

Product name:	TBX1 Rabbit Polyclonal Antibody
Cat number:	ABN18698
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human TBX1. AA range:311-360
Reactivity:	Human,Rat,Mouse
Applications:	WB 1:500-1:2000,IHC 1:100-1:300,ICC/IF 1:200-1:1000,ELISA 1:10000-1:20000
Molecular Weight:	43kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

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 RefSeq, Jul 2008],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 dysmorphism. VCFS is marked by the association of congenital conotruncal heart defects, cleft palate or velar insufficiency, facial dysmorphology and learning difficulties. It is now accepted that these two syndromes represent two forms of clinical expression of the same entity manifesting at different stages of life.,function:Probable transcriptional regulator involved in developmental processes. Is required for normal development of the pharyngeal arch arteries.,similarity:Contains 1 T-box DNA-binding domain.,