

Recombinant Human CEP290 protein ab164215

1 Image

Description	
Product name	Recombinant Human CEP290 protein
Expression system	Wheat germ
Accession	<u>O15078</u>
Protein length	Protein fragment
Animal free	No
Nature	Recombinant
Species	Human
Sequence	MAIFKIAALQKVVDNSVSLSELELANKQYNELTAKYRDILQ KDNMLVQRT SNLEHLECENISLKEQVESINKELEITKEKLHTIEQAWEQET KLGNESSM DKAKKSITNSDMVSISKKITMLEMKELNERQRAEHCQKMYE HLRTSLKQM EERNFELETKFAEV
Amino acids	941 to 1104
Tags	GST tag N-Terminus

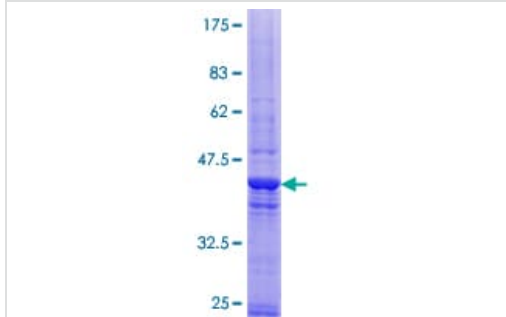
Specifications	
Our <u>Abpromise guarantee</u> covers the use of ab164215 in the following tested applications.	
The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.	
Applications	ELISA Western blot
Form	Liquid
Additional notes	

Preparation and Storage	
Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80°C. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.31% Glutathione, 0.79% Tris HCl

General Info

Function	Activates ATF4-mediated transcription. Required for the correct localization of ciliary and phototransduction proteins in retinal photoreceptor cells; may play a role in ciliary transport processes.
Tissue specificity	Ubiquitous. Expressed strongly in placenta and weakly in brain.
Involvement in disease	<p>Defects in CEP290 are a cause of Joubert syndrome type 5 (JBTS5) [MIM:610188]. Joubert syndrome is an autosomal recessive disease characterized by cerebellar vermis hypoplasia with prominent superior cerebellar peduncles (the 'molar tooth sign' on axial magnetic resonance imaging), psychomotor delay, hypotonia, ataxia, oculomotor apraxia and neonatal breathing abnormalities. JBTS5 shares the neurologic and neuroradiologic features of Joubert syndrome together with severe retinal dystrophy and/or progressive renal failure characterized by nephronophthisis.</p> <p>Defects in CEP290 are a cause of Senior-Loken syndrome type 6 (SLSN6) [MIM:610189]. Senior-Loken syndrome is also known as juvenile nephronophthisis with Leber amaurosis. It is an autosomal recessive renal-retinal disorder, characterized by progressive wasting of the filtering unit of the kidney, with or without medullary cystic renal disease, and progressive eye disease.</p> <p>Defects in CEP290 are the cause of Leber congenital amaurosis type 10 (LCA10) [MIM:611755]. LCA designates a clinically and genetically heterogeneous group of childhood retinal degenerations, generally inherited in an autosomal recessive manner. Affected infants have little or no retinal photoreceptor function as tested by electroretinography. LCA represents the most common genetic cause of congenital visual impairment in infants and children.</p> <p>Defects in CEP290 are the cause of Meckel syndrome type 4 (MKS4) [MIM:611134]. MKS4 is an autosomal recessive disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly.</p> <p>Note=Antibodies against CEP290 are present in sera from patients with cutaneous T-cell lymphomas, but not in the healthy control population.</p> <p>Defects in CEP290 are the cause of Bardet-Biedl syndrome type 14 (BBS14) [MIM:209900]. A syndrome characterized by usually severe pigmentary retinopathy, early-onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for disease manifestation in some cases (triallelic inheritance).</p>
Cellular localization	Cytoplasm > cytoskeleton > centrosome. Nucleus. Cell projection > cilium. Connecting cilium of photoreceptor cells, base of cilium in kidney intramedullary collecting duct cells.

Images



SDS-PAGE - Recombinant Human CEP290 protein
(ab164215)

ab164215 on a 12.5% SDS-PAGE stained with Coomassie Blue.

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