# Recombinant Human TBX1 Protein, MYC/DDK-tagged

**Cat. No.** TBX1-415H **Lot. No.** (See product label)

## SPECIFICATION

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| **Product Overview** | Recombinant human TBX1 protein, fused to MYC/DDK tag at C-terminus, was expressed in HEK293. |
| **Species** | Human |
| **Source** | HEK293 |
| **Description** | This gene is a member of a phylogenetically conserved family of genes that share a common DNA-binding domain, the T-box. T-box genes encode transcription factors involved in the regulation of developmental processes. This gene product shares 98% amino acid sequence identity with the mouse ortholog. DiGeorge syndrome (DGS)/velocardiofacial syndrome (VCFS), a common congenital disorder characterized by neural-crest-related developmental defects, has been associated with deletions of chromosome 22q11.2, where this gene has been mapped. Studies using mouse models of DiGeorge syndrome suggest a major role for this gene in the molecular etiology of DGS/VCFS. Several alternatively spliced transcript variants encoding different isoforms have been described for this gene. [provided by RefSeq, Jul 2008]. |
| **Form** | 25 mM Tris.HCl, pH 7.3, 100 mM glycine, 10 % glycerol. |
| **Molecular Mass** | 52.5 kDa |
| **Purity** | > 80% as determined by SDS-PAGE and Coomassie blue staining |

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| **Concentration** | >50 ug/mL as determined by microplate BCA method |

## GENE INFORMATION

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| **Gene Name** | T-box 1 [ Homo sapiens ] |
| **Official Symbol** | TBX1 |
| **Synonyms** | CAFS; CATCH22; CTHM; DGCR; DGS; DORV; TBX1C; TGA; VCF; VCFS |
| **Gene ID** | 6899 |
| **mRNA Refseq** | NM_005992.1 |
| **Protein Refseq** | NP_005983.1 |
| **MIM** | 602054 |
| **UniProt ID** | O43435 |

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