

Recombinant Human CEP104 Protein, Myc/DDK-tagged, C13 and N15-labeled

Cat. No. CEP104-4036H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	KIAA0562 MS Standard C13 and N15-labeled recombinant protein (NP_055519) with a C-terminal MYC/DDK tag, was expressed in HEK293 cells.
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.
Molecular Mass	104.3 kDa
AA Sequence	MPHKIGFVVVSSSGHEDGFSARELMIHAPTVSGWRSRPRFCQFPQEIVLQMVERCRI RKLQLLAHQYMISSKIEFYISESLPEYFAPYQAERFRRLGYVSLCDNEKTGCKARELK

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SVYVDAVGQFLKLIFHQNHVKNYNIYNQVALVAINIIGDPADFSDESNTASREKLIDHY
 LGHNSDPALEGTARKSDYISPLDDLAFDMYQDPEVAQIIRKLDERKREAVQKERY
 DYAKKLKQAIADLQKVGERLGRYEVEKRCAVEKEDYDLAKEKKQQMEQYRAEVYE
 QLELHSLDAELMRPFDLPLQPLARSGSPCHQKPMPSLPQLEERGTEHQFAEPFL
 QEKPSYSLSISPQHSADVPLLPATDHPKINAESLPYDERPLPAIRKHGEAVVEPE
 MSNADISDARRGGMGEPEPLTEKALREASSAIDVLGETLVAEAYCKTWSYREDALL
 ALSKKLMEMPVGTPKEDLKNTLRASVFLVRRRAIKDIVTSVFQASLKLLKMITQYIPKH
 KLSKLETAHCVERTIPVLLTRTGDSSARLRVTAANFIQEMALFKEVKSQIIPSYLVQP
 LKANSSVHLAMSQMGLLARLLKDLGTGSSGFTIDNVMKFSVSALEHRVYEVRETAV
 RIILDMYRQHQASILEYLPPDDSNTRRNILYKTIFEGFAKIDGRATDAEMRARRKAATE
 EAEKQKKEEIKALQGQLAALKEIQAEVQEKESDAVKPKNQDIQGGKAAPAEALGIPD
 EHYLDNLCIFCGERSESFTEEGDLHYWKHCLMLTRCDHCKQVVEISSLTEHLLTEC
 DKKDGFGKCYRCSEAVFKEELPRHIKHKDCNPAKPEKLANRCPLCHENFSPGEEAW
 KAHLMPAGCTMNLRKTHILQKAPALQPGKSSAVAASGPLGSKAGSKIPTPKGGLS
 KSSSRTYAKRTRTRPLEQKLISEEDLAANDILDYKDDDDKV

Purity > 80% as determined by SDS-PAGE and Coomassie blue staining

Stability Stable for 3 months from receipt of products under proper storage and handling conditions.

Storage Store at -80 centigrade. Avoid repeated freeze-thaw cycles.

Concentration 50 µg/mL as determined by BCA

Storage Buffer 100 mM glycine, 25 mM Tris-HCl, pH 7.3.

GENE INFORMATION

Gene Name CEP104 centrosomal protein 104kDa [Homo sapiens (human)]

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Official Symbol	CEP104
Synonyms	CEP104; centrosomal protein 104kDa; KIAA0562; centrosomal protein of 104 kDa; GlyBP; glycine; glutamate; thienylcyclohexylpiperidine binding protein; RP1 286D6.4; RP1-286D6.4;
Gene ID	9731
mRNA Refseq	NM_014704
Protein Refseq	NP_055519
MIM	616690
UniProt ID	O60308
SDS-PAGE	