

GTF2IRD2/2B (I-14): sc-162916

BACKGROUND

The TFII-I family contains two highly homologous 949 amino acid proteins, GTF2IRD2 (GTF2I repeat domain containing 2) and GTF2IRD2B (general transcription factor II-I repeat domain-containing protein 2B). Localizing to the nucleus, these proteins are ubiquitously expressed and contain two GTF2I-like repeats. Encoded by a gene mapping to human chromosome 7q11.23, GTF2IRD2 and GTF2IRD2B are located in the Williams-Beuren syndrome (WBS) critical region. The deletion of genes located within this region results in WBS, possibly due to the unequal crossing over of highly homologous low-copy repeat sequences that flank the deleted region. WBS is an autosomal dominant genetic condition that is characterized by physical, cognitive and behavioral traits including facial dysmorphism, vascular stenoses, growth deficiencies, dental anomalies and neurologic and musculoskeletal abnormalities.

REFERENCES

1. Tipney, H.J., et al. 2004. Isolation and characterisation of GTF2IRD2, a novel fusion gene and member of the TFII-I family of transcription factors, deleted in Williams-Beuren syndrome. *Eur. J. Hum. Genet.* 12: 551-560.
2. Makeyev, A.V., et al. 2004. GTF2IRD2 is located in the Williams-Beuren syndrome critical region 7q11.23 and encodes a protein with two TFII-I-like helix-loop-helix repeats. *Proc. Natl. Acad. Sci. USA* 101: 11052-11057.
3. Hinsley, T.A., et al. 2004. Comparison of TFII-I gene family members deleted in Williams-Beuren syndrome. *Protein Sci.* 13: 2588-2599.
4. Del Campo, M., et al. 2006. Hemizygoty at the NCF1 gene in patients with Williams-Beuren syndrome decreases their risk of hypertension. *Am. J. Hum. Genet.* 78: 533-542.
5. Eckert, M.A., et al. 2006. The neurobiology of Williams syndrome: cascading influences of visual system impairment? *Cell. Mol. Life Sci.* 63: 1867-1875.
6. Osborne, L.R., et al. 2006. Williams-Beuren syndrome diagnosis using fluorescence *in situ* hybridization. *Methods Mol. Med.* 126: 113-128.
7. Ohazama, A., et al. 2007. TFII-I gene family during tooth development: candidate genes for tooth anomalies in Williams syndrome. *Dev. Dyn.* 236: 2884-2888.
8. Edelman, L., et al. 2007. An atypical deletion of the Williams-Beuren syndrome interval implicates genes associated with defective visuospatial processing and autism. *J. Med. Genet.* 44: 136-143.

CHROMOSOMAL LOCATION

Genetic locus: GTF2IRD2/GTF2IRD2B (human) mapping to 7q11.23; Gtf2ird2 (mouse) mapping to 5 G2.

SOURCE

GTF2IRD2/2B (I-14) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the C-terminus of GTF2IRD2 of human origin.

STORAGE

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

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-162916 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-162916 X, 200 µg/0.1 ml.

APPLICATIONS

GTF2IRD2/2B (I-14) is recommended for detection of human GTF2IRD2 isoforms 1 and 6; human GTF2IRD2B isoform 1; and mouse GTF2IRD2 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); non cross-reactive with human GTF2IRD2 isoforms 2-5 or human GTF2IRD2B isoform 2.

Suitable for use as control antibody for GTF2IRD2 siRNA (m): sc-145822, GTF2IRD2 shRNA Plasmid (m): sc-145822-SH and GTF2IRD2 shRNA (m) Lentiviral Particles: sc-145822-V.

GTF2IRD2/2B (I-14) X TransCruz antibody is recommended for Gel Supershift and ChIP applications.

Molecular Weight of GTF2IRD2/2B: 107 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.

RESEARCH USE

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

PROTOCOLS

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