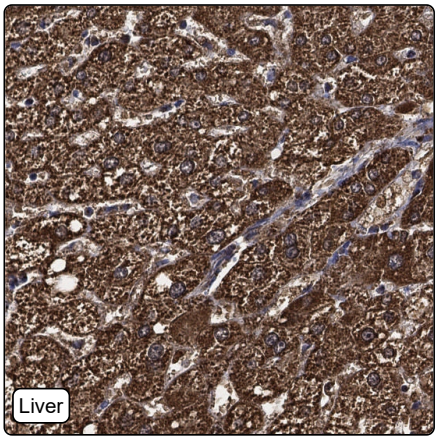
CPTC-FH-1 (CAB080323)

Uniprot ID: P07954
Protein name: FUMH_HUMAN
Full name: Fumarate hydratase, mitochondrial
Tissue specificity: Expressed in red blood cells; underexpressed in red blood cells (cytoplasm) of patients with hereditary non-spherocytic hemolytic anemia of unknown etiology.
Function: Catalyzes the reversible stereospecific interconversion of fumarate to L-malate (PubMed:30761759). Experiments in other species have demonstrated that specific isoforms of this protein act in defined pathways and favor one direction over the other (Probable). [Isoform Mitochondrial]: Catalyzes the hydration of fumarate to L-malate in the tricarboxylic acid (TCA) cycle to facilitate a transition step in the production of energy in the form of NADH. [Isoform Cytoplasmic]: Catalyzes the dehydration of L-malate to fumarate (By similarity). Fumarate metabolism in the cytosol plays a role during urea cycle and arginine metabolism; fumarate being a by-product of the urea cycle and amino-acid catabolism (By similarity). Also plays a role in DNA repair by promoting non-homologous end-joining (NHEJ) (PubMed:20231875, PubMed:26237645). In response to DNA damage and phosphorylation by PRKDC, translocates to the nucleus and accumulates at DNA double-strand breaks (DSBs): acts by catalyzing formation of fumarate, an inhibitor of KDM2B histone demethylase activity, resulting in enhanced dimethylation of histone H3 'Lys-36' (H3K36me2) (PubMed:26237645).
Subcellular location:
Isoform Cytoplasmic:
Cytoplasm > Cytosol (*experimental evidence*)
Nucleus (*experimental evidence*)
Chromosome (*experimental evidence*)
NOTE: Translocates to the nucleus in response to DNA damage: localizes to DNA double-strand breaks (DSBs) following phosphorylation by PRKDC.
Isoform Mitochondrial:
Mitochondrion (*experimental evidence*)
Protein existence: Experimental evidence at protein level

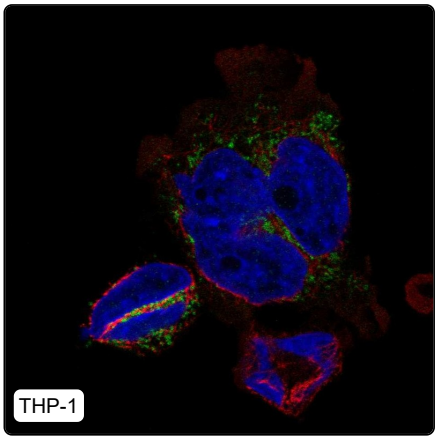
Comment:

Immunohistochemistry



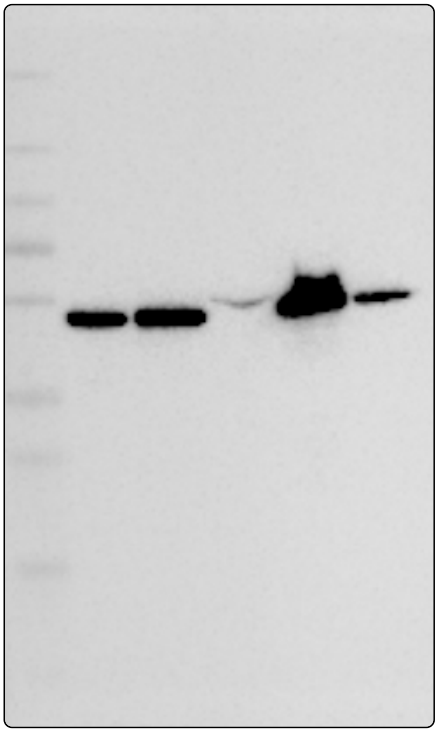
IHC protocol:	HIER pH6, Dilution 1:4000
IHC test staining:	Cytoplasmic positivity in most tissues.
Literature conformance:	Partly consistent with extensive gene/protein characterization data
Literature significance:	
RNA similarity:	Medium consistency between antibody staining and RNA expression data
RNA tissue specificity:	Tissue enhanced (liver)
RNA tissue distribution:	Detected in all
IHC Sibling similarity:	Other antibody shows similar IHC staining pattern

Immunofluorescence



IF Overlay:	antibody (green), anti-tubulin (red) and DAPI (blue)
IF main location:	Mitochondria - 1 [3]: **Supportive** (auto)
IF additional location:	
IF approved for publication on HPA:	Yes
IF in THP-1:	Mitochondria
IF in U2OS:	Mitochondria

Western blot



WB Size markers (kDa):	250, 130, 100, 70, 55, 35, 25, 15, 10
WB Lanes:	Marker (1), RT-4 (2), U-251MG (3), Plasma (4), Liver (5), Tonsil (6)
WB Target weight (kDa):	55
WB Validation:	Supported (Single band corresponding to the predicted size in kDa (+/-20%).)