GJB6 polyclonal antibody

Catalog # PAB17269 Size 100 ug

Specification

Product Description	Goat polyclonal antibody raised against synthetic peptide of GJB6.
Immunogen	A synthetic peptide corresponding to amino acids at internal region of human GJB6.
Sequence	C-RSKRAQTQKNHPNH
Host	Goat
Theoretical MW (kDa)	30.4
Specificity	Reported variants represent identical protein: NP_006774.2, NP_001103691.1, NP_001103689.1, NP_001103690.1
Form	Liquid
Purification	Antigen affinity purification
Concentration	0.5 mg/mL
Recommend Usage	ELISA (1:16000) The optimal working dilution should be determined by the end user.
Storage Buffer	In Tris saline, pH 7.3 (0.5% BSA, 0.02% sodium azide)
Storage Instruction	Store at -20°C. Aliquot to avoid repeated freezing and thawing.
Note	This product contains sodium azide: a POISONOUS AND HAZARDOUS SUBSTANCE which shoul d be handled by trained staff only.

Applications

- Enzyme-linked Immunoabsorbent Assay

Gene Info — GJB6

Entrez GeneID	10804
Protein Accession#	NP_006774.2
Gene Name	GJB6
Gene Alias	CX30, DFNA3, ED2, EDH, HED
Gene Description	gap junction protein, beta 6, 30kDa
Omim ID	129500 601544 604418
Gene Ontology	Hyperlink
Gene Summary	Gap junctions allow the transport of ions and metabolites between the cytoplasm of adjacent cells. They are formed by two hemichannels, made up of six connexin proteins assembled in groups. E ach connexin protein has four transmembrane segments, two extracellular loops, a cytoplasmic lo op formed between the two inner transmembrane segments, and the N- and C-terminus both bein g in the cytoplasm. The specificity of the gap junction is determined by which connexin proteins co mprise the hemichannel. In the past, connexin protein names were based on their molecular weig ht, however the new nomenclature uses sequential numbers based on which form (alpha or beta) of the gap junction is present. This gene encodes one of the connexin proteins. Mutations in this g ene have been found in some forms of deafness and in some families with hidrotic ectodermal dy splasia. [provided by RefSeq
Other Designations	OTTHUMP00000018096|OTTHUMP00000176870|OTTHUMP00000176871|OTTHUMP000001 76872|connexin 30|ectodermal dysplasia 2, hidrotic (Clouston syndrome)|gap junction protein, bet a 6|gap junction protein, beta 6 (connexin 30)

Publication Reference

- Mutation in gap and tight junctions in patients with non-syndromic hearing loss.

  Belguith H, Tlili A, Dhouib H, Ben Rebeh I, Lahmar I, Charfeddine I, Driss N, Ghorbel A, Ayadi H, Masmoudi S.

  Biochemical and Biophysical Research Communications 2009 Feb; 385(1):1.

Disease

- Auditory Perception
- Cholesteatoma

- Chronic Disease
- Cytomegalovirus Infections
- Deafness
- Disease Progression
- Genetic Predisposition to Disease
- Hearing Loss
- Presbycusis
- Rhinitis
- Sinusitis
- Speech Perception
- Syndrome
- Usher Syndromes
- Vestibulocochlear Nerve Diseases