

Product name:	ATPAF2 Rabbit Polyclonal Antibody
Cat number:	ABN07346
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human ATPAF2. AA range:21-70
Reactivity:	Human,Mouse
Applications:	WB 1:500-1:2000,ELISA 1:20000-1:40000
Molecular Weight:	35kDa
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:

ATP synthase mitochondrial F1 complex assembly factor 2(ATPAF2) Homo sapiens This gene encodes an assembly factor for the F(1) component of the mitochondrial ATP synthase. This protein binds specifically to the F1 alpha subunit and is thought to prevent this subunit from forming nonproductive homooligomers during enzyme assembly. This gene is located within the Smith-Magenis syndrome region on chromosome 17. An alternatively spliced transcript variant has been described, but its biological validity has not been determined. [provided by RefSeq, Jul 2008],disease:Defects in ATPAF2 are the cause of complex V mitochondrial respiratory chain ATPAF2 subunit deficiency (ATPAF2 deficiency) [MIM:604273]; also called ATP synthase deficiency or ATPase deficiency. ATPAF2 deficiency seems to be an early presenting disease in which lactic acidosis, dysmorphic features, and methyl glutaconic aciduria can be major clues in the diagnosis. Dysmorphic features include a large mouth, prominent nasal bridge, micrognathia, rocker-bottom feet and flexion contractures of the limbs associated with camptodactyly. Patients are hypertonic and have an enlarged liver, hypoplastic kidneys and elevated lactate levels in urine, plasma and cerebro spinal fluid (CSF).,function:May play a role in the assembly of the F1 component of the mitochondrial ATP synthase (ATPase).,similarity:Belongs to the ATP12 family.,subunit:Interacts with ATP5A1.,tissue specificity:Widely expressed.,