Recombinant Human CEP104 Protein, MYC/DDK-tagged

Cat. No. CEP104-3182H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Human CEP104 protein, fused to MYC/DDK-tagged at C-terminus, was expressed in HEK293.
Species	Human
Source	HEK293
Description	This gene encodes a centrosomal protein required for ciliogenesis and for ciliary tip structural integrity. The mammalian protein contains three amino-terminal hydrophobic domains, two glycosylation sites, four cysteine-rich motifs, and two regions with homology to the glutamate receptor ionotropic, NMDA 1 protein. During ciliogenesis, the encoded protein translocates from the distal tips of the centrioles to the tip of the elongating cilium. Knockdown of the protein in human retinal pigment cells results in severe defects in ciliogenesis with structural deformities at the ciliary tips. Allelic variants of this gene are associated with the autosomal-recessive disorder Joubert syndrome, which is characterized by a distinctive mid-hindbrain and cerebellar malformation, oculomotor apraxia, irregular breathing, developmental delay, and ataxia.
Form	25 mM Tris.HCl, pH 7.3, 100 mM glycine, 10% glycerol.
Molecular Mass	104.3 kDa
Purity	> 80% as determined by SDS-PAGE and Coomassie blue staining

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Concentration	>50 ug/mL as determined by microplate BCA method

GENE INFORMATION

Gene Name	CEP104 centrosomal protein 104kDa [Homo sapiens]
Official Symbol	CEP104
Synonyms	CFAP256; GlyBP; JBTS25; KIAA0562; ROC22
Gene ID	9731
mRNA Refseq	NM_014704
Protein Refseq	NP_055519
UniProt ID	O60308

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