# connexin 30 (h): 293T Lysate: sc-115048



The Power to Question

# **BACKGROUND**

The connexin family of proteins form hexameric complexes called connexons that facilitate movement of low molecular weight proteins between cells via GAP junctions. Connexin proteins share a common topology of four transmembrane  $\alpha$ -helical domains, two extracellular loops, a cytoplasmic loop and cytoplasmic N- and C-termini. Many of the key functional differences between connexins arise from specific amino-acid substitutions in the most highly conserved domains: the transmembrane and extracellular regions. Connexin 30, also known as GJB6 (GAP junction  $\beta$  6), ED2, EDH, HED or DFNA3, is a 261 amino acid multi-pass membrane protein that localizes to the cell junction and belongs to the connexin family. Functioning as a hexamer with other connexin proteins, connexin 30 facilitates the diffusion of low molecular weight materials from one cell to another. Defects in the gene encoding connexin 30 are the cause of ectodermal dysplasia type 2 (ED2) and non-syndromatic sensorineural deafness autosomal dominant type 3 (DFNA3), the former of which is characterized by abnormal development of ectodermal structures (such as skin and nails).

# **REFERENCES**

- Kelley, P.M., Abe, S., Askew, J.W., Smith, S.D., Usami, S. and Kimberling, W.J. 1999. Human connexin 30 (GJB6), a candidate gene for nonsyndromic hearing loss: molecular cloning, tissue-specific expression, and assignment to chromosome 13q12. Genomics 62: 172-176.
- Smith, F.J., Morley, S.M. and McLean, W.H. 2002. A novel connexin 30 mutation in Clouston syndrome. J. Invest. Dermatol. 118: 530-532.
- 3. Del Castillo, I., Moreno-Pelayo, M.A., Del Castillo, F.J., Brownstein, Z., Marlin, S., Adina, Q., Cockburn, D.J., Pandya, A., Siemering, K.R., Chamberlin, G.P., Ballana, E., Wuyts, W., Maciel-Guerra, A.T., Alvarez, A., Villamar, M., Shohat, M., Abeliovich, D., Dahl, H.H., Estivill, X., Gasparini, P., Hutchin, T., et al. 2003. Prevalence and evolutionary origins of the del(GJB6-D13S1830) mutation in the DFNB1 locus in hearing-impaired subjects: a multicenter study. Am. J. Hum. Genet. 73: 1452-1458.
- Essenfelder, G.M., Larderet, G., Waksman, G. and Lamartine, J. 2005.
  Gene structure and promoter analysis of the human GJB6 gene encoding connexin 30. Gene 350: 33-40.
- 5. Wilch, E., Zhu, M., Burkhart, K.B., Regier, M., Elfenbein, J.L., Fisher, R.A. and Friderici, K.H. 2006. Expression of GJB2 and GJB6 is reduced in a novel DFNB1 allele. Am. J. Hum. Genet. 79: 174-179.
- Yum, S.W., Zhang, J., Valiunas, V., Kanaporis, G., Brink, P.R., White, T.W. and Scherer, S.S. 2007. Human connexin 26 and connexin 30 form functional heteromeric and heterotypic channels. Am. J. Physiol., Cell Physiol. 293: C1032-C1048.
- 7. Gurtler, N., Egenter, C., Bosch, N. and Plasilova, M. 2008. Mutation analysis of the Cx26, Cx30, and Cx31 genes in autosomal recessive nonsyndromic hearing impairment. Acta Otolaryngol. 128: 1056-1062.
- 8. Evirgen, N., Solak, M., Dereköy, S., Erdogan, M., Yildiz, H., Eser, B., Arikan, S. and Erkoç, A. 2008. Genotyping for Cx26 and Cx30 mutations in cases with congenital hearing loss. Genet. Test. 12: 253-256.

#### **CHROMOSOMAL LOCATION**

Genetic locus: GJB6 (human) mapping to 13q12.11.

# **PRODUCT**

connexin 30 (h): 293T Lysate represents a lysate of human connexin 30 transfected 293T cells and is provided as 100  $\mu$ g protein in 200  $\mu$ l SDS-PAGE buffer.

# **APPLICATIONS**

connexin 30 (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive connexin 30 antibodies. Recommended use: 10-20  $\mu$ l per lane.

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

# **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.

# **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.

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