

## 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.
Stability:	12 months from date of despatch
Locus ID:	2184
UniProt ID:	<a href="#">P16930</a>
Summary:	Fumarylacetoacetase belongs to the FAH family. Fumarylacetoacetase is primary 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 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.



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