene: 489190Human

Unigene: 24513Mouse

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

Citrin是一种Mitochondrion内钙结合天冬氨酸/谷氨酸载体(Aspartate/ Glutamate Carrier,
AGC)蛋白，在尿素循环及其他代谢过程中发挥重要作用。Citrin缺乏症包含成年发作Ⅱ型瓜氨酸血症(Adult Onset Type Ⅱ Citrullinemia ,
CTLN2)和Citrin缺乏所致新生儿肝内胆汁淤积症(Neonatal Intrahepatic Cholestasis caused by Citrin Deficiency, NICCD)两种不同表型，为常染色体隐性遗传。
Citrin分子量约为74kDa，含675个氨基酸，在肝脏、肾脏及心脏中均有表达，位于Mitochondrion内膜。Citrin的N端有4个EF手型结构域，可结合钙离子，C端作为Mitochondrion载体活性部位有6个跨膜结构。研究还发现，Citrin的类似物－Aralar，同为天冬氨酸/谷氨酸载体蛋白，虽氨基酸序列与Citrin有77.8%的同源性，但组织分布明显不同，Citrin主要在肝脏而Aralar
主要在骨骼肌和脑中表达，提示Citrin缺乏症是一种局限于肝脏的疾病。
Citrin作为肝内主要的天冬氨酸/谷氨酸载体蛋白，其功能有3方面；
其一，将Mitochondrion中天冬氨酸转运至胞浆中，参与尿素、蛋白和核酸的合成。
其二，将天冬氨酸转运至胞浆，作为苹果酸/天冬氨酸穿梭的一个环节，将胞浆中糖酵解生成的NADH还原当量运至Mitochondrion内，参与能量、氨基酸、糖和脂代谢。
其三，在NADH形成及利用的同时促进乳糖糖异生。

CTLN2患者多死于脑水肿，脑损伤机制尚不明确，高血氨并非唯一致病因素，局部缺血、能量耗竭、神经毒性及代谢失调引起氧化应激反应，均可加速脑损伤。CTLN2患者多并发肝脏Tumour，其中大部分为肝细胞癌
。体外研究提示瓜氨酸的积聚对肝细胞的增生有促进作用，游离脂肪酸聚集造成的氧化应激和脂质过氧化反应对Tumour发生也有重要作用。