





# **TBX1 Monoclonal Antibody**

Catalog No	YP-mAb-02076
Isotype	IgG
Reactivity	Human;Rat;Mouse;
Applications	WB
Gene Name	TBX1
Protein Name	T-box transcription factor TBX1
lmmunogen	The antiserum was produced against synthesized peptide derived from human TBX1. AA range:311-360
Specificity	TBX1 Monoclonal Antibody detects endogenous levels of TBX1 protein.
Formulation	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.
Source	Monoclonal, Mouse,IgG
Purification	The antibody was affinity-purified from mouse antiserum by affinity-chromatography using epitope-specific immunogen.
Dilution	WB 1:500-1:2000
Concentration	1 mg/ml
Purity	≥90%
Storage Stability	-20°C/1 year
Synonyms	TBX1; T-box transcription factor TBX1; T-box protein 1; Testis-specific T-box protein
Observed Band	43kD
Cell Pathway	Nucleus .
Tissue Specificity	Skeletal muscle,Testis,
Function	disease:Defects in TBX1 are a cause of conotruncal heart malformations (CTHM) [MIM:217095]. CTHM consist of cardiac outflow tract defects, such as tetralogy of Fallot, pulmonary atresia, double-outlet right ventricle, truncus arteriosus communis, and aortic arch anomalies.,disease:Defects in TBX1 are a cause of DiGeorge syndrome (DGS) [MIM:188400].,disease:Defects in TBX1 are a cause of velocardiofacial syndrome (VCFS) [MIM:192430].,disease:Haploinsufficiency of the TBX1 gene is responsible for most of the physical malformations present in DiGeorge syndrome (DGS) and velocardiofacial syndrome (VCFS) [MIM:188400, 192430]. DGS is characterized by the association of several malformations: hypoplastic thymus and parathyroid glands, congenital conotruncal cardiopathy, and a subtle but characteristic facial dysmorphology. VCFS is marked by the association of congenital conotruncal heart defect



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## **Background**

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 PofSeg. Jul 2008] described for this gene. [provided by RefSeq, Jul 2008],

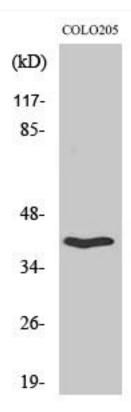
#### matters needing attention

Avoid repeated freezing and thawing!

### Usage suggestions

This product can be used in immunological reaction related experiments. For more information, please consult technical personnel.

# **Products Images**



Western Blot analysis of various cells using TBX1 Monoclonal Antibody