



SZABO SCANDIC

Part of Europa Biosite

Produktinformation



Forschungsprodukte & Biochemikalien



Zellkultur & Verbrauchsmaterial



Diagnostik & molekulare Diagnostik



Laborgeräte & Service

Weitere Information auf den folgenden Seiten!
See the following pages for more information!



Lieferung & Zahlungsart

siehe unsere [Liefer- und Versandbedingungen](#)

Zuschläge

- Mindermengenzuschlag
- Trockeneiszuschlag
- Gefahrgutzuschlag
- Expressversand

SZABO-SCANDIC HandelsgmbH

Quellenstraße 110, A-1100 Wien

T. +43(0)1 489 3961-0

F. +43(0)1 489 3961-7

mail@szabo-scandic.com

www.szabo-scandic.com

[linkedin.com/company/szaboscandic](https://www.linkedin.com/company/szaboscandic) 

PHYH (m): 293T Lysate: sc-127330

BACKGROUND

PHYH (phytanoyl-CoA 2-hydroxylase), also known as RD, LN1, PAHX or LNAP1, is a 338 amino acid protein that localizes to the peroxisome and plays an important role in fatty acid metabolism. Expressed in kidney, liver and T cells, PHYH uses iron and ascorbate as cofactors to catalyze the conversion of phytanoyl-CoA to 2-hydroxyphytanoyl-CoA, a reaction that is involved in the α -oxidation of 3-methyl branched fatty acids. Defects in the gene encoding PHYH are associated with Refsum disease (RD), an autosomal recessive disorder that is characterized by retinitis pigmentosa, peripheral neuropathy, cerebellar ataxia, nerve deafness, anosmia, skeletal abnormalities, ichthyosis, cataracts and cardiac impairment, all of which usually develop during the second or third decade of life.

REFERENCES

- Jansen, G.A., Wanders, R.J., Watkins, P.A. and Mihalik, S.J. 1997. Phytanoyl-coenzyme A hydroxylase deficiency — the enzyme defect in Refsum's disease. *N. Engl. J. Med.* 337: 133-134.
- Mihalik, S.J., Morrell, J.C., Kim, D., Sacksteder, K.A., Watkins, P.A. and Gould, S.J. 1997. Identification of PAHX, a Refsum disease gene. *Nat. Genet.* 17: 185-189.
- Jansen, G.A., Ferdinandusse, S., Hogenhout, E.M., Verhoeven, N.M., Jakobs, C. and Wanders, R.J. 1999. Phytanoyl-CoA hydroxylase deficiency. Enzymological and molecular basis of classical Refsum disease. *Adv. Exp. Med. Biol.* 466: 371-376.
- Mukherji, M., Chien, W., Kershaw, N.J., Clifton, I.J., Schofield, C.J., Wierzbicki, A.S. and Lloyd, M.D. 2001. Structure-function analysis of phytanoyl-CoA 2-hydroxylase mutations causing Refsum's disease. *Hum. Mol. Genet.* 10: 1971-1982.
- Kee, H.J., Koh, J.T., Yang, S.Y., Lee, Z.H., Baik, Y.H. and Kim, K.K. 2003. A novel murine long-chain acyl-CoA synthetase expressed in brain participates in neuronal cell proliferation. *Biochem. Biophys. Res. Commun.* 305: 925-933.
- Foulon, V., Asselberghs, S., Geens, W., Mannaerts, G.P., Casteels, M. and Van Veldhoven, P.P. 2003. Further studies on the substrate spectrum of phytanoyl-CoA hydroxylase: implications for Refsum disease? *J. Lipid Res.* 44: 2349-2355.
- Jansen, G.A., Waterham, H.R. and Wanders, R.J. 2004. Molecular basis of Refsum disease: sequence variations in phytanoyl-CoA hydroxylase (PHYH) and the PTS2 receptor (PEX7). *Hum. Mutat.* 23: 209-218.
- McDonough, M.A., Kavanagh, K.L., Butler, D., Searls, T., Oppermann, U. and Schofield, C.J. 2005. Structure of human phytanoyl-CoA 2-hydroxylase identifies molecular mechanisms of Refsum disease. *J. Biol. Chem.* 280: 41101-41110.
- Searls, T., Butler, D., Chien, W., Mukherji, M., Lloyd, M.D. and Schofield, C.J. 2005. Studies on the specificity of unprocessed and mature forms of phytanoyl-CoA 2-hydroxylase and mutation of the iron binding ligands. *J. Lipid Res.* 46: 1660-1667.

CHROMOSOMAL LOCATION

Genetic locus: *Phyh* (mouse) mapping to 2 A1.

PRODUCT

PHYH (m): 293T Lysate represents a lysate of mouse PHYH transfected 293T cells and is provided as 100 μ g protein in 200 μ l SDS-PAGE buffer.

APPLICATIONS

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

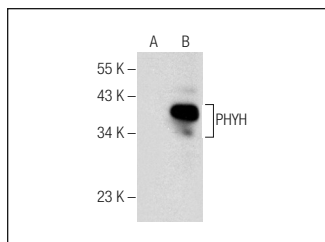
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

PHYH (E-8): sc-376727 is recommended as a positive control antibody for Western Blot analysis of enhanced mouse PHYH expression in PHYH transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgG κ BP-HRP: sc-516102 or m-IgG κ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048.

DATA



PHYH (E-8): sc-376727. Western blot analysis of PHYH expression in non-transfected: sc-117752 (A) and mouse PHYH transfected: sc-127330 (B) 293T whole cell lysates.

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.

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.