

GEN1 (S-14): sc-167970

BACKGROUND

GEN1 (flap endonuclease GEN homolog 1) is a 908 amino acid nuclear protein that belongs to the XPG/RAD2 endonuclease family and GEN subfamily. GEN1 cleaves flap structures at the junction between single-stranded DNA and double-stranded DNA and binds two magnesium ions per subunit. The gene encoding GEN1 maps to human chromosome 2, which consists of 237 million bases, encodes over 1,400 genes and makes up approximately 8% of the human genome. A number of genetic diseases are linked to genes on chromosome 2. Harlequin ichthyosis, a rare and morbid skin deformity, is associated with mutations in the ABCA12 gene. The lipid metabolic disorder sitosterolemia is associated with ABCG5 and ABCG8. An extremely rare recessive genetic disorder, Alstrom syndrome is due to mutations in the ALMS1 gene.

REFERENCES

- Ijdo, J.W., Baldini, A., Ward, D.C., Reeders, S.T. and Wells, R.A. 1991. Origin of human chromosome 2: an ancestral telomere-telomere fusion. *Proc. Natl. Acad. Sci. USA* 88: 9051-9055.
- Avarello, R., Pedicini, A., Caiulo, A., Zuffardi, O. and Fraccaro, M. 1992. Evidence for an ancestral alphoid domain on the long arm of human chromosome 2. *Hum. Genet.* 89: 247-249.
- Ishikawa, G., Kanai, Y., Takata, K., Takeuchi, R., Shimanouchi, K., Ruike, T., Furukawa, T., Kimura, S. and Sakaguchi, K. 2004. DmGEN, a novel RAD2 family endo-exonuclease from *Drosophila melanogaster*. *Nucleic Acids Res.* 32: 6251-6259.
- Hillier, L.W., Graves, T.A., Fulton, R.S., Fulton, L.A., Pepin, K.H., Minx, P., Wagner-McPherson, C., Layman, D., Wylie, K., Sekhon, M., Becker, M.C., Fewell, G.A., Delehaunty, K.D., Miner, T.L., Nash, W.E., Krenitzki, C., Oddy, L., Du, H., Sun, H., Bradshaw-Cordum, H., Ali, J., Carter, J., Cordes, M., Harris, A., Isak, A., van Brunt, A., Nguyen, C., et al. 2005. Generation and annotation of the DNA sequences of human chromosomes 2 and 4. *Nature* 434: 724-731.
- Thomas, A.C., Cullup, T., Norgett, E.E., Hill, T., Barton, S., Dale, B.A., Sprecher, E., Sheridan, E., Taylor, A.E., Wilroy, R.S., DeLozier, C., Burrows, N., Goodyear, H., Fleckman, P., Stephens, K.G., Mehta, L., Watson, R.M., Graham, R., Wolf, R., Slavotinek, A., Martin, M., Bourn, D., Mein, C.A., O'Toole, E.A. and Kelsell, D.P. 2006. ABCA12 is the major harlequin ichthyosis gene. *J. Invest. Dermatol.* 126: 2408-2413.
- Akiyama, M., Sakai, K., Sato, T., McMillan, J.R., Goto, M., Sawamura, D. and Shimizu, H. 2007. Compound heterozygous ABCA12 mutations including a novel nonsense mutation underlie harlequin ichthyosis. *Dermatology* 215: 155-159.
- Marshall, J.D., Beck, S., Maffei, P. and Naggert, J.K. 2007. Alström syndrome. *Eur. J. Hum. Genet.* 15: 1193-1202.
- Marshall, J.D., Hinman, E.G., Collin, G.B., Beck, S., Cerqueira, R., Maffei, P., Milan, G., Zhang, W., Wilson, D.I., Hearn, T., Tavares, P., Vettor, R., Veronese, C., Martin, M., So, W.V., Nishina, P.M. and Naggert, J.K. 2007. Spectrum of ALMS1 variants and evaluation of genotype-phenotype correlations in Alström syndrome. *Hum. Mutat.* 28: 1114-1123.

CHROMOSOMAL LOCATION

Genetic locus: GEN1 (human) mapping to 2p24.2; Gen1 (mouse) mapping to 12 A1.1.

SOURCE

GEN1 (S-14) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of GEN1 of human origin.

PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-167970 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

Available as TransCruz reagent for Gel Supershift and ChIP applications, sc-167970 X, 200 µg/0.1 ml.

APPLICATIONS

GEN1 (S-14) is recommended for detection of GEN1 of mouse, rat and 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 solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

GEN1 (S-14) is also recommended for detection of GEN1 in additional species, including equine.

Suitable for use as control antibody for GEN1 siRNA (h): sc-94802, GEN1 siRNA (m): sc-145381, GEN1 shRNA Plasmid (h): sc-94802-SH, GEN1 shRNA Plasmid (m): sc-145381-SH, GEN1 shRNA (h) Lentiviral Particles: sc-94802-V and GEN1 shRNA (m) Lentiviral Particles: sc-145381-V.

GEN1 (G-14) X TransCruz antibody is recommended for Gel Supershift and ChIP applications.

Molecular Weight of GEN1: 103 kDa.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (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) Immunofluorescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (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.

RESEARCH USE

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