abcam

Product datasheet

Recombinant Human CaSR protein ab114274

画像数1

製品の詳細

製品名 Recombinant Human CaSR protein

 発現系
 Wheat germ

 アクセッション番号
 P41180

タンパク質長 Protein fragment

Animal free No

由来 Recombinant

生物種 Human

配列 GPDQRAQKKGDIILGGLFPIHFGVAAKDQDLKSRPESVECIR

YNFRGFRW

LQAMIFAIEEINSSPALLPNLTLGYRIFDTCNTVSKALEATL

SFVAQNKI DSLNLDEFCN

予測される分子量 38 kDa including tags

領域 21 to 130

特性

Our Abpromise guarantee covers the use of ab114274 in the following tested applications.

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

アプリケーション ELISA

SDS-PAGE

Western blot

製品の状態 Liquid

備考 This product was previously labelled as Calcium Sensing Receptor.

前処理および保存

保存方法および安定性 Shipped on dry ice. Upon delivery aliquot and store at -80°C. Avoid freeze / thaw cycles.

pH: 8.00

Constituents: 0.3% Glutathione, 0.79% Tris HCI

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関連情報

機能

Senses changes in the extracellular concentration of calcium ions. The activity of this receptor is mediated by a G-protein that activates a phosphatidylinositol-calcium second messenger system.

組織特異性

Expressed in the temporal lobe, frontal lobe, parietal lobe, hippocampus, and cerebellum. Also found in kidney, lung, liver, heart, skeletal muscle, placenta.

関連疾患

Defects in CASR are the cause of familial hypocalciuric hypercalcemia type 1 (FHH) [MIM:145980]. FHH is characterized by altered calcium homeostasis. Affected individuals exhibit mild or modest hypercalcemia, relative hypocalciuria, and inappropriately normal PTH levels. Defects in CASR are the cause of neonatal severe primary hyperparathyroidism (NSHPT) [MIM:239200]. NSHPT is a rare autosomal recessive life-threatening disorder characterized by very high serum calcium concentrations, skeletal demineralization, and parathyroid hyperplasia. In some instances NSHPT has been demonstrated to be the homozygous form of FHH. Defects in CASR are a cause of familial isolated hypoparathyroidism (FIH) [MIM:146200]; also called autosomal dominant hypoparathyroidism or autosomal dominant hypocalcemia. FIH is characterized by hypocalcemia and hyperphosphatemia due to inadequate secretion of parathyroid hormone. Symptoms are seizures, tetany and cramps. An autosomal recessive form of FIH also exists.

Defects in CASR are the cause of idiopathic generalized epilepsy type 8 (IGE8) [MIM:612899]; also known as EIG8. A disorder characterized by recurring generalized seizures in the absence of detectable brain lesions and/or metabolic abnormalities. Seizure types are variable, but include myoclonic seizures, absence seizures, febrile seizures, complex partial seizures, and generalized tonic-clonic seizures.

Note=Homozygous defects in CASR can be a cause of primary hyperparathyroidism in adulthood. Patients suffer from osteoporosis and renal calculi, have marked hypercalcemia and increased serum PTH concentrations.

配列類似性

Belongs to the G-protein coupled receptor 3 family.

翻訳後修飾

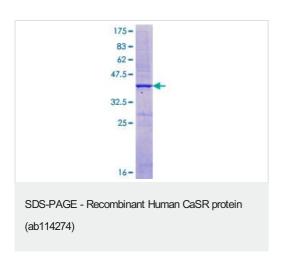
N-glycosylated.

Ubiquitinated by RNF19A; which induces proteasomal degradation.

細胞内局在

Cell membrane.

画像



12.5% SDS-PAGE analysis of ab114274 stained with Coomassie Blue.

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