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Rabbit Anti-TBX1 antibody

SL21502R

Product Name:	TBX1
Chinese Name:	先心病相关蛋白TBX1抗体
Alias:	CAFS; CTHM; DGCR; DGS; DORV; T box 1; T box 1 transcription factor; T box 1 transcription factor C; T box; T box protein 1; T box transcription factor TBX 1; T box transcription factor TBX1; T-box 1; T-box protein 1; T-box transcription factor TBX1; TBX 1; TBX 1C; tbx1; TBX1_HUMAN; TBX1C; Testis specific T box protein; Testis-specific T-box protein; TGA; VCFS.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Pig,Horse,Rabbit,Sheep,
Applications:	WB=1:500-2000ELISA=1:500-1000 not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	43kDa
Cellular localization:	The nucleus
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human TBX1:271-370/398
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed:	PubMed
Product Detail:	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].

Function:
Probable transcriptional regulator involved in developmental processes. Is required for normal development of the pharyngeal arch arteries. Subunit : Interacts with DSCR6.

Subunit:
Interacts with DSCR6.

Subcellular Location:
Nucleus.

DISEASE:
Note=Haploinsufficiency of the TBX1 gene is responsible for most of the physical malformations present in DiGeorge syndrome (DGS) and velocardiofacial syndrome (VCFS). 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 defects, cleft palate or velar insufficiency, facial dysmorpholgy 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.
DiGeorge syndrome (DGS) [MIM:188400]: A congenital syndrome characterized by a wide spectrum of characteristics including parathyroid hypoplasia resulting in hypocalcemia, thymic hypoplasia resulting in T-cell immunodeficiency, defects in the outflow tract of the heart, and craniofacial anomalies. Disturbance of cervical neural crest migration into the derivatives of the pharyngeal arches and pouches can account for the phenotype. Note=The disease is caused by mutations affecting the gene represented in this entry.
Velocardiofacial syndrome (VCFS) [MIM:192430]: A syndrome characterized by abnormal pharyngeal arch development that results in defective development of the parathyroid glands, thymus, and conotruncal region of the heart. The phenotype is highly variable, with no single clinical feature present in every patient. Affected individuals may present with structural or functional palatal abnormalities, cardiac defects, unique facial characteristics, hypernasal speech, hypotonia, and defective thymic development associated with impaired immune function. In addition, affected individuals may present with learning disabilities, overt developmental delay, and psychiatric disorders. Note=The disease is caused by mutations affecting the gene represented in this entry.
Conotruncal heart malformations (CTHM) [MIM:217095]: A group of congenital heart defects involving the outflow tracts. Examples include truncus arteriosus communis, double-outlet right ventricle and transposition of great arteries. Truncus arteriosus

communis is characterized by a single outflow tract instead of a separate aorta and pulmonary artery. In transposition of the great arteries, the aorta arises from the right ventricle and the pulmonary artery from the left ventricle. In double outlet of the right ventricle, both the pulmonary artery and aorta arise from the right ventricle. Note=The disease is caused by mutations affecting the gene represented in this entry.

Similarity:
Contains 1 T-box DNA-binding domain.

SWISS:
O43435

Gene ID:
6899

Database links:

Entrez Gene: 6899Human

Entrez Gene: 21380Mouse

Entrez Gene: 360737Rat

Omim: 602054Human

SwissProt: O43435Human

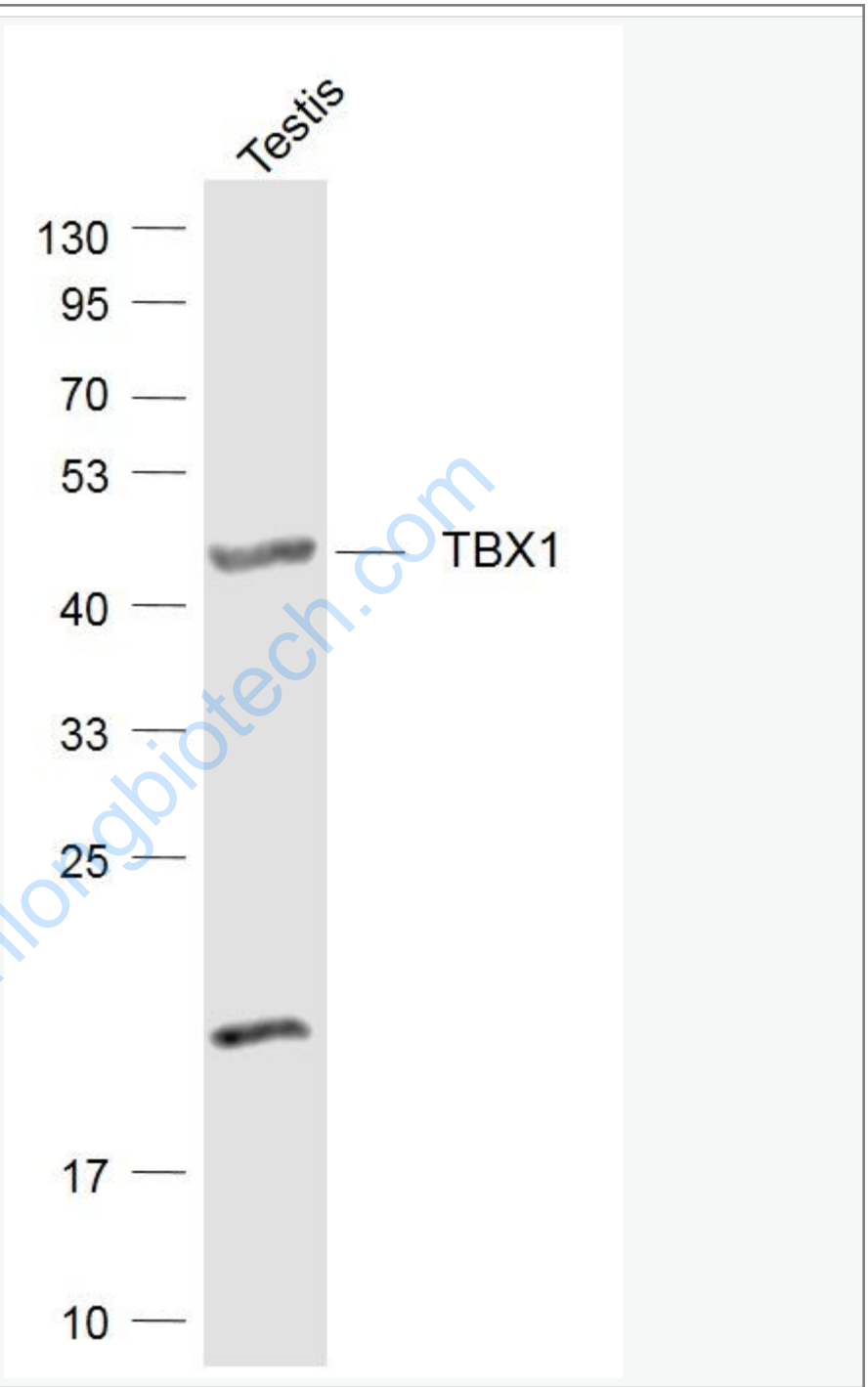
SwissProt: P70323Mouse

Unigene: 173984Human

Unigene: 295194Mouse

Unigene: 41347Rat

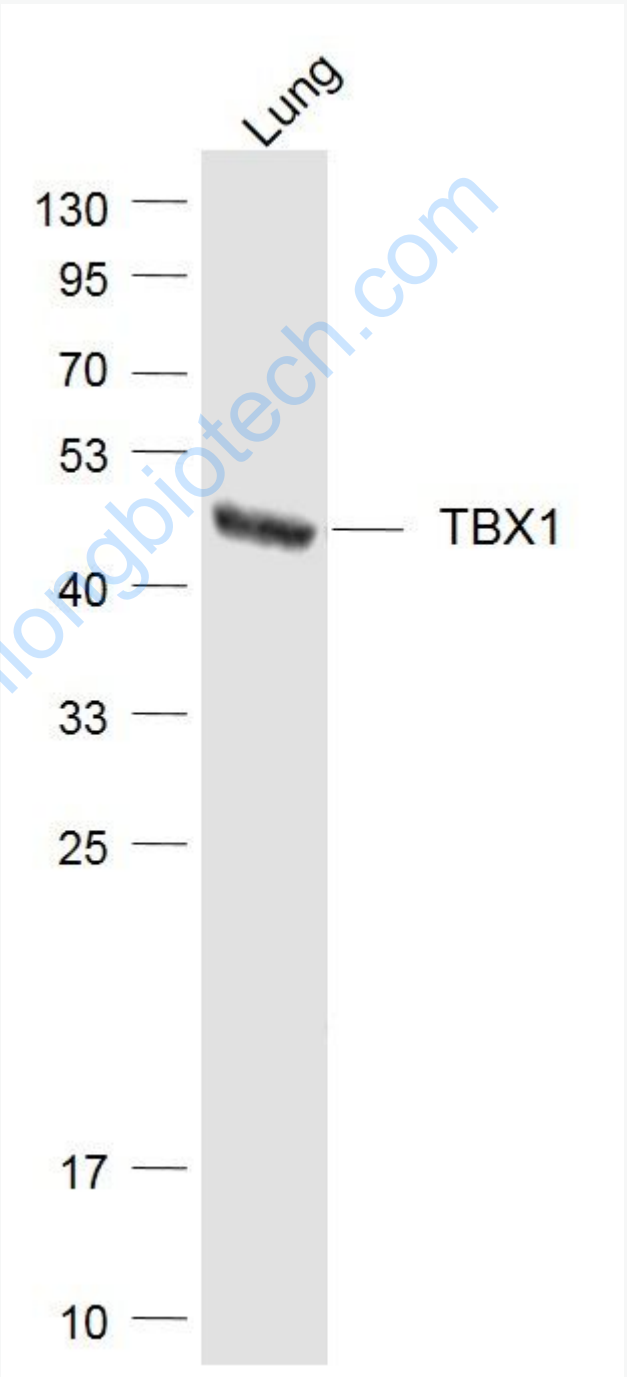
Important Note:
This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

Picture:	 Sample: Testis (Mouse) Lysate at 40 ug Primary: Anti-TBX1 (SL21502R) at 1/1000 dilution

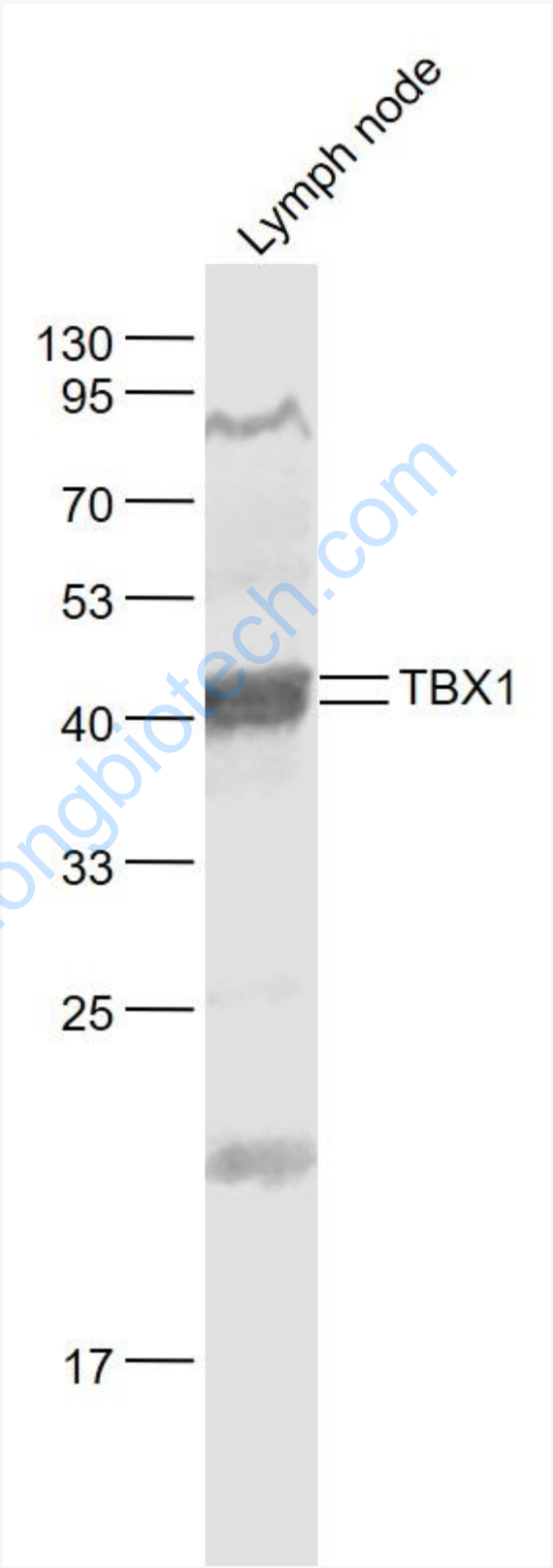
Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution

Predicted band size: 43 kD

Observed band size: 43 kD



	Sample: Lung (Mouse) Lysate at 40 ug Primary: Anti-TBX1 (SL21502R) at 1/1000 dilution Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution Predicted band size: 43 kD Observed band size: 43 kD



Sample:

	Lymph node (Mouse) Lysate at 40 ug Primary: Anti- TBX1 (SL21502R) at 1/1000 dilution Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution Predicted band size: 43 kD Observed band size: 41'43 kD