10320 Camino Santa Fe, Suite G
San Diego, CA 92121
Tel: 858.875.1900 Fax: 858.875.1999

SLC12A3 Rabbit pAb

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Catalog # AP58719

Product Information

Application	IHC-P, IHC-F, IF, E
Primary Accession	P55017
Reactivity	Human
Predicted	Rat, Mouse, Rabbit, Dog
Host	Rabbit
Clonality	Polyclonal
Calculated MW	113139
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human SLC12A3/NCCT
Epitope Specificity	951-1021/1021
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Membrane.
DISEASE	Defects in SLC12A3 are the cause of Gitelman syndrome (GS). GS is an autosomal recessive disorder characterized by hypokalemic alkalosis in combination with hypomagnesemia, low urinary calcium, and increased renin activity associated with normal blood pressure. Patients are often asymptomatic or present transient periods of muscular weakness and tetany, usually accompanied by abdominal pain, vomiting and fever. The phenotype is highly heterogeneous in terms of age at onset and severity. Cardinal features such as hypocalciuria and hypomagnesemia might also change during the life cycle of a given patient. GS has overlapping features with Bartter syndrome.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Na-K-Cl cotransporters (NKCC) are channel proteins that aid in the transcellular movement of chloride across both secretory and absorptive epithelia. NKCC1 is expressed in muscle cells, neurons, and red blood cells. In the basolateral membrane of secretory epithelia, NKCC1 mediates active chloride secretion. The gene encoding human NKCC1 maps to chromosome 5q23.3. In mice, disruption of the NKCC1 gene leads to deafness and impaired balance. NKCC2 is specifically expressed in the kidney where it mediates active reabsorption of sodium chloride in the thick ascending limb of the loop of Henle. NKCC2 is sensitive to the clinically important diuretics furosemide and bumetanide. The gene encoding human NKCC2 maps to chromosome 15q15-q21 and mutations in this gene lead to Bartter's syndrome, an inherited hypokalaemic alkalosis. NCCT is a thiazide-sensitive Na-Cl cotransporter that is primarily expressed in the distal convoluted tubule of the kidney where it accounts for a significant fraction of net renal sodium reabsorption. The gene for human NCCT map to chromosome 16q13. Mutations in the gene encoding NCCT cause Gitelman's syndrome, a subset of Bartter's syndrome.

Additional Information

Gene ID	6559
Other Names	Solute carrier family 12 member 3, Na-Cl cotransporter, NCC, Na-Cl symporter, Thiazide-sensitive sodium-chloride cotransporter, SLC12A3 {ECO:0000303|PubMed:8812482, ECO:0000312|HGNC:HGNC:10912}
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500,ELISA=1:5000-10000
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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.

Protein Information

Name	SLC12A3 {ECO:0000303|PubMed:8812482, ECO:0000312|HGNC:HGNC:10912}
Function	Electroneutral sodium and chloride ion cotransporter, which acts as a key mediator of sodium and chloride reabsorption in kidney distal convoluted tubules (PubMed:18270262, PubMed:21613606, PubMed:22009145, PubMed:36351028, PubMed:36792826). Also acts as a receptor for the pro-inflammatory cytokine IL18, thereby contributing to IL18-induced cytokine production, including IFNG, IL6, IL18 and CCL2 (By similarity). May act either independently of IL18R1, or in a complex with IL18R1 (By similarity).
Cellular Location	Cell membrane; Multi-pass membrane protein. Apical cell membrane {ECO:0000250|UniProtKB:P59158}; Multi-pass membrane protein
Tissue Location	Predominantly expressed in the kidney (at protein level) (PubMed:29993276, PubMed:8812482). Localizes to the distal convoluted tubules (at protein level) (PubMed:29993276). Not detected in normal aorta, but abundantly expressed in fatty streaks and advanced atherosclerotic lesions (at protein level) (PubMed:26099046)

Background

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