TBX1 (Human) Cell-Based ELISA Kit

Catalog # KA6278 Size 1 Kit

Specification

Product Description	TBX1 (Human) Cell-Based ELISA Kit is an indirect enzyme-linked immunoassay for qualitative determination of TBX1 expression in cultured cells.
Suitable Sample	Attached Cell, Loosely Attached Cell, Suspension Cell.
Label	HRP-conjugated
Detection Method	Colorimetric
Assay Type	Qualitative
Reactivity	Human
Regulatory Status	For research use only (RUO)
Storage Instruction	Store at 4°C for six months.

Applications

- Qualitative

Gene Info — TBX1

Entrez GeneID	6899
Protein Accession#	O43435
Gene Name	TBX1
Gene Alias	CAFS, CTHM, DGCR, DGS, DORV, TBX1C, TGA, VCFS
Gene Description	T-box 1

Omim ID	188400 192430 217095 602054
Gene Ontology	Hyperlink
Gene Summary	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
Other Designations	T-box 1 transcription factor C|Testis-specific T-box protein|brachyury

Disease

- Abnormalities
- Alagille Syndrome
- Chromosome Deletion
- Cleft Lip
- Cleft Palate
- Down Syndrome
- Ductus Arteriosus
- Genetic Predisposition to Disease
- Heart Defects
- Infant
- Mood Disorders
- Psychotic Disorders
- Schizophrenia
- Syndrome
- Tetralogy of Fallot