

Product name:	ERAB Rabbit Polyclonal Antibody
Cat number:	ABN10561
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human ERAB. AA range:111-160
Reactivity:	Human,Mouse,Rat,Monkey
Applications:	WB 1:500-1:2000,IHC 1:100-1:300,ICC/IF 1:50-1:200,ELISA 1:20000-1:40000
Molecular Weight:	27kDa
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 3-hydroxyacyl-CoA dehydrogenase type II, a member of the short-chain dehydrogenase/reductase superfamily. The gene product is a mitochondrial protein that catalyzes the oxidation of a wide variety of fatty acids and steroids, and is a subunit of mitochondrial ribonuclease P, which is involved in tRNA maturation. The protein has been implicated in the development of Alzheimer disease, and mutations in the gene are the cause of 17beta-hydroxysteroid dehydrogenase type 10 (HSD10) deficiency. Several alternatively spliced transcript variants have been identified, but the full-length nature of only two transcript variants has been determined. [provided by RefSeq, Aug 2014], catalytic activity:(2S,3S)-3-hydroxy-2-methylbutanoyl-CoA + NAD(+) = 2-methylacetoacetyl-CoA + NADH., catalytic activity:(S)-3-hydroxyacyl-CoA + NAD(+) = 3-oxoacyl-CoA + NADH., disease:A chromosomal microduplication involving HSD17B10 and HUWE1 is the cause of mental retardation X-linked type 17 (MRX17) [MIM:300705]; also known as mental retardation X-linked type 31 (MRX31). Mental retardation is characterized by significantly sub-average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. In contrast to syndromic or specific X-linked mental retardation which also present with associated physical, neurological and/or psychiatric manifestations, intellectual deficiency is the only primary symptom of non-syndromic X-linked mental retardation., disease:Defects in HSD17B10 are the cause of 2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency (MHBD deficiency) [MIM:300438]. MHBD deficiency leads to neurological abnormalities, including psychomotor retardation, and, in virtually all patients, loss of mental and motor skills., disease:Defects in HSD17B10 are the cause of mental retardation syndromic X-linked type 10 (MRXS10) [MIM:300220]. MRXS10 is characterized by mild mental retardation, choreoathetosis and abnormal behavior., function:Functions in mitochondrial tRNA maturation. Part of mitochondrial ribonuclease P, an enzyme composed of MRPP1/RG9MTD1, MRPP2/HSD17B10 and MRPP3/KIAA0391, which cleaves tRNA molecules in their 5'-ends. By interacting with intracellular amyloid-beta, it may contribute to the neuronal dysfunction associated with Alzheimer disease (AD)., similarity:Belongs to the short-chain dehydrogenases/reductases (SDR) family., subunit:Homotetramer (By similarity). Interacts with MRPP1/RG9MTD1 and MRPP3/KIAA0391., tissue specificity:Expressed in normal tissues but is overexpressed in neurons affected in AD.,