SANTA CRUZ BIOTECHNOLOGY, INC.

DPYS (h): 293T Lysate: sc-114343

BACKGROUND

DPYS (dihydropyrimidinase), also known as DHPase, hydantoinase or DHP, is a 519 amino acid protein that is expressed in liver and kidney tissue and belongs to the DHOase family. Functioning as a homotetramer, DPYS uses zinc as a cofactor to catalyze the second step of reductive pyrimidine degradation, namely the conversion of 5,6-dihydrouracil to 3-ureidopropionate. DPYS is subject to post-translational carbamylation, an event which enhances the ability of DPYS to bind zinc ions. Defects in the gene encoding DPYS are the cause of DHP deficiency, an autosomal recessive disorder that is characterized by epileptic or convulsive attacks, dysmorphic features and severe developmental delay and congenital microvillous atrophy.

REFERENCES

1. Berger, R., et al. 1984. Dihydropyrimidine dehydrogenase deficiency leading to thymine-uraciluria. An inborn error of pyrimidine metabolism. Clin. Chim. Acta 141: 227-234.
2. Duran, M., et al. 1991. Dihydropyrimidinuria: a new inborn error of pyrimidine metabolism. J. Inherit. Metab. Dis. 14: 367-370.
3. Hamajima, N., et al. 1996. A novel gene family defined by human dihydropyrimidinase and three related proteins with differential tissue distribution. Gene 180: 157-163.
4. van Gennip, A.H., et al. 1997. Dihydropyrimidinase deficiency: confirmation of the enzyme defect in dihydropyrimidinuria. J. Inherit. Metab. Dis. 20: 339-342.
5. Assmann, B., et al. 1997. Dihydropyrimidinase deficiency and congenital microvillous atrophy: coincidence or genetic relation? J. Inherit. Metab. Dis. 20: 681-688.
6. Hamajima, N., et al. 1998. Dihydropyrimidinase deficiency: structural organization, chromosomal localization, and mutation analysis of the human dihydropyrimidinase gene. Am. J. Hum. Genet. 63: 717-726.
7. Online Mendelian Inheritance in Man, OMIM™. 2006. Johns Hopkins University, Baltimore, MD. MIM Number: 222748. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/
8. van Kuilenburg, A.B., et al. 2007. Clinical, biochemical and genetic findings in two siblings with a dihydropyrimidinase deficiency. Mol. Genet. Metab. 91: 157-164.
9. Thomas, H.R., et al. 2007. Genetic regulation of dihydropyrimidinase and its possible implication in altered uracil catabolism. Pharmacogenet. Genomics 17: 973-987.

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.

PROTOCOLS

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

CHROMOSOMAL LOCATION

Genetic locus: DPYS (human) mapping to 8q22.3.

PRODUCT

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

APPLICATIONS

DPYS (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive DPYS 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.