

## TESPA1 (C-20): sc-247266

### BACKGROUND

KIAA0748, also known as HSPC257, is a 521 amino acid protein that exists as 3 alternatively spliced isoforms and is encoded by a gene that maps to human chromosome 12q13.2. Encoding over 1,100 genes within 132 million bases, chromosome 12 makes up about 4.5% of the human genome. A number of skeletal deformities are linked to chromosome 12, including hypochondrogenesis, achondrogenesis and Kniest dysplasia. Noonan syndrome, which includes heart and facial developmental defects among its primary symptoms, is caused by a mutant form of the PTPN11 gene product, SH-PTP2. Chromosome 12 is also home to a homeobox gene cluster, which encodes crucial transcription factors for morphogenesis, and the natural killer complex gene cluster, encoding C-type lectin proteins which mediate the NK cell response to MHC I interaction. Trisomy 12p leads to facial development defects, seizure disorders and a host of other symptoms which vary in severity depending on the extent of mosaicism. It is most severe in cases of complete trisomy.

### REFERENCES

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- Zumkeller, W., et al. 2004. Genotype/phenotype analysis in a patient with pure and complete trisomy 12p. *Am. J. Med. Genet. A* 129: 261-264.
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- Segel, R., et al. 2006. The natural history of trisomy 12p. *Am. J. Med. Genet. A* 140: 695-703.
- Stein, R. 2007. Genetics of Noonan syndrome-a new gene, and the search is still on. *Clin. Genet.* 72: 402-404.

### CHROMOSOMAL LOCATION

Genetic locus: TESPA1 (human) mapping to 12q13.2.

### SOURCE

TESPA1 (C-20) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the C-terminus of TESPA1 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-247266 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

### APPLICATIONS

TESPA1 (C-20) is recommended for detection of TESPA1 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 solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Molecular Weight of TESPA1 isoforms: 59/44/45 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.

### PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) or our catalog for detailed protocols and support products.