



Human TBX1 blocking peptide (DAG-P1226)

This product is for research use only and is not intended for diagnostic use.

PRODUCT INFORMATION

Antigen 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]
Conjugate	Unconjugated
Applications	BL
Sequence Similarities	Contains 1 T-box DNA-binding domain.
Format	Liquid
Preservative	None
Storage	Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles. Information available upon request.

GENE INFORMATION

Gene Name	TBX1 T-box 1 [Homo sapiens (human)]
Official Symbol	TBX1
Synonyms	TBX1; T-box 1; DGS; TGA; VCF; CAFS; CTHM; DGCR; DORV; VCFS; TBX1C; CATCH22; T-

box transcription factor TBX1; brachyury; Testis-specific T-box protein; T-box 1 transcription factor C;

Entrez Gene ID	6899
mRNA Refseq	NM_005992.1
Protein Refseq	NP_005983.1
UniProt ID	O43435
Chromosome Location	22q11.21
Pathway	Heart Development, organism-specific biosystem;
Function	DNA binding; protein dimerization activity; protein homodimerization activity; sequence-specific DNA binding; NOT sequence-specific DNA binding transcription factor activity;