SANTA CRUZ BIOTECHNOLOGY, INC.

Fumarylacetoacetase siRNA (h): sc-62356

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The Power to Question

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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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.
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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.
8. McKiernan, P.J. 2006. Nitisinone in the treatment of hereditary tyrosinaemia type 1. Drugs 66: 743-750.

CHROMOSOMAL LOCATION

Genetic locus: FAH (human) mapping to 15q25.1.

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.

PRODUCT

Fumarylacetoacetase siRNA (h) is a pool of 3 target-specific 19-25 nt siRNAs designed to knock down gene expression. Each vial contains 3.3 nmol of lyophilized siRNA, sufficient for a 10 µM solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see Fumarylacetoacetase shRNA Plasmid (h): sc-62356-SH and Fumarylacetoacetase shRNA (h) Lentiviral Particles: sc-62356-V as alternate gene silencing products.

For independent verification of Fumarylacetoacetase (h) gene silencing results, we also provide the individual siRNA duplex components. Each is available as 3.3 nmol of lyophilized siRNA. These include: sc-62356A, sc-62356B and sc-62356C.

STORAGE AND RESUSPENSION

Store lyophilized siRNA duplex at -20° C with desiccant. Stable for at least one year from the date of shipment. Once resuspended, store at -20° C, avoid contact with RNAses and repeated freeze thaw cycles.

Resuspend lyophilized siRNA duplex in 330 µl of the RNAse-free water provided. Resuspension of the siRNA duplex in 330 µl of RNAse-free water makes a 10 µM solution in a 10 µM Tris-HCl, pH 8.0, 20 mM NaCl, 1 mM EDTA buffered solution.

APPLICATIONS

Fumarylacetoacetase siRNA (h) is recommended for the inhibition of Fumarylacetoacetase expression in human cells.

SUPPORT REAGENTS

For optimal siRNA transfection efficiency, Santa Cruz Biotechnology's siRNA Transfection Reagent: sc-29528 (0.3 ml), siRNA Transfection Medium: sc-36868 (20 ml) and siRNA Dilution Buffer: sc-29527 (1.5 ml) are recommended. Control siRNAs or Fluorescein Conjugated Control siRNAs are available as 10 µM in 66 µl. Each contain a scrambled sequence that will not lead to the specific degradation of any known cellular mRNA. Fluorescein Conjugated Control siRNAs include: sc-36869, sc-44239, sc-44240 and sc-44241. Control siRNAs include: sc-37007, sc-44230, sc-44231, sc-44232, sc-44233, sc-44234, sc-44235, sc-44236, sc-44237 and sc-44238.

RT-PCR REAGENTS

Semi-quantitative RT-PCR may be performed to monitor Fumarylacetoacetase gene expression knockdown using RT-PCR Primer: Fumarylacetoacetase (h)-PR: sc-62356-PR (20 µl). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.