

Recombinant Human SLC4A1 cell lysate

Cat. No. SLC4A1-1635HCL Lot. No. (See product label)

SPECIFICATION

Product Overview

Species	Human
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Description

The protein encoded by this gene is part of the anion exchanger (AE) family and is expressed in the erythrocyte plasma membrane, where it functions as a chloride/bicarbonate exchanger involved in carbon dioxide transport from tissues to lungs. The protein comprises two domains that are structurally and functionally distinct. The N-terminal 40kDa domain is located in the cytoplasm and acts as an attachment site for the red cell skeleton by binding ankyrin. The glycosylated C-terminal membrane-associated domain contains 12-14 membrane spanning segments and carries out the stilbene disulphonate-sensitive exchange transport of anions. The cytoplasmic tail at the extreme C-terminus of the membrane domain binds carbonic anhydrase II. The encoded protein associates with the red cell membrane protein glycophorin A and this association promotes the correct folding and translocation of the exchanger. This protein is predominantly dimeric but forms tetramers in the presence of ankyrin. Many mutations in this gene are known in man, and these mutations can lead to two types of disease: destabilization of red cell membrane leading to hereditary spherocytosis, and defective kidney acid secretion leading to distal renal tubular acidosis. Other mutations that do not give rise to disease result in novel blood group antigens, which form the Diego blood group system. Southeast Asian ovalocytosis (SAO, Melanesian ovalocytosis) results from the heterozygous presence of a deletion in the encoded protein and is common in areas where Plasmodium falciparum malaria is endemic. One null mutation in this gene is known, resulting in very severe anemia and nephrocalcinosis.

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Size	100 ul
Storage Buffer	1X Sample Buffer (50 mM Tris-HCl, 2% SDS, 10% glycerol, 300 mM 2-mercaptoethanol, 0.01% Bromophenol blue)
Applications	Western Blot;
GENE INFORMATION	
Gene Name	SLC4A1 solute carrier family 4, anion exchanger, member 1 (erythrocyte membrane protein band 3, Diego blood group) [Homo sapiens]
Official Symbol	SLC4A1
Synonyms	SLC4A1; solute carrier family 4, anion exchanger, member 1 (erythrocyte membrane protein band 3, Diego blood group); AE1, DI, EPB3, Waldner blood group , WD; band 3 anion transport protein; CD233; FR; Froese blood group; RTA1A; SW; Swann blood group; WR; Wright blood group; anion exchanger 1; anion exchanger-1; Waldner blood group; anion exchange protein 1; erythroid anion exchange protein; erythrocyte membrane protein band 3; solute carrier family 4, anion exchanger, number 1; DI; WD; AE1; WD1; BND3; EPB3; EMPB3; MGC116750; MGC116753; MGC126619; MGC126623;
Gene ID	6521
mRNA Refseq	NM_000342
Protein Refseq	NP_000333
UniProt ID	P02730

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Chromosome Location	17q12-q21
Pathway	Bicarbonate transporters, organism-specific biosystem; Collecting duct acid secretion, organism-specific biosystem; Collecting duct acid secretion, conserved biosystem; SLC-mediated transmembrane transport, organism-specific biosystem; Transmembrane transport of small molecules, organism-specific biosystem; Transport of inorganic cations/anions and amino acids/oligopeptides, organism-specific biosystem;
Function	actin binding; anion transmembrane transporter activity; anion:anion antiporter activity; ankyrin binding; chloride transmembrane transporter activity; inorganic anion exchanger activity; protein C-terminus binding; protein anchor; protein binding; protein homodimerization activity; transporter activity;

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