

ALK-1 (MM0015-8G33): sc-101555

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., Robinson, C.E., Fong, H.W., Crabtree, J.S., Rodriguez, B.R., Roe, B.A. and Gimble, J.M. 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., Montagner, R., Fonsatti, E., Colizzi, F., Cattarossi, I., Brasoveanu, L.I., Nicotra, M.R., Cattelan, A., Natali, P.G. and Maio, M. 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., Klaus, D.J., Yeh, E.Y., Stenzel, T.T., Xue, Y., Anthony, K.B., McAllister, K.A., Baldwin, M.A., Berg, J.N., Lux, A., Smith, J.D., Vary, C.P., Craigen, W.J., Westermann, C.J., Warner, M.L., Miller, Y.E., Jackson, C.E., Gutmacher, A.E. and Marchuk, D.A. 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., Gallione, C.J., Anthony, K., Yeh, E.Y., Yu, J., Lux, A., Johnson, D.W. and Marchuk, D.A. 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., Faughnan, M.E. and Letarte, M. 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. Bourdeau, A., Faughnan, M.E., McDonald, M.L., Paterson, A.D., Wanless, I.R. and Letarte, M. 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.

CHROMOSOMAL LOCATION

Genetic locus: ACVRL1 (human) mapping to 12q13.13.

SOURCE

ALK-1 (MM0015-8G33) is a mouse monoclonal antibody raised against the extracellular domain of ALK-1 of human origin.

PRODUCT

Each vial contains 100 μ g IgG₂ in 1.0 ml PBS with < 0.1% sodium azide and 0.1% gelatin.

APPLICATIONS

ALK-1 (MM0015-8G33) is recommended for detection of ALK-1 of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and immunohistochemistry (including paraffin-embedded sections) (starting dilution 1:50, dilution range 1:50-1:500).

Suitable for use as control antibody for ALK-1 siRNA (h): sc-40212, ALK-1 shRNA Plasmid (h): sc-40212-SH and ALK-1 shRNA (h) Lentiviral Particles: sc-40212-V.

Molecular Weight of ALK-1: 53 kDa.

Positive Controls: human platelet extract: sc-363773.

STORAGE

Store at 4° C, **DO NOT FREEZE**. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

RESEARCH USE

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

PROTOCOLS

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