SANTA CRUZ BIOTECHNOLOGY, INC.

PHEX (C-12): sc-47323

BACKGROUND

The PHEX gene (phosphate regulating gene with homologies to endopeptidases on the X chromosome) encodes a 749-amino acid protein that putatively consists of an intracellular, transmembrane, and extracellular domain. PHEX mutations have been observed in 60-80% of hypophosphatemic rickets patients. The PHEX protein, which is a single-pass membrane protein, is also designated HYP, X-linked hypophosphatemia protein or metalloendo peptidase homolog PEX. PHEX plays an active role in bone and dentin mineralization and renal phosphate re-absorption. X-linked hypophosphatemic rickets, also designated HYP, is an X-linked dominant disorder characterized by impaired phosphate uptake in the kidney, which is likely to be caused by abnormal regulation of sodium phosphate cotransport in the proximal tubules. Clinical manifestations include skeletal deformities, growth failure, craniosynostosis, paravertebral calcifications, pseudofractures in lower extremities, and muscular hypotonia with onset in early childhood.

REFERENCES

1. Francis, F., Strom, T.M., Hennig, S., Boddrich, A., Lorenz, B., Brandau, O., Mohnike, K.L., Cagnoli, M., Steffens, C., Klages, S., Borzym, K., Pohl, T., Oudet, C., Econs, M.J., Rowe, P.S., Reinhardt, R., Meitinger, T. and Lehrach, H. 1997. Genomic organization of the human PEX gene mutated in X-linked dominant hypophosphatemic rickets. Genome Res. 7: 573-585.
2. Guo, R. and Quarles, L.D. 1997. Cloning and sequencing of human PEX from a bone cDNA library: evidence for its developmental stage-specific regulation in osteoblasts. J. Bone. Miner. Res. 12: 1009-1017.
3, Sato, K., Tajima, T., Nakae, J., Adachi, M., Asakura, Y., Tachibana, K., Suwa, S., Katsumata, N., Tanaka, T., Hayashi, Y., Abe, S., Murashita, M., Okuhara, K., Shinohara, N. and Fujieda, K. 2000. Three novel PHEX gene mutations in Japanese patients with X-linked hypophosphatemic rickets. Pediatr. Res. 48: 536-540.
4. Tyynismaa, H., Kaitila, I., Nanto-Salonen, K., Ala-Houhala, M. and Alitalo, T. 2000. Identification of fifteen novel PHEX gene mutations in Finnish patients with hypophosphatemic rickets. Hum. Mutat. 15: 383-384.
5. Tanaka, H. 2001. Hypophosphatemia and rickets/osteomalacia. Clin. Calcium 11: 1282-1289.
6. Liu, S., Guo, R., Simpson, L.G., Xiao, Z.S., Burnham, C.E. and Quarles, L.D. 2003. Regulation of fibroblastic growth factor 23 expression but not degradation by PHEX. J. Biol. Chem. 278: 37419-37426.
7. Liu, S., Brown, T.A., Zhou, J., Xiao, Z.S., Awad, H., Guilak, F., Quarles, L.D. 2005. Role of matrix extracellular phosphoglycoprotein in the pathogenesis of X-linked hypophosphatemia. J. Am. Soc. Nephrol. 16: 1645-1653.
8. Chou, Y.Y., Chao, S.C., Tsai, S.C., Lin, S.J. 2005. Novel PHEX gene mutations in two Taiwanese patients with hypophosphatemic rickets. J. Formos. Med. Assoc. 104: 198-202.

CHROMOSOMAL LOCATION

Genetic locus: PHEX (human) mapping to Xp22.11; Phex (mouse) mapping to X F4.

SOURCE

PHEX (C-12) is an affinity purified goat polyclonal antibody raised against a peptide mapping at the C-terminus of PHEX of human origin.

PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-47323 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

PHEX (C-12) is recommended for detection of PHEX of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

PHEX (C-12) is also recommended for detection of PHEX in additional species, including equine and canine.

Suitable for use as control antibody for PHEX siRNA (h): sc-61334, PHEX siRNA (m): sc-61335, PHEX shRNA Plasmid (h): sc-61334-SH, PHEX shRNA Plasmid (m): sc-61335-SH, PHEX shRNA (h) Lentiviral Particles: sc-61334-V and PHEX shRNA (m) Lentiviral Particles: sc-61335-V.

Molecular Weight of PHEX: 97 kDa.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluorescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

STORAGE

Store at 4° C, **DO NOT FREEZE**. Stable for one year from the date of shipment. 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 or our catalog for detailed protocols and support products.