abcam

Product datasheet

Anti-CEP290 antibody ab77479

1 Abreviews

Overview

Product name	Anti-CEP290 antibody
Description	Goat polyclonal to CEP290
	This product is a fast track antibody. It has been affinity purified and shows high titre values against the immunizing peptide by ELISA. Read the terms of use »
Host species	Goat
Species reactivity	**Predicted to work with:** Human
Immunogen	Synthetic peptide: QSGAESTIPDADQ (Human) from the internal region of the protein sequence according to NP_079390.3. Run BLAST with Run BLAST with
Positive control	Human kidney, ovary and thymus lysates

Properties

Form	Liquid
Storage instructions	Shipped at 4°C. Upon delivery aliquot and store at -20°C. Avoid freeze / thaw cycles.
Storage buffer	Preservative: 0.02% Sodium Azide Constituents: 0.5% BSA, Tris buffered saline, pH 7.3
Purity	Immunogen affinity purified
Purification notes	Purified from goat serum by ammonium sulphate precipitation followed by antigen affinity chromatography using the immunizing peptide.
Clonality	Polyclonal
Isotype	IgG

Applications

Fast track antibodies constitute a diverse group of products that have been released to accelerate your research, but are not yet fully characterized. They have all been affinity purified and show high titre values against the immunizing peptide (by ELISA).

Application	Abreviews	Notes
ELISA		1/8000.

Fast track terms of use

Target

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

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.
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.
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.
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.
Note=Antibodies against CEP290 are present in sera from patients with cutaneous T-cell lymphomas, but not in the healthy control population.
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).

Cellular localization

Cytoplasm > cytoskeleton > centrosome. Nucleus. Cell projection > cilium. Connecting cilium of photoreceptor cells, base of cilium in kidney intramedullary collecting duct cells.

Terms and conditions

- Guarantee only valid for products bought direct from Abcam or one of our authorized distributors