abcam

Product datasheet

Recombinant Human CaSR protein ab114274

1 Image

Description

Product name Recombinant Human CaSR protein

Expression system Wheat germ

Accession P41180

Protein length Protein fragment

Animal free No

Nature Recombinant

Species Human

Sequence GPDQRAQKKGDIILGGLFPIHFGVAAKDQDLKSRPESVECI

RYNFRGFRW

LQAMIFAIEEINSSPALLPNLTLGYRIFDTCNTVSKALEATLS

FVAQNKI DSLNLDEFCN

Predicted molecular weight 38 kDa including tags

Amino acids 21 to 130

Specifications

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.

Applications ELISA

SDS-PAGE Western blot

Form Liquid

Additional notes This product was previously labelled as Calcium Sensing Receptor.

Preparation and Storage

Stability and Storage 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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General Info

Function

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.

Tissue specificity

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

Involvement in disease

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.

Sequence similarities

Belongs to the G-protein coupled receptor 3 family.

Post-translational modifications

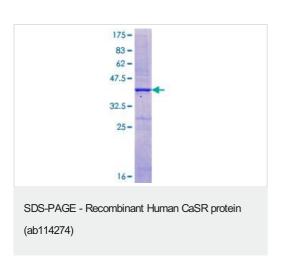
N-glycosylated.

Ubiquitinated by RNF19A; which induces proteasomal degradation.

Cellular localization

Cell membrane.

Images



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

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