SANTA CRUZ BIOTECHNOLOGY, INC.

Fumarylacetoacetase (m): 293T Lysate: sc-126870

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

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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.
6. Arranz, A., et al. 2006. Gene symbol: FAH. Disease: tyrosinaemia 1. Hum. Genet. 118: 537-537.
7. Langlois, C., et al. 2006. Evaluation of dichloroacetate treatment in a murine model of hereditary tyrosinemia type 1. Biochem. Pharmacol. 71: 1648-1661.
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CHROMOSOMAL LOCATION

Genetic locus: Fah (mouse) mapping to 7 D3.

PRODUCT

Fumarylacetoacetase (m): 293T Lysate represents a lysate of mouse Fumarylacetoacetase transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

APPLICATIONS

Fumarylacetoacetase (m): 293T Lysate is suitable as a Western Blotting positive control for mouse reactive Fumarylacetoacetase antibodies. Recommended use: 10-20 µl per lane.

Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

RESEARCH USE

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

PROTOCOLS

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