Recombinant Human KIAA1279 Protein (His & GST Tag)

Catalog Number:PKSH030514

Note: Centrifuge before opening to ensure complete recovery of vial contents.

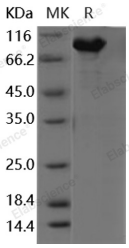
Description

Synonyms	KBP;KIAA1279;TTC20
Species	Human
Expression Host	Baculovirus-Insect Cells
Sequence	Met 1-Thr621
Accession	Q96EK5
Calculated Molecular Weight	99.6 kDa
Observed molecular weight	92-102 kDa
Tag	N-His-GST

Properties

Purity	> 90 % as determined by reducing SDS-PAGE.
Endotoxin	< 1.0 EU per μg of the protein as determined by the LAL method.
Storage	Generally, lyophilized proteins are stable for up to 12 months when stored at -20 to -80℃. Reconstituted protein solution can be stored at 4-8℃ for 2-7 days. Aliquots of reconstituted samples are stable at < -20℃ for 3 months.
Shipping	This product is provided as lyophilized powder which is shipped with ice packs.
Formulation	Lyophilized from sterile 20mM Tris, 500mM NaCl, 10% glycerol, pH 7.4 Normally 5 % - 8 % trehalose, mannitol and 0.01% Tween80 are added as protectants before lyophilization. Please refer to the specific buffer information in the printed manual.
Reconstitution	Please refer to the printed manual for detailed information.

Data



> 90 % as determined by reducing SDS-PAGE.

Background

KIFBP (Kinesin Family Binding Protein, also known as KIAA1279 and KIF1BP) is a Protein Coding gene. This gene encodes a kinesin family member 1 binding protein that is characterized by two tetratricopeptide repeats. The encoded protein localizes to the mitochondria and may be involved in regulating the transport of the mitochondria. Homozygous nonsense mutations in KIAA1279 at 1q22.1, encoding a protein with two tetratricopeptide repeats, underlie this syndromic form of Hirschsprung disease and generalized polymicrogyria, establishing the importance of KIAA1279 in both enteric and central nervous system development. KIAA1279 is widely expressed in the brain, testis, and other tissues. Diseases associated with KIFBP include Goldberg-Shprintzen Syndrome and Shprintzen-Goldberg Craniosynostosis Syndrome.

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