

CDKL5 (h): 293T Lysate: sc-173758

BACKGROUND

Cell cycle progression is controlled in part by a family of cyclin proteins and cyclin-dependent kinases (Cdks). Cdk proteins work in concert with the cyclins to phosphorylate key substrates involved in each phase of cell cycle progression. Another family of proteins, Cdk inhibitors, also plays a role in regulating the cell cycle by binding to cyclin-Cdk complexes and modulating their activity. CDKL5 (cyclin-dependent kinase-like 5) is a 1,030 amino acid protein that belongs to the CMGC Ser/Thr protein kinase family. Expressed in brain, lung, kidney, prostate, ovary, placenta, pancreas and testis, CDKL5 is thought to play a role in cell cycle regulation. Defects in CDKL5 are a cause of several disorders, such as X-linked infantile spasm syndrome and Rett syndrome.

REFERENCES

1. Tao, J., et al. 2004. Mutations in the X-linked cyclin-dependent kinase-like 5 (CDKL5/STK9) gene are associated with severe neurodevelopmental retardation. *Am. J. Hum. Genet.* 75: 1149-1154.
2. Buoni, S., et al. 2006. Myoclonic encephalopathy in the CDKL5 gene mutation. *Clin. Neurophysiol.* 117: 223-227.
3. Nectoux, J., et al. 2006. Maternal origin of a novel C-terminal truncation mutation in CDKL5 causing a severe atypical form of Rett syndrome. *Clin. Genet.* 70: 29-33.
4. Bertani, I., et al. 2006. Functional consequences of mutations in CDKL5, an X-linked gene involved in infantile spasms and mental retardation. *J. Biol. Chem.* 281: 32048-32056.
5. Archer, H.L., et al. 2006. CDKL5 mutations cause infantile spasms, early onset seizures, and severe mental retardation in female patients. *J. Med. Genet.* 43: 729-734.
6. Van Esch, H., et al. 2007. Encephalopathy and bilateral cataract in a boy with an interstitial deletion of Xp22 comprising the CDKL5 and NHS genes. *Am. J. Med. Genet. A* 143A: 364-369.
7. Grosso, S., et al. 2007. Seizures and electroencephalographic findings in CDKL5 mutations: case report and review. *Brain Dev.* 29: 239-242.
8. Bahi-Buisson, N., et al. 2008. The three stages of epilepsy in patients with CDKL5 mutations. *Epilepsia* 49: 1027-1037.

CHROMOSOMAL LOCATION

Genetic locus: CDKL5 (human) mapping to Xp22.13.

PRODUCT

CDKL5 (h): 293T Lysate represents a lysate of human CDKL5 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.

RESEARCH USE

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

APPLICATIONS

CDKL5 (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive CDKL5 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.

PROTOCOLS

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