

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

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

## SPECIFICATION

<b>Product Overview</b>	KIAA0562 MS Standard C13 and N15-labeled recombinant protein (NP_055519) with a C-terminal MYC/DDK tag, was expressed in HEK293 cells.
<b>Species</b>	Human
<b>Source</b>	HEK293
<b>Description</b>	<p>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.</p>
<b>Molecular Mass</b>	104.3 kDa
<b>AA Sequence</b>	MPHKIGFVVVSSSGHEDGFSARELMIHAPT VSGWRSRPRFCQFPQEIVLQMVERCRI RKLQLLAHQYMISSKIEFYISESLPEYFAPYQAERFRRLGYVSLCDNEKTGCKARELK

 Tel: 1-631-559-9269    1-516-512-3133

 Email: [info@creative-biomart.com](mailto:info@creative-biomart.com)     Fax: 1-631-938-8127

 45-1 Ramsey Road, Shirley, NY 11967, USA

SVYVDAVGQFLKLIFHQNHVKNYNIYNQVALVAINIIGDPADFSDESNTASREKLIDHY  
 LGHNSDPALEGTYARKSDYISPLDDLAFDMYQDPEVAQIIRKLDERKREAVQKERY  
 DYAKKLKQAIADLQKVGERLGRYEVEKRCAVEKEDYDLAKEKKQQMEQYRAEVYE  
 QLELHSLDDAELMRRPFDLPLQPLARSGSPCHQKPMPSLPQLEERGTEHQFAEPFL  
 QEKPSSYSLTISPQHSADVPLLPATDPHPKINAECLPYDERPLPAIRKHGAEVVEPE  
 MSNADISDARRGGMLGEPEPLTEKALREASSAIDLGETLVAEAYCKTWSYREDALL  
 ALSKKLMEMPVGTPKEDLKNLTLRASVFLVRRRAIKDIVTSVFQASLKLKMIITQYIPKH  
 KLSKLETAHCVERTIPVLLTRTGDSARLRVTAANFIQEMALFKEVKSLQIIPSYLVQP  
 LKANSSVHLAMSQMGLLARLLKDLGTGSSGFTIDNVMKFSVSALEHRVYEVRETAV  
 RIILDMYRQHQASILEYLPPDDSNTRRNILYKTIFEGFAKIDGRATDAEMRARRKAATE  
 EAEKQKKEEIKALQGQLAALKEIQAEVQEKESDAVKPKNQDIQGGKAAPAEALGIPD  
 EHYLDNLCIFCGERSESFTEEGDLHYWKHCLMLTRCDHCKQVVEISSLTEHLLTEC  
 DKKDGFGKCYRCSEAVFKEELPRHIKHKDCNPAKPEKLANRCPLCHENFSPGEEAW  
 KAHLMGPACTMNLRKTHILQKAPALQPGKSSAVAASGPLGSKAGSKIPTPKGGLS  
 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) ]

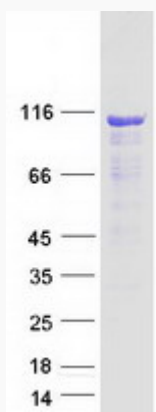
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<b>Official Symbol</b>	CEP104
<b>Synonyms</b>	CEP104; centrosomal protein 104kDa; KIAA0562; centrosomal protein of 104 kDa; GlyBP; glycine; glutamate; thienylcyclohexylpiperidine binding protein; RP1 286D6.4; RP1-286D6.4;
<b>Gene ID</b>	9731
<b>mRNA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

**SDS-PAGE**



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