

WBSCR11 (h): 293 Lysate: sc-113214

BACKGROUND

Williams-Beuren syndrome (WBS) is a developmental disorder caused by the hemizygous microdeletion on chromosome 7q11.23. WBS is an autosomal dominant genetic condition that is characterized by physical, cognitive and behavioral traits. The physical traits associated with WBS include facial dysmorphism, vascular stenoses, growth deficiencies, dental anomalies and neurologic and musculoskeletal abnormalities. Mild retardation, a weakness in visual-spatial skills, anxiety and a short attention span are typical cognitive and behavioral traits of WBS patients. The WBSCR11 gene is located within the WBS deletion and may contribute to the developmental symptoms found in WBS because of a loss of the encoded transcription factor. WBSCR11 is also designated GRF2IRD1, GTF3, Cream1, and MustRD1 in human and BEN in mouse, due to slight differences in gene structure. WBSCR11 is expressed in all adult tissues as several variants and has discrete spatial and temporal expression during embryogenesis. The amino terminus of WBSCR11 interacts with transcriptional machinery proteins, while the carboxy terminus has been shown to bind the retinoblastoma protein to possibly regulate the cell cycle.

REFERENCES

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CHROMOSOMAL LOCATION

Genetic locus: GTF2IRD1 (human) mapping to 7q11.23.

PRODUCT

WBSCR11 (h): 293 Lysate represents a lysate of human WBSCR11 transfected 293 cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

WBSCR11 (h): 293 Lysate is suitable as a Western Blotting positive control for human reactive WBSCR11 antibodies. Recommended use: 10-20 µl per lane.

Control 293 Lysate: sc-110760 is available as a Western Blotting negative control lysate derived from non-transfected 293 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.