

Product name:	ALMS1 Rabbit Polyclonal Antibody
Cat number:	ABN06789
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	Synthesized peptide derived from ALMS1 . at AA range: 1530-1610
Reactivity:	Human,Rat,Mouse
Applications:	WB 1:500-1:2000,IHC 1:100-1:300,ICC/IF 1:50-1:200,ELISA 1:20000-1:40000
Molecular Weight:	460kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

This gene encodes a protein containing a large tandem-repeat domain as well as additional low complexity regions. The encoded protein functions in microtubule organization, particularly in the formation and maintenance of cilia. Mutations in this gene cause Alstrom syndrome. There is a pseudogene for this gene located adjacent in the same region of chromosome 2. Alternative splice variants have been described but their full length nature has not been determined. [provided by RefSeq, Apr 2014],developmental stage:Widely expressed in fetal tissues. Detected in fetal pancreas, skeletal muscle, liver, kidney and brain (at protein level). Expressed in fetal aorta and brain.,disease:Defects in ALMS1 are the cause of Alstrom syndrome (ALMS) [MIM:203800]. Alstrom syndrome is a rare autosomal recessive disorder characterized by progressive cone-rod retinal dystrophy, neurosensory hearing loss, early childhood obesity and type 2 diabetes mellitus. Dilated cardiomyopathy, acanthosis nigricans, male hypogonadism, hypothyroidism, developmental delay and hepatic dysfunction can also be associated with the syndrome.,function:Possible role in intracellular trafficking.,PTM:Phosphorylated upon DNA damage, probably by ATM or ATR.,subcellular location:Associated with centrosomes and basal body at the base of primary cilia. During mitosis localizes to both spindle poles.,tissue specificity:Expressed in all tissues tested including adipose and pancreas. Expressed by beta-cells of the islets in the pancreas (at protein level),.