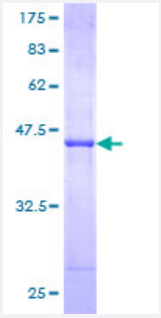
Full-Length

GTF2H2 (Human) Recombinant Protein (P01)

Catalog # H00002966-P01 Size 25 ug, 10 ug

Applications



Specification

Product Description	Human GTF2H2 full-length ORF (AAH05345, 1 a.a. - 165 a.a.) recombinant protein with GST-tag at N-terminal.
Sequence	MDEEPERTKRWEGGYERTWEILKEDESGSLKATIEDILFKAKRKRVFEHHGQVRLGMMRHLYVVV DGSRTMEDQDLKPNRLTCTLKLLEYFVEEYFDQNPISQIGIIVTKSKRAEKLTELSGNPRKHITSLKK AVDMTCHGEPSLYNSLSIAMQTLKLVLYIMYN
Host	Wheat Germ (in vitro)
Theoretical MW (kDa)	43.89
Preparation Method	*in vitro* wheat germ expression system
Purification	Glutathione Sepharose 4 Fast Flow
Quality Control Testing	12.5% SDS-PAGE Stained with Coomassie Blue.
Storage Buffer	50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.
Storage Instruction	Store at -80°C. Aliquot to avoid repeated freezing and thawing.
Note	Best use within three months from the date of receipt of this protein.

Applications

- Enzyme-linked Immunoabsorbent Assay
- Western Blot (Recombinant protein)
- Antibody Production
- Protein Array

Gene Info — GTF2H2

Entrez GeneID	2966
GeneBank Accession#	BC005345
Protein Accession#	AAH05345
Gene Name	GTF2H2
Gene Alias	BTF2, BTF2P44, MGC102806, T-BTF2P44, TFIIH
Gene Description	general transcription factor IIH, polypeptide 2, 44kDa
Omim ID	601748
Gene Ontology	Hyperlink
Gene Summary	This gene is part of a 500 kb inverted duplication on chromosome 5q13. This duplicated region contains at least four genes and repetitive elements which make it prone to rearrangements and deletions. The repetitiveness and complexity of the sequence have also caused difficulty in determining the organization of this genomic region. This gene is within the telomeric copy of the duplication. Deletion of this gene sometimes accompanies deletion of the neighboring SMN1 gene in spinal muscular atrophy (SMA) patients but it is unclear if deletion of this gene contributes to the SMA phenotype. This gene encodes the 44 kDa subunit of RNA polymerase II transcription initiation factor IIH which is involved in basal transcription and nucleotide excision repair. Transcript variants for this gene have been described, but their full length nature has not been determined. A second copy of this gene within the centromeric copy of the duplication has been described in the literature. It is reported to be different by either two or four base pairs; however, no sequence data is currently available for the centromeric copy of the gene. [provided by RefSeq
Other Designations	general transcription factor IIH, polypeptide 2 (44kD subunit)|general transcription factor IIH, polypeptide 2, 44kD subunit

Pathway

- Basal transcription factors
- Nucleotide excision repair

Disease

- Spinal Muscular Atrophies of Childhood
- Spinal muscular atrophy