# abcam

# Product datasheet

# Recombinant Human CaSR protein ab114274

## 1 图像

描述

产品名称 重组人CaSR蛋白

表达系统 Wheat germ

Accession P41180

**蛋白长度** Protein fragment

无动物成分 No

性质 Recombinant

**种属** Human

序列 GPDQRAQKKGDIILGGLFPIHFGVAAKDQDLKSRPESVECIR

YNFRGFRW

LQAMIFAIEEINSSPALLPNLTLGYRIFDTCNTVSKALEATL

SFVAQNKI DSLNLDEFCN

预测分子量 38 kDa including tags

**氨基酸** 21 to 130

技术指标

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.

应**用** ELISA

SDS-PAGE

Western blot

形式 Liquid

**补充说明** This product was previously labelled as Calcium Sensing Receptor.

制备和贮存

稳定性和存储 Shipped on dry ice. Upon delivery aliquot and store at -80℃. Avoid freeze / thaw cycles.

pH: 8.00

Constituents: 0.3% Glutathione, 0.79% Tris HCI

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#### 常规信息

功能

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.

组织特异性

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

疾病相关

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.

序列相似性

Belongs to the G-protein coupled receptor 3 family.

翻译后修饰

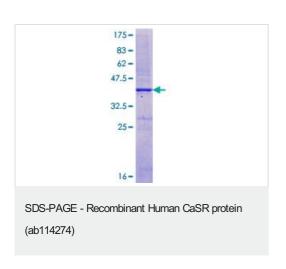
N-glycosylated.

Ubiquitinated by RNF19A; which induces proteasomal degradation.

细胞定位

Cell membrane.

# 图片



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

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