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Product datasheet

Recombinant Human GJB2 protein ab152424

1 Image

Description

Product name	Recombinant Human GJB2 protein
Expression system	Wheat germ
Accession	**P29033**
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	MDWGTLQTILGGVNKHSTSIGKIWLTVLFIFRIMILVVAAKEV WGDEQAD FVCNTLQPGCKNVCYDHYFPISHIRLWALQLIFVSTPALLV AMHVAYRRH EKKRKFIKGEIKSEFKDIEEIKTQKVRIEGSLWWTYTSSIFFR VIFEAAF MYVFYVMYDGFSMQRLVKCNAWPCPNTVDCFVSRPTEK TVFTVFMIAVSG ICILLNVTELCYLLIRYCSGKSKKPV
Predicted molecular weight	51 kDa including tags
Amino acids	1 to 226

Specifications

Our **Abpromise guarantee** covers the use of **ab152424** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	ELISA
	SDS-PAGE
	Western blot
Form	Liquid
Additional notes	

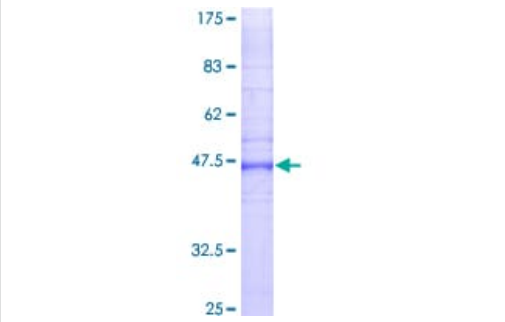
Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.31% Glutathione, 0.79% Tris HCl

General Info

Function	One gap junction consists of a cluster of closely packed pairs of transmembrane channels, the connexons, through which materials of low MW diffuse from one cell to a neighboring cell.
Involvement in disease	Defects in GJB2 are the cause of deafness autosomal recessive type 1A (DFNB1A) [MIM:220290]. DFNB1A is a form of sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. Defects in GJB2 are the cause of deafness autosomal dominant type 3A (DFNA3A) [MIM:601544]. Defects in GJB2 are a cause of Vohwinkel syndrome (VS) [MIM:124500]. VS is an autosomal dominant disease characterized by hyperkeratosis, constriction on finger and toes and congenital deafness. Defects in GJB2 are a cause of palmoplantar keratoderma with deafness (PPKDFN) [MIM:148350]. PPKDFN is an autosomal dominant disorder characterized by the association of palmoplantar hyperkeratosis with progressive, bilateral, high-frequency, sensorineural deafness. Defects in GJB2 are a cause of keratitis-ichthyosis-deafness syndrome (KID syndrome) [MIM:148210]; an autosomal dominant form of ectodermal dysplasia. Ectodermal dysplasias (EDs) constitute a heterogeneous group of developmental disorders affecting tissues of ectodermal origin. EDs are characterized by abnormal development of two or more ectodermal structures such as hair, teeth, nails and sweat glands, with or without any additional clinical sign. Each combination of clinical features represents a different type of ectodermal dysplasia. KID syndrome is characterized by the association of hyperkeratotic skin lesions with vascularizing keratitis and profound sensorineural hearing loss. Clinical features include deafness, ichthyosis, photobia, absent or decreased eyebrows, sparse or absent scalp hair, decreased sweating and dysplastic finger and toenails. Defects in GJB2 are the cause of Bart-Pumphrey syndrome (BPS) [MIM:149200]. BPS is an autosomal dominant disorder characterized by sensorineural hearing loss, palmoplantar keratoderma, knuckle pads, and leukonychia, It shows considerable phenotypic variability. Defects in GJB2 are the cause of ichthyosis hystrix-like with deafness syndrome (HID syndrome) [MIM:602540]. HID syndrome is an autosomal-dominant inherited keratinizing disorder characterized by sensorineural deafness and spiky hyperkeratosis affecting the entire skin. HID syndrome is considered to differ from the similar KID syndrome in the extent and time of occurrence of skin symptoms and the severity of the associated keratitis.
Sequence similarities	Belongs to the connexin family. Beta-type (group I) subfamily.
Cellular localization	Cell membrane. Cell junction > gap junction.

Images



SDS-PAGE - Recombinant Human GJB2 protein (ab152424)

12.5% SDS-PAGE analysis of ab152424 stained with Coomassie Blue.

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