

Recombinant Human FAH, His-tagged

Cat. No. FAH-85H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Human Fumarylacetoacetase/FAH is produced by our mammalian expression system in human cells. The target protein is expressed with sequence (Ser2-Ser419) of Human FAH fused with a polyhistidine tag at the C-terminus.
Species	Human
Source	HEK293
ProteinLength	2-419 a.a.
Description	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.
Form	Ser2-Ser419
AA Sequence	MSFIPVAEDSDFPIHNLPYGVFSTRGDPRPRIGVAIGDQILDLSIIKHLFTGPVLSKHQ DVFNQP TLNSFMGLGQAAWKEARVFLQNLLSVSQARLRDDTELKCAFISQASAT MHLPATIGDYTDYSS RQHATNVGIMFRDKENALMPNWLHLPVGYHGRASSVVVS GTPIRRPMPGQMKPDDSKPPVYGACKL LDMELEMAFFVGPNGRLGEPIPISKAHEHI FGMVLMDWSARDIQKWEYVPLGPFLGKSFGTTVS PWVVPMDALMPFAVNPKNQ

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DPRPLPYLCHDEPYTFDINLSVNLKGEGMSQAATICKSNFKYMYWT MLQQLTHHSV
 NGCNLRPGDLLASGTISGPEPENFGSMLELSWKGTKPIDLGNGQTRKFLLDGDEV II
 TGYCQGDGYRIGFGQCAGKVLPALLPSVDHHHHHH

Endotoxin Less than 0.1 ng/μg (1 IEU/μg).

Purity Greater than 95% as determined by SEC-HPLC and reducing SDS-PAGE.

GENE INFORMATION

Gene Name FAH fumarylacetoacetate hydrolase (fumarylacetoacetase) [Homo sapiens]

Official Symbol FAH

Synonyms FAH; fumarylacetoacetate hydrolase (fumarylacetoacetase); fumarylacetoacetase; FAA; beta-diketonase; FLJ51912;

Gene ID 2184

mRNA Refseq NM_000137

Protein Refseq NP_000128

MIM 613871

UniProt ID P16930

Chromosome Location 15q23-q25

Pathway Metabolic pathways, organism-specific biosystem; Metabolism, organism-specific

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biosystem; Metabolism of amino acids and derivatives, organism-specific biosystem; Phenylalanine and tyrosine catabolism, organism-specific biosystem; Tyrosine degradation, tyrosine => homogentisate, organism-specific biosystem; Tyrosine degradation, tyrosine =>

Function

catalytic activity; fumarylacetoacetase activity; fumarylacetoacetase activity; hydrolase activity; metal ion binding;

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