

Product datasheet for **TP726928**

Fumarylacetoacetate hydrolase (FAH) Human Recombinant Protein

Product data:

Product Type:	Recombinant Proteins
Description:	Recombinant Human Fumarylacetoacetase/FAH (C-6His)
Species:	Human
Expression cDNA Clone or AA Sequence:	Ser2-Ser419
Tag:	C-His
Buffer:	Lyophilized from a 0.2 um filtered solution of 20mM Tris-HCl, 150mM NaCl, pH8.5.
Note:	Recombinant Human Fumarylacetoacetase is produced by our Mammalian expression system and the target gene encoding Ser2-Ser419 is expressed with a 6His tag at the C-terminus.
Storage:	Lyophilized protein should be stored at < -20°C, though stable at room temperature for 3 weeks. Reconstituted protein solution can be stored at 4-7°C for 2-7 days. Aliquots of reconstituted samples are stable at < -20°C for 3 months.
Stability:	12 months from date of despatch
Locus ID:	2184
UniProt ID:	P16930
Synonyms:	Fumarylacetoacetase; FAA; Beta-Diketonase; Fumarylacetoacetate Hydrolase; FAH
Summary:	Fumarylacetoacetase belongs to the FAH family. Fumarylacetoacetase is primarily expressed in liver and kidney. It exists as a homodimer and catalyzes the hydrolysis of 4-fumarylacetoacetate into fumarate and acetoacetate. Defects in Fumarylacetoacetase cause tyrosinemia type 1, which is a congenital metabolism defect characterized by elevated levels of tyrosine in the blood and urine, and hepatorenal manifestations. Typical features include renal tubular injury, self-mutilation, hepatic necrosis, episodic weakness, and seizures.
Protein Families:	Druggable Genome
Protein Pathways:	Metabolic pathways, Tyrosine metabolism


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