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| Product name: | BTR1 Rabbit Polyclonal Antibody |
| Cat number: | ABN07692 |
| Conjugate: | Unconjugated |
| Size: | 100µL |
| Clone: | Polyclonal |
| Concentration: | 1mg/ml |
| Host: | Rabbit |
| Isotype: | IgG |
| Immunogen: | The antiserum was produced against synthesized peptide derived from human SLC4A11. AA range:291-340 |
| Reactivity: | Human,Rat,Mouse |
| Applications: | WB 1:500-1:2000,ELISA 1:10000-1:20000 |
| Molecular Weight: | 100kDa |
| Purification: | Affinity purification |
| Form: | Liquid |
| Buffer: | Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N. |
| Storage: | Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles. |

Background:

This gene encodes a voltage-regulated, electrogenic sodium-coupled borate cotransporter that is essential for borate homeostasis, cell growth and cell proliferation. Mutations in this gene have been associated with a number of endothelial corneal dystrophies including recessive corneal endothelial dystrophy 2, corneal dystrophy and perceptive deafness, and Fuchs endothelial corneal dystrophy. Multiple transcript variants encoding different isoforms have been described. [provided by RefSeq, Mar 2010],disease:Defects in SLC4A11 are the cause of corneal dystrophy and perceptive deafness (CDPD) [MIM:217400]; also known as corneal dystrophy and sensorineural deafness or Harboyan syndrome. CDPD consists of congenital corneal endothelial dystrophy and progressive perceptive deafness. Inheritance is autosomal recessive.,disease:Defects in SLC4A11 are the cause of corneal endothelial dystrophy type 2 (CHED2) [MIM:217700]; also known as congenital hereditary endothelial dystrophy of cornea. This bilateral corneal dystrophy is characterized by corneal opacification and nystagmus. Inheritance is autosomal recessive.,function:Transporter involved in borate homeostasis. In the absence of borate, it functions as a Na(+) and OH(-)(H(+)) channel. In the presence of borate functions as an electrogenic Na(+) coupled borate cotransporter.,PTM:Glycosylated.,similarity:Belongs to the anion exchanger (TC 2.A.31) family.,tissue specificity:Widely expressed. Highly expressed in kidney, testis, salivary gland, thyroid, trachea and corneal endothelium. Not detected in retina and lymphocytes.,