Recombinant Human CEP290

Catalog No: #GP12757

Package Size: #GP12757-1 100ug



Orders: order@signalwayantibody.com Support: tech@signalwayantibody.com

Description

Product Name	Recombinant Human CEP290
Brief Description	Recombinant Protein
Immunogen Description	Fusion protein corresponding to a region derived from 941-1104 amino acids of human CEP290
Target Name	centrosomal protein 290
Other Names	CT87; MKS4; POC3; rd16; BBS14; JBTS5; LCA10; NPHP6; SLSN6; 3H11Ag
Accession No.	Swissprot:O15078Gene Accession:BC008641
Uniprot	O15078
GeneID	80184;
Storage	-20~-80°C, pH 7.6 PBS

Background

This gene encodes a protein with 13 putative coiled-coil domains, a region with homology to SMC chromosome segregation ATPases, six KID motifs, three tropomyosin homology domains and an ATP/GTP binding site motif A. The protein is localized to the centrosome and cilia and has sites for N-glycosylation, tyrosine sulfation, phosphorylation, N-myristoylation, and amidation. Mutations in this gene have been associated with Joubert syndrome and nephronophthisis and the presence of antibodies against this protein is associated with several forms of cancer.

References

Note: For in vitro research use only, not for diagnostic or therapeutic use. This product is not a medical device.

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