



**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](mailto:mail@szabo-scandic.com)

[www.szabo-scandic.com](http://www.szabo-scandic.com)

[linkedin.com/company/szaboscandic](http://linkedin.com/company/szaboscandic)



# CLN5 (h5): 293T Lysate: sc-372537

## BACKGROUND

Neuronal ceroid-lipofuscinose (NCL), also designated Batten disease, comprises a group of recessively inherited, progressive neurodegenerative diseases found in children. NCL is characterized by atrophy of the brain and an accumulation of lysosome derived fluorescent bodies found in many cells, especially neurons. Symptoms of NCL include a failure of psychomotor development, seizures, impaired vision and premature death. The eight genes/proteins associated with NCL are designated CLN1-CLN8. Mutations in six of these genes results in a distinct type of NCL-disease; the six genes/proteins are CLN1 (encoding PPT1, a protein thiolesterase), CLN2 (encoding the serine protease TPP1), CLN3, CLN5, CLN6 and CLN8. A single base duplication mutation in dog and cow CLN5 has been shown to cause NCL.

## REFERENCES

1. Nardocci, N. and Cardona, F. 1998. Neuronal ceroid lipofuscinoses: a review. *Ital. J. Neurol. Sci.* 19: 271-276.
2. Wisniewski, K.E., Kida, E., Connell, F. and Zhong, N. 2000. Neuronal ceroid lipofuscinoses: research update. *Neurol. Sci.* 21: S49-S56.
3. Zhong, N. 2000. Neuronal ceroid lipofuscinoses and possible pathogenic mechanism. *Mol. Genet. Metab.* 71: 195-206.
4. Heinonen, O., Salonen, T., Jalanko, A., Peltonen, L. and Copp, A. 2000. CLN1 and CLN5, genes for infantile and variant late infantile neuronal ceroid lipofuscinoses, are expressed in the embryonic human brain. *J. Comp. Neurol.* 426: 406-412.
5. Wisniewski, K.E., Zhong, N. and Philipart, M. 2001. Pheno/genotypic correlations of neuronal ceroid lipofuscinoses. *Neurology* 57: 576-581.
6. Ranta, S., Savukoski, M., Santavuori, P. and Haltia, M. 2001. Studies of homogenous populations: CLN5 and CLN8. *Adv. Genet.* 45: 123-140.
7. Melville, S.A., Wilson, C.L., Chiang, C.S., Studdert, V.P., Lingaas, F. and Wilton, A.N. 2005. A mutation in canine CLN5 causes neuronal ceroid lipofuscinosis in Border collie dogs. *Genomics* 86: 287-294.
8. Mole, S.E., Williams, R.E. and Goebel, H.H. 2005. Correlations between genotype, ultrastructural morphology and clinical phenotype in the neuronal ceroid lipofuscinoses. *Neurogenetics* 63: 107-126.
9. Pineda-Trujillo, N., Cornejo, W., Carrizosa, J., Wheeler, R.B., Munera, S., Valencia, A., Agudelo-Arango, J., Cogollo, A., Anderson, G., Bedoya, G., Mole, S.E. and Ruiz-Linares, A. 2005. A CLN5 mutation causing an atypical neuronal ceroid lipofuscinosis of juvenile onset. *Neurology* 64: 740-742.

## CHROMOSOMAL LOCATION

Genetic locus: CLN5 (human) mapping to 13q22.3.

## PRODUCT

CLN5 (h5): 293T Lysate represents a lysate of human CLN5 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

## RESEARCH USE

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

## APPLICATIONS

CLN5 (h5): 293T Lysate is suitable as a Western Blotting positive control for human reactive CLN5 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.

## 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](http://www.scbt.com) for detailed protocols and support products.