

Endoglin (h): 293T Lysate: sc-170141

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

1. Wu, X., et al. 1995. Cloning and characterization of the murine Activin receptor like kinase-1 (ALK-1) homolog. *Biochem. Biophys. Res. Commun.* 216: 78-83.
2. Altomonte, M., et al. 1996. Expression and structural features of Endoglin (CD105), a transforming growth factor β 1 and β 3 binding protein, in human melanoma. *Br. J. Cancer* 74: 1586-1591.
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. 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 (human) mapping to 9q34.11.

RESEARCH USE

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

PRODUCT

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

APPLICATIONS

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

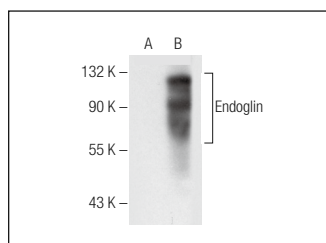
Endoglin (A-8): sc-376381 is recommended as a positive control antibody for Western Blot analysis of enhanced human Endoglin expression in Endoglin transfected 293T cells (starting dilution 1:100, dilution range 1:100-1:1,000).

RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended:

1) Western Blotting: use m-IgG κ BP-HRP: sc-516102 or m-IgG κ 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



Endoglin (A-8): sc-376381. Western blot analysis of Endoglin expression in non-transfected: sc-117752 (A) and human Endoglin transfected: sc-170141 (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.