

# Recombinant Full Length Human GTF2H2 Protein, GST-tagged

**Cat. No.** GTF2H2-3387HF    **Lot. No.** (See product label)

## SPECIFICATION

**Product Overview**      Human GTF2H2 full-length ORF ( AAH05345, 1 a.a. - 165 a.a.) recombinant protein with GST-tag at N-terminal.

**Species**                      Human

**Source**                        In Vitro Cell Free System

**ProteinLength**              165 amino acids

**Description**

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

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<b>Molecular Mass</b>	43.89 kDa
<b>AA Sequence</b>	MDEEPTKRWEGGYERTWEILKEDESGSLKATIEDILFKAKRKRVFEEHHGQVRLG MMRHLYVVVDGSRMEDQDLKPNRLTCTLKLLEYFVEEYFDQNPISQIGIIVTKSKRA EKLTELSGNPRKHITSKKAVDMTCHGEPSTLYNSLSIAMQTLKLVLYIMYN
<b>Applications</b>	Enzyme-linked Immunoabsorbent Assay Western Blot (Recombinant protein) Antibody Production Protein Array
<b>Notes</b>	Best use within three months from the date of receipt of this protein.
<b>Storage</b>	Store at -8 centigrade. Aliquot to avoid repeated freezing and thawing.
<b>Storage Buffer</b>	50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.

## GENE INFORMATION

<b>Gene Name</b>	GTF2H2 general transcription factor IIH, polypeptide 2, 44kDa [ Homo sapiens ]
<b>Official Symbol</b>	GTF2H2
<b>Synonyms</b>	GTF2H2; general transcription factor IIH, polypeptide 2, 44kDa; general transcription factor IIH, polypeptide 2 (44kD subunit); general transcription factor IIH subunit 2; BTF2; BTF2P44; p44; T BTF2P44; TFIIH; BTF2 p44; basic transcription factor 2 44 kDa subunit; general transcription factor IIH polypeptide 2; TFIIH basal transcription factor complex p44 subunit; general transcription factor IIH, polypeptide 2, 44kD subunit; T-BTF2P44; MGC102806;
<b>Gene ID</b>	2966

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mRNA Refseq	NM_001515
Protein Refseq	NP_001506
MIM	601748
UniProt ID	Q13888

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