



Endoglin (SN6/N1-3A1): sc-80539

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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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.
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CHROMOSOMAL LOCATION

Genetic locus: ENG (human) mapping to 9q34.11.

RESEARCH USE

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

SOURCE

Endoglin (SN6/N1-3A1) is a mouse monoclonal antibody raised against partially purified cell membrane antigens of human origin.

PRODUCT

Each vial contains 200 μ g IgG₁ in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Available as phycoerythrin (sc-80539 PE) or fluorescein (sc-80539 FITC) conjugates for flow cytometry, 100 tests.

APPLICATIONS

Endoglin (SN6/N1-3A1) is recommended for detection of Endoglin cell surface antigen of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 μ g per 100-500 μ g of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and flow cytometry (1 μ g per 1 x 10⁶ cells).

Suitable for use as control antibody for Endoglin siRNA (h): sc-35302, Endoglin shRNA Plasmid (h): sc-35302-SH and Endoglin shRNA (h) Lentiviral Particles: sc-35302-V.

Molecular Weight of reduced Endoglin: 84 kDa.

Molecular Weight of non-reduced Endoglin: 130 kDa.

Positive Controls: HL-60 whole cell lysate: sc-2209, HEL 92.1.7 cell lysate: sc-2270 or HUV-EC-C cells.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use goat anti-mouse IgG-HRP: sc-2005 (dilution range: 1:2000-1:32,000) or Cruz Marker™ compatible goat anti-mouse IgG-HRP: sc-2031 (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) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) Immunofluorescence: use goat anti-mouse IgG-FITC: sc-2010 (dilution range: 1:100-1:400) or goat anti-mouse IgG-TR: sc-2781 (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.

PROTOCOLS

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