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

ATP6V0A2 Rabbit pAb

ATP6V0A2 Rabbit pAb
Catalog # AP54955

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q9Y487
Predicted	Rat, Pig, Dog, Human, Mouse, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	98082
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human ATP6V0A2
Epitope Specificity	1-100/856
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	Cell membrane. Endosome membrane. In kidney proximal tubules, also detected in subapical vesicles.
DISEASE	Defects in ATP6V0A2 are the cause of cutis laxa autosomal recessive type 2A (ARCL2A) [MIM:219200]. An autosomal recessive disorder characterized by an excessive congenital skin wrinkling, a large fontanelle with delayed closure, a typical facial appearance with downslanting palpebral fissures, a general connective tissue weakness, and varying degrees of growth and developmental delay and neurological abnormalities. Some affected individuals develop seizures and mental deterioration later in life, whereas the skin phenotype tends to become milder with age. At the molecular level, an abnormal glycosylation of serum proteins is observed in many cases. Defects in ATP6V0A2 are a cause of wrinkly skin syndrome (WSS) [MIM:278250]. WSS is rare autosomal recessive disorder characterized by wrinkling of the skin of the dorsum of the hands and feet, an increased number of palmar and plantar creases, wrinkled abdominal skin, multiple musculoskeletal abnormalities, microcephaly, growth failure and developmental delay.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	The protein encoded by this gene is a subunit of the vacuolar ATPase (v-ATPase), an heteromultimeric enzyme that is present in intracellular vesicles and in the plasma membrane of specialized cells, and which is essential for the acidification of diverse cellular components. V-ATPase is comprised of a membrane peripheral V(1) domain for ATP hydrolysis, and an integral membrane V(0) domain for proton translocation. The subunit encoded by this gene is a component of the V(0) domain. Mutations in this gene are a cause of both cutis laxa type II and wrinkly skin syndrome. [provided by RefSeq, Jul 2009]

Additional Information

Gene ID	23545
Other Names	V-type proton ATPase 116 kDa subunit a 2, V-ATPase 116 kDa subunit a 2, Lysosomal H(+)-transporting ATPase V0 subunit a 2, TJ6, Vacuolar proton translocating ATPase 116 kDa subunit a isoform 2, ATP6V0A2
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500
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	ATP6V0A2
Function	Subunit of the V0 complex of vacuolar(H+)-ATPase (V-ATPase), a multisubunit enzyme composed of a peripheral complex (V1) that hydrolyzes ATP and a membrane integral complex (V0) that translocates protons (By similarity). V-ATPase is responsible for acidifying and maintaining the pH of intracellular compartments and in some cell types, is targeted to the plasma membrane, where it is responsible for acidifying the extracellular environment (By similarity). Essential component of the endosomal pH-sensing machinery (PubMed:16415858). May play a role in maintaining the Golgi functions, such as glycosylation maturation, by controlling the Golgi pH (PubMed:18157129). In aerobic conditions, involved in intracellular iron homeostasis, thus triggering the activity of Fe(2+) prolyl hydroxylase (PHD) enzymes, and leading to HIF1A hydroxylation and subsequent proteasomal degradation (PubMed:28296633).
Cellular Location	Cell membrane; Multi-pass membrane protein. Endosome membrane. Note=In kidney proximal tubules, also detected in subapical vesicles.

Background

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.