

Fumarylacetoacetase (C-20): sc-66223

BACKGROUND

Fumarylacetoacetase is a 419 amino acid protein encoded by the human gene FAH. Fumarylacetoacetase catalyzes the hydrolysis of 4-fumarylacetoacetate, an intermediate in the metabolism of tyrosine, into acetoacetate and fumarate. Defects in FAH are the cause of tyrosinemia type I. It is an autosomal recessive inborn error of metabolism that occurs in both an acute and a chronic form. Clinical characteristics of the acute form include hepatic failure and death in infancy, whereas children with the chronic form have renal tubular dysfunction and hypophosphatemic rickets, progressive liver disease with development of hepatocellular carcinoma. Dietary treatment with restriction of tyrosine and phenylalanine alleviates the rickets, but liver transplantation has so far been the only definite treatment. Tyrosinemia type I is a rare condition, except in the Saguenay-lac-St-Jean region (province of Quebec, Canada) where the frequency is 1/1,846 newborns as the result of a founder effect.

REFERENCES

1. Phaneuf, D., et al. 1992. Type 1 hereditary tyrosinemia. Evidence for molecular heterogeneity and identification of a causal mutation in a French Canadian patient. *J. Clin. Invest.* 90: 1185-1192.
2. Bergeron, A., et al. 2001. Structural and functional analysis of missense mutations in fumarylacetoacetate hydrolase, the gene deficient in hereditary tyrosinemia type 1. *J. Biol. Chem.* 276: 15225-15231.
3. Dreumont, N., et al. 2004. Cytoplasmic nonsense-mediated mRNA decay for a nonsense (W262X) transcript of the gene responsible for hereditary tyrosinemia, fumarylacetoacetate hydrolase. *Biochem. Biophys. Res. Commun.* 324: 186-192.
4. Bergeron, A., et al. 2006. Involvement of endoplasmic reticulum stress in hereditary tyrosinemia type I. *J. Biol. Chem.* 281: 5329-5334.
5. Jacobs, S.M., et al. 2006. Kidneys of mice with hereditary tyrosinemia type I are extremely sensitive to cytotoxicity. *Pediatr. Res.* 59: 365-370.

CHROMOSOMAL LOCATION

Genetic locus: FAH (human) mapping to 15q23-q25; Fah (mouse) mapping to 7 D3.

STORAGE

Store at 4° C, ****DO NOT FREEZE****. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

PROTOCOLS

See our web site at www.scbt.com or our catalog for detailed protocols and support products.

SOURCE

Fumarylacetoacetase (C-20) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the C-terminus of Fumarylacetoacetase of human origin.

PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-66223 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

Fumarylacetoacetase (C-20) is recommended for detection of Fumarylacetoacetase of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1–2 µg per 100–500 µg of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Suitable for use as control antibody for Fumarylacetoacetase siRNA (h): sc-62356 and Fumarylacetoacetase siRNA (m): sc-62357.

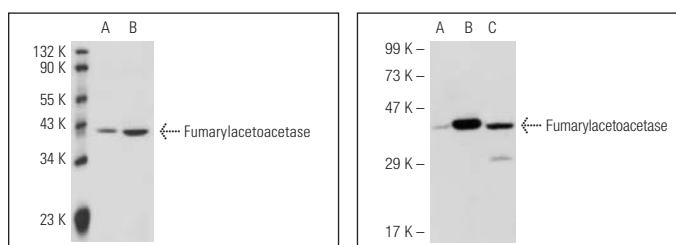
Molecular Weight of Fumarylacetoacetase: 46 kDa.

Positive Controls: Hep G2 cell lysate: sc-2227 or mouse liver extract: sc-2256.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) Immunofluorescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

DATA



Fumarylacetoacetase (C-20): sc-66223. Western blot analysis of Fumarylacetoacetase expression in Hep G2 whole cell lysate (A) and mouse liver tissue extract (B).

Fumarylacetoacetase (C-20): sc-66223. Western blot analysis of Fumarylacetoacetase expression in non-transfected 293T: sc-117752 (A), mouse Fumarylacetoacetase transfected 293T: sc-126870 (B) and Hep G2 (C) whole cell lysates.

RESEARCH USE

For research use only, not for use in diagnostic procedures.