

UROD (h2): 293T Lysate: sc-170721

BACKGROUND

Uroporphyrinogen decarboxylase, also known as UROD or UPD, is a 367 amino acid protein that exists as a homodimer. UROD is the fifth enzyme in the human heme biosynthetic pathway and is responsible for the conversion of uroporphyrinogen to coproporphyrinogen through the removal of four carboxymethyl side chains. Mutations in the UROD gene are responsible for three autosomal disorders in humans: familial porphyria cutanea tarda (f-PCT), sporadic porphyria cutanea tarda (s-PCT) and hepatoerythropoietic porphyria (HEP). F-PCT is an autosomal dominant disorder characterized by late-onset light-sensitive dermatitis. High levels of uroporphyrin excretion in the urine and varying degrees of liver damage are associated with this disease. S-PCT is an idiosyncratic form of PCT that is characterized by a reduction of liver enzymes. HEP is an autosomal recessive disorder that affects infants. It is characterized by excessive excretion of acetate-substituted porphyrins and accumulation of protoporphyrin in erythrocytes.

REFERENCES

1. Moran-Jimenez, M.J., et al. 1996. Uroporphyrinogen decarboxylase: complete human gene sequence and molecular study of three families with hepatoerythropoietic porphyria. *Am. J. Hum. Genet.* 58: 712-721.
2. Phillips, J.D., et al. 1997. Characterization and crystallization of human uroporphyrinogen decarboxylase. *Protein Sci.* 6: 1343-1346.
3. Akhtar, R.A. and Smith, A.G. 1998. Chromosomal linkage analysis of porphyria in mice induced by hexachlorobenzene-iron synergism: a model of sporadic porphyria cutanea tarda. *Pharmacogenetics* 8: 485-494.
4. Christiansen, L., et al. 1999. Screening for mutations in the uroporphyrinogen decarboxylase gene using denaturing gradient gel electrophoresis. Identification and characterization of six novel mutations associated with familial PCT. *Hum. Mutat.* 14: 222-232.
5. Phillips, J.D., et al. 2001. Functional consequences of naturally occurring mutations in human uroporphyrinogen decarboxylase. *Blood* 98: 3179-3185.
6. Online Mendelian Inheritance in Man, OMIM™. 2002. Johns Hopkins University, Baltimore, MD. MIM Number: 176100. World Wide Web URL: <http://www.ncbi.nlm.nih.gov/omim/>

CHROMOSOMAL LOCATION

Genetic locus: UROD (human) mapping to 1p34.1.

PRODUCT

UROD (h2): 293T Lysate represents a lysate of human UROD 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.

PROTOCOLS

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

APPLICATIONS

UROD (h2): 293T Lysate is suitable as a Western Blotting positive control for human reactive UROD antibodies. Recommended use: 10-20 µl per lane.

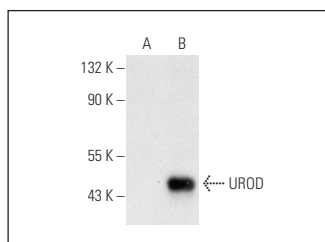
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

UROD (C-4): sc-365297 is recommended as a positive control antibody for Western Blot analysis of enhanced human UROD expression in UROD transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

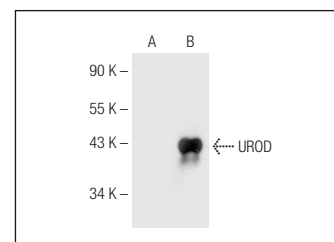
RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended:
1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ 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



UROD (C-4): sc-365297. Western blot analysis of UROD expression in non-transfected: sc-117752 (A) and human UROD transfected: sc-170721 (B) 293T whole cell lysates.



UROD (G-7): sc-374318. Western blot analysis of UROD expression in non-transfected: sc-117752 (A) and human UROD transfected: sc-170721 (B) 293T whole cell lysates.

RESEARCH USE

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