

Product datasheet for **RC401077**

FH (NM_000143) Human Mutant ORF Clone

Product data:

Product Type:	Mutant ORF Clones
Product Name:	FH (NM_000143) Human Mutant ORF Clone
Mutation Description:	Q47X
Affected Codon#:	47
Affected NT#:	139
Nucleotide Mutation:	FH Mutant (Q47X), Myc-DDK-tagged ORF clone of Homo sapiens fumarate hydratase (FH), nuclear gene encoding mitochondrial protein as transfection-ready DNA
Effect:	Muliple leiomyomosis
Symbol:	FH
Synonyms:	FMRD; HLRCC; HsFH; LRCC; MCL; MCUL1
E. coli Selection:	Kanamycin (25 ug/mL)
Mammalian Cell Selection:	Neomycin
Vector:	pCMV6-Entry (PS100001)
Tag:	Myc-DDK
ACCN:	NM_000143
ORF Size:	138 bp
Restriction Sites:	SgfI-MluI
ORF Nucleotide Sequence:	>RC401077 representing NM_000143 Red=Cloning site Blue=ORF Green=Tags(s)

TTTTGTAATACGACTCACTATAGGGCGGCCGGGAATTCGTCGACTGGATCCGGTACCGAGGAGATCTGCC
GCC**GCGATCGCC**

ATGTACCGAGCACTTCGGCTCCTCGCGGCTCGCGTCCCCTCGTGCGGGCTCCAGCCGACGCTTAGCTT
CGGCTCCCGGCTTGGGTGGCGCGCCGTGCCCTCGTTTTGGCCTCCGAACGCGGCTCGAATGGCAAGC

AGCGGACCGACGCGTACGCGCCGCTCGAGCAGAACTCATCTCAGAAGAGGATCTGGCAGCAAATGATATCC
TGGATTACAAGGATGACGACGA TAAGGTTTAA



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Locus ID:	2271
Cytogenetics:	1q43
Domains:	lyase_1
Protein Families:	Druggable Genome
Protein Pathways:	Citrate cycle (TCA cycle), Metabolic pathways, Pathways in cancer, Renal cell carcinoma
MW:	5.1 kDa
Gene Summary:	<p>The protein encoded by this gene is an enzymatic component of the tricarboxylic acid (TCA) cycle, or Krebs cycle, and catalyzes the formation of L-malate from fumarate. It exists in both a cytosolic form and an N-terminal extended form, differing only in the translation start site used. The N-terminal extended form is targeted to the mitochondrion, where the removal of the extension generates the same form as in the cytoplasm. It is similar to some thermostable class II fumarases and functions as a homotetramer. Mutations in this gene can cause fumarase deficiency and lead to progressive encephalopathy. [provided by RefSeq, Jul 2008]</p>