Recombinant Human HTT Protein, GST-tagged

Cat. No. HTT-4637H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Human HD partial ORF (NP_002102, 81 a.a. - 190 a.a.) recombinant protein with GST-tag at N-terminal.
Species	Human
Source	Wheat Germ
Description	Huntingtin is a disease gene linked to Huntington's disease, a neurodegenerative disorder characterized by loss of striatal neurons. This is thought to be caused by an expanded, unstable trinucleotide repeat in the huntingtin gene, which translates as a polyglutamine repeat in the protein product. A fairly broad range of trinucleotide repeats (9-35) has been identified in normal controls, and repeat numbers in excess of 40 have been described as pathological. The huntingtin locus is large, spanning 180 kb and consisting of 67 exons. The huntingtin gene is widely expressed and is required for normal development. It is expressed as 2 alternatively polyadenylated forms displaying different relative abundance in various fetal and adult tissues. The larger transcript is approximately 13.7 kb and is expressed predominantly in adult and fetal brain whereas the smaller transcript of approximately 10.3 kb is more widely expressed. The genetic defect leading to Huntington's disease may not necessarily eliminate transcription, but may confer a new property on the mRNA or alter the function of the protein. One candidate is the huntingtin-associated protein-1, highly expressed in brain, which has increased affinity for huntingtin protein with expanded polyglutamine repeats. This gene contains an upstream open reading frame in the 5' UTR that inhibits expression of the huntingtin gene product through translational

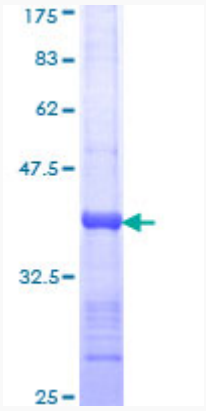
Tel: 1-631-559-9269 1-516-512-3133
Email: info@creative-biomart.com Fax: 1-631-938-8127
45-1 Ramsey Road, Shirley, NY 11967, USA

	repression. [provided by RefSeq, Jul 2016]
Molecular Mass	37.84 kDa
AA Sequence	AVAEEPLHRPKKELSATKKDRVNHCLTICENIVAQSVRNSPEFQKLLGIAMELFLLCS DDAESDVRMVADECLNKVIKALMDSNLPRLQLELYKEIKKNGAPRSLRAALW
Applications	Enzyme-linked Immunoabsorbent Assay Western Blot (Recombinant protein) Antibody Production Protein Array
Notes	Best use within three months from the date of receipt of this protein.
Storage	Store at -8 centigrade. Aliquot to avoid repeated freezing and thawing.
Storage Buffer	50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.

GENE INFORMATION

Gene Name	HTT huntingtin [Homo sapiens]
Official Symbol	HTT
Synonyms	HTT; huntingtin; HD, huntingtin (Huntington disease); IT15; huntington disease protein; HD;
Gene ID	3064
mRNA Refseq	NM_002111

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Protein Refseq	NP_002102
MIM	613004
UniProt ID	P42858
Quality Control Testing	 12.5% SDS-PAGE Stained with Coomassie Blue.

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