INPUT
Variant
Chr17:59477596 G>A

Gene
TBX2

DATABASES
OMIM
ExAC / Geno2MP
ClinVar
DGV / DECIPHER

MODEL ORGANISMS
Predicted Orthologs
Protein Domain
Protein Alignment

Human Gene Description (OMIM)
T-BOX 2; TBX2

MIM number: 600747

Description: The TBX2 gene encodes a transcription factor that belongs to the family of T-box factor proteins that bind DNA, including TBX1 (602054) and TBX3 (601621). TBX2 is expressed in a variety of tissues and organs during embryogenesis (summary by Harrelson et al., 2004).

Gene-Phenotype Relationships
OMIM

Phenotype	Phenotype MIM number	Inheritance
Vertebral anomalies and variable endocrine and T-cell dysfunction	618223	Autosomal dominant

Reported Alleles From OMIM
TBX2

Phenotype	Mutation	dbSNP
VERTEBRAL ANOMALIES AND VARIABLE ENDOCRINE AND T-CELL DYSFUNCTION	TBX2, ARG20GLN	rs1364709483
VERTEBRAL ANOMALIES AND VARIABLE ENDOCRINE AND T-CELL DYSFUNCTION	TBX2, ARG305HIS	rs1555877071

Control Population Gene Summary
TBX2 (ExAC Gene Table)

Constraint from ExAC	Expected no. variants	Observed no. variants	ConstraintMetric
Synonymous	170.6	96	z=3.54
Missense	289.1	172	z=3.37
LoF	14.5	1	pLI=0.96
CNV	4.2	0	z=0.92

Disease Population (Geno2MP Database)
Chr17:59477596 G>A

No matches found

Gene-Phenotype Relationships (Geno2MP)
Chr17:59477596 G>A

No matches found

Population Allele Frequencies (ExAC Database)
Chr17:59477596 G>A

No matches found

Population Allele Frequencies (GnomAD Database)
Chr17:59477596 G>A

Allele Count	2
Allele Number	179042
Homozygous count	0
Allele frequency	0.000011171
Gene	RP11-332H18.4

Benign (ClinVar)	Likely Benign (ClinVar)	Pathogenic (ClinVar)	Likely Pathogenic (ClinVar)	Risk Factor (ClinVar)	Uncertain Significance (ClinVar)	Conflicting Interpretations (ClinVar)
0	0	18	0	0	2	0

Reported Alleles From ClinVar
TBX2 / Chr17:59477596 G>A

Variation	Location	Condition(s)	Frequency	Clinical Significance	Review Status
NC_000017.10:g.17711738_217748468del200036731	**Chr17:17711738-217748468**	Smith-Magenis syndrome		Pathogenic	criteria provided, single submitter
NC_000017.10:g.(?_56321134)_(62080001_?)dup	**Chr17:56321134-62080001**	See cases		Pathogenic	criteria provided, single submitter
GRCh38/hg38 17q23.2(chr17:61331901-61424019)x3	**Chr17:59409262-59501380**	See cases		Uncertain significance	no assertion criteria provided
GRCh38/hg38 17q23.1-25.1(chr17:36449220-75053130)x3	**Chr17:57595736-73049225**	See cases		Pathogenic	no assertion criteria provided
GRCh37/hg19 17q23-24(chr17:59209629-64222315)x3	**Chr17:59209629-64222315**	See cases		Pathogenic	criteria provided, single submitter
GRCh38/hg38 17q23.2-25.3(chr17:36449220-83086677)x3	**Chr17:58617905-81044553**	See cases		Pathogenic	criteria provided, single submitter
GRCh38/hg38 17q23.1-23.2(chr17:60043448-62148729)x1	**Chr17:58120809-60226090**	See cases		Pathogenic	criteria provided, single submitter
GRCh37/hg19 17q23(chr17:58934659-60395826)x1	**Chr17:58934659-60395826**	See cases		Pathogenic	criteria provided, single submitter

Copy Number Variation In Control Population (DGV Database)
TBX2

Show 10 entries Search:

Position	Size	Type	Subtype	Frequency	Gain	Loss	Sample Size	References
17 59323466 59562984	239518	CNV	loss	0.02105263	0	2	95	17160897
17 59454601 59495300	40699	CNV	deletion	1.00000000	0	1	1	24416366
17 59456589 59491281	34692	CNV	gain	0.00049358	1	0	2026	19592680
17 59476600 59478200	1600	CNV	deletion	0.50000000	0	1	2	24896259
17 59486746 59487226	480	CNV	gain	0.09677419	3	0	31	20364138

Showing 1 to 5 of 5 entries Previous 1 Next

Common Copy Number Variants (DECIPHER Database)
17:59477596

No matches found

Gene Function Table
TBX2

Show only best DIOPT v6 score gene

	Homolog	DIOPT Score	Expression	Molecular function	Cellular component	Biological process
Human	TBX2 PubMed Monarch	NA	• adrenal gland • breast • caudate • cerebellum • cerebral cortex • cervix, uterine • colon • epididymis • fallopian tube • kidney • nasopharynx • placenta • rectum • skeletal muscle • testis • thyroid gland • urinary bladder Show all / GTEx	• contributes_toRNA polymerase II core promoter proximal region sequence-specific DNA binding • transcriptional repressor activity, RNA polymerase II core promoter proximal region sequence-specific binding • protein binding • contributes_tosequence-specific DNA binding	• nucleus	• negative regulation of transcription from RNA polymerase II promoter • cell aging • positive regulation of cell proliferation • negative regulation of transcription, DNA-templated • cellular senescence
Rat	Tbx2 PubMed	7/11	Show all (9)	No term based on experiment	No term based on experiment	No term based on experiment
Mouse	Tbx2 PubMed IMPC	13/13	• extraembryonic component • embryo ectoderm • cardiovascular system • branchial arches • alimentary system Show 12 more Open on MGI	• DNA binding • transcription factor activity, sequence-specific DNA binding • protein binding	• nucleus • transcription factor complex	• heart morphogenesis • outflow tract septum morphogenesis • outflow tract morphogenesis • endocardial cushion morphogenesis • regulation of transcription from RNA polymerase II promoter involved in myocardial precursor cell differentiation Show 15 more
Zebrafish	tbx2b PubMed	11/12	• brain • cranial ganglion • epiphysis • retinal ganglion cell layer • retinal neural layer Show 51 more Open on ZFIN	No term based on experiment	No term based on experiment	• heart looping • cardiac chamber development • cell adhesion • brain development • regulation of heart contraction Show 15 more
	tbx2a PubMed	9/12	• cardiac ventricle • hindbrain • hypothalamus • olfactory placode • optic vesicle Show 22 more Open on ZFIN	• protein binding	No term based on experiment	• cardiac chamber development • embryonic heart tube development • pharyngeal system development
Drosophila	bi PubMed	10/12	• Eye • Brain Show all (25)	• DNA binding • protein binding	• nucleus	• negative regulation of transcription from RNA polymerase II promoter • compound eye morphogenesis • wing disc morphogenesis • imaginal disc-derived wing morphogenesis • organ growth Show 1 more
C Elegans	tbx-2 PubMed	8/12	Open on WormBase	• enzyme binding	• nematode larval development • nucleus • cytoplasm	• regulation of transcription from RNA polymerase II promoter • regulation of protein localization • locomotion

Human Gene Protein Domains (DIOPT V6)
TBX2

Index	Domain name	Domain start	Domain stop	Domain description	Protein ID	External ID
hs1	TBOX	106	289	T-box DNA binding domain of the T-box family of transcriptional regulators. The T-box family is an ancient group that appears to play a critical role in development in all animal species. These genes were uncovered on the basis of similarity to the DNA...; cd00182	NP_005985.3	CDD:29144
hs1	Repression domain 1 (RD1)	518	601	propagated from UniProtKB/Swiss-Prot (Q13207.3)	NP_005985.3	
hs1	TBX	305	382	T-box transcription factor; pfam12598	NP_005985.3	CDD:204975

Multiple Protein Alignment (DIOPT V6)
TBX2

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hs1 ------------GLAGAAAAAAAAAAAAEAGLHVSALG----PHPPAAH----------------------------LRSLKSLEPEDE-VEDDPKVTLEAKELWDQFHKLGTEMVITK[129]
mm1 ------------GLAGAAAAAAAAAAAAEAGLHVSALG----PHPPAAH----------------------------LRSLKSLEPEDE-VEDDPKVTLEAKELWDQFHKLGTEMVITK[129]
rn1 ------------------------------------------------------------------------------------------------------------MVITK[5]
dr1 ------------TLAG----------AAEAGLH-PALS----HHHQAAH----------------------------LRSLKSLEPEEE-VEDDPKVTLEAKDLWDQFHKLGTEMVITK[118]
dr2 ------------ALSG----------AAEAGLH-AALG----HHHQAAH----------------------------LRSFKGLEPEED-VEDDPKVTLEAKELWDQFHKIGTEMVITK[117]
dm1 PPPPYFPAAALAALAG--------SPAGPHPGLY-PGGGLRFPPHHPGAHPHAHHLGSAYTTAEDVVLASAVAHQLHPAMRPLRALQPEDDGVVDDPKVTLEGKDLWEKFHKLGTEMVITK[352]
ce1 ------------------------------------------------------------------------------------------PEDDGVTDDPKVELDERELWQQFSQCGTEMVITK[85]
                                                                                                                        *****
hs1 SGRRMFPPFKVRVSGLDKKAKYILLMDIVAADDCRYKFHNSRWMVAGKADPEMPKRMYIHPDSPATGEQWMAKPVAFHKLKLTNNISDKHGF---TILNSMHKYQPRFHIVRANDILKLP[246]
mm1 SGRRMFPPFKVRVSGLDKKAKYILLMDIVAADDCRYKFHNSRWMVAGKADPEMPKRMYIHPDSPATGEQWMAKPVAFHKLKLTNNISDKHGF---TILNSMHKYQPRFHIVRANDILKLP[246]
rn1 SGRRMFPPFKVRVSGLDKKAKYILLMDIVAADDCRYKFHNSRWMVAGKADPEMPKRMYIHPDSPATGEQWMAKPVAFHKLKLTNNISDKHGF---TILNSMHKYQPRFHIVRANDILKLP[122]
dr1 SGRRMFPPFKVRINGLDKKAKYILLMDIVAADDCRYKFHNSRWMVAGKADPEMPKRMYIHPDSPATGEQWMAKPVAFHKLKLTNNISDKHGF---TILNSMHKYQPRFHIVRANDILKLP[235]
dr2 SGRRMFPPFKVRVNGLDKKAKYILLMDIVAADDCRYKFHNSRWMVAGKADPEMPKRMYIHPDSPATGEQWMAKPVAFHKLKLTNNISDKHGF---TILNSMHKYQPRFHIVRANDILKLP[234]
dm1 SGRQMFPQMKFRVSGLDAKAKYILLLDIVAADDYRYKFHNSRWMVAGKADPEMPKRMYIHPDSPTTGEQWMQKVVSFHKLKLTNNISDKHGFVSTTILNSMHKYQPRFHLVRANDILKLP[472]
ce1 SGRRIFRAYRVKISGLDKKSQYFVMMDLVRADFHRYKFNNSRWMIAGKADPEMPKTLYIHPDSRSTGEHWMSKGANFHKLKLTNNISDKHGY---TILNSMHKYQPRLHVVRGADRHNLM[202]
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Highlight from Integer (1~n) **to** Integer (1~n) **for** ☑ hs ☐ rn ☐ mm ☐ dr ☐ dm ☐ ce