

## **Product datasheet for RC401079**

## 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: R101X

Affected Codon#: 101

Affected NT#: 301

Nucleotide Mutation: FH Mutant (R101X), 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 Neomycin

Selection:

**Vector:** pCMV6-Entry (PS100001)

Tag: Myc-DDK
ACCN: NM 000143

ORF Size: 300 bp
Restriction Sites: Sgfl-Mlul

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## FH (NM\_000143) Human Mutant ORF Clone - RC401079

ORF Nucleotide Sequence:

>RC401079 representing NM\_000143

Red=Cloning site Blue=ORF Green=Tags(s)

 ${\tt TTTTGTAATACGACTCACTATAGGGCGGCCGGGAATTCGTCGACTGGATCCGGTACCGAGGAGATCTGCCGCCGCGATCGCC}$ 

AGCGGACCGACGCGTACGCGGCCGCTCGAGCAGAAACTCATCTCAGAAGAGGATCTGGCAGCAAATGATATCCTGGATTACAAGGATGACGACGA TAAGGTTTAA

**Protein Sequence:** 

>RC401079 representing NM\_000143 Red=Cloning site Green=Tags(s)

 ${\tt MYRALRLLARSRPLVRAPAAALASAPGLGGAAVPSFWPPNAARMASQNSFRIEYDTFGELKVPNDKYYGA} \\ {\tt OTVRSTMNFKIGGVTERMPTPVIKAFGILK} \\$ 

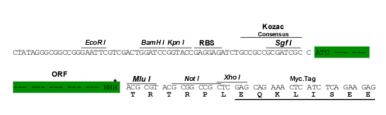
**SGPTRTRRL**EQKLISEEDLAANDILDYKDDDDK**V** 

**Restriction Sites:** 

Sgfl-Mlul

**Cloning Scheme:** 





GAT CTG GCA GCA AAT GAT ATC CTG GAT TAC AAG GAT GAC GAC GAT AAG GTT TAA ACGCCCGCCC

| D | L | A | A | N | D | I | L | D | Y | K | D | D | D | K | V | stop

<sup>\*</sup> The last codon before the Stop codon of the ORF



**OTI Disclaimer:** 

Due to the inherent nature of this plasmid, standard methods to replicate additional amounts of DNA in E. coli are highly likely to result in mutations and/or rearrangements. Therefore, OriGene does not guarantee the capability to replicate this plasmid DNA. Additional amounts of DNA can be purchased from OriGene with batch-specific, full-sequence verification at a reduced cost. Please contact our customer care team at <a href="mailto:customercom">customercom</a> or by calling 301.340.3188 option 3 for pricing and delivery.

The molecular sequence of this clone aligns with the gene accession number as a point of reference only. However, individual transcript sequences of the same gene can differ through naturally occurring variations (e.g. polymorphisms), each with its own valid existence. This clone is substantially in agreement with the reference, but a complete review of all prevailing variants is recommended prior to use. <u>More info</u>

OTI Annotation: This clone was engineered to express the complete ORF with an expression tag. Expression

varies depending on the nature of the gene.

**Components:** The ORF clone is ion-exchange column purified and shipped in a 2D barcoded Matrix tube

containing 10ug of transfection-ready, dried plasmid DNA (reconstitute with 100 ul of water).

**RefSeq:** NP 000134

**RefSeq Size**: 300 bp **RefSeq ORF**: 1533 bp

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:** 11 kDa

**Gene Summary:** 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]