

Endoglin (m): 293T Lysate: sc-126793

BACKGROUND

Hereditary hemorrhagic telangiectasia (HHT) is an autosomal dominant disorder characterized by vascular abnormalities such as dilated vessels, hemorrhages, liver and lung congestion, and brain or heart ischemia. Mutations in two genes, Endoglin (also designated CD105) and ALK-1 (activin receptor-like kinase-1, also designated TGF β superfamily RI), are responsible for HHT. Endoglin is mutated in HHT1 and ALK-1 is mutated in HHT2, both of which are thought to be caused by haploinsufficiency. Endoglin and ALK-1 are type III and type I members of the TGF β receptor superfamily, respectively, that are expressed on vascular endothelial cells. Endoglin can only bind ligands of the TGF β superfamily via association with the respective ligand binding receptors for TGF β 1, TGF β 3, Activin A, BMP-2 and BMP-7. The human ALK-1 gene encodes two protein species which exist as a result of either glycosylation or alternative splicing events. ALK-1 preferentially binds TGF β 1 and is expressed in bone marrow stromal cells, lung, brain, kidney and spleen.

REFERENCES

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3. Gallione, C.J., et al. 1998. Mutation and expression analysis of the Endoglin gene in hereditary hemorrhagic telangiectasia reveals null alleles. *Hum. Mutat.* 11: 286-294.
4. Klaus, D.J., et al. 1998. Novel missense and frameshift mutations in the Activin receptor-like kinase-1 gene in hereditary hemorrhagic telangiectasia. *Mutations in brief no. 164. Online. Hum. Mutat.* 12: 137.
5. Bourdeau, A., et al. 2000. Endoglin-deficient mice, a unique model to study hereditary hemorrhagic telangiectasia. *Trends Cardiovasc. Med.* 10: 279-285.
6. Azuma, H. 2000. Genetic and molecular pathogenesis of hereditary hemorrhagic telangiectasia. *J. Med. Invest.* 47: 81-90.
7. Gallione, C.J., et al. 2000. Two common Endoglin mutations in families with hereditary hemorrhagic telangiectasia in the Netherlands Antilles: evidence for a founder effect. *Hum. Genet.* 107: 40-44.
8. Bourdeau, A., et al. 2001. Potential role of modifier genes influencing transforming growth factor β 1 levels in the development of vascular defects in Endoglin heterozygous mice with hereditary hemorrhagic telangiectasia. *Am. J. Pathol.* 158: 2011-2020.
9. Howe, J.R., et al. 2007. ENG mutations in MADH4/BMPRI-A mutation negative patients with juvenile polyposis. *Clin. Genet.* 71: 91-92.

CHROMOSOMAL LOCATION

Genetic locus: Eng (mouse) mapping to 2 B.

RESEARCH USE

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

PRODUCT

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

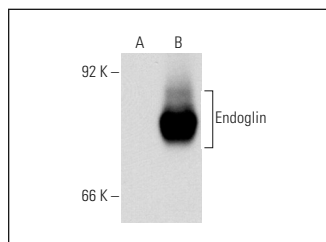
APPLICATIONS

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

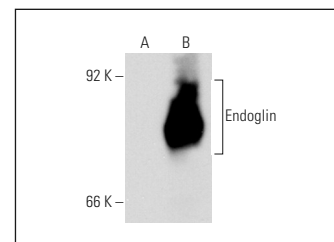
Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

Endoglin (RM0030-6J9): sc-101443 is recommended as a positive control antibody for Western Blot analysis of enhanced mouse Endoglin expression in Endoglin transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

DATA



Endoglin (RM0030-6J9): sc-101443. Western blot analysis of Endoglin expression in non-transfected: sc-117752 (A) and mouse Endoglin transfected: sc-126793 (B) 293T whole cell lysates.



Endoglin (201707): sc-71042. Western blot analysis of Endoglin expression in non-transfected: sc-117752 (A) and mouse Endoglin transfected: sc-126793 (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.

PROTOCOLS

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