

## Recombinant Human BEST1, GST-tagged

Cat. No. BEST1-10208H Lot. No. (See product label)

### SPECIFICATION

**Product Overview** Recombinant Human BEST1 protein, fused to GST-tag, was expressed in E.coli and purified by GSH-sepharose.

**Species** Human

**Source** E.coli

**ProteinLength** C-term-180a.a.

**Description** This gene encodes a member of the bestrophin gene family. This small gene family is characterized by proteins with a highly conserved N-terminus with four to six transmembrane domains. Bestrophins may form chloride ion channels or may regulate voltage-gated L-type calcium-ion channels. Bestrophins are generally believed to form calcium-activated chloride-ion channels in epithelial cells but they have also been shown to be highly permeable to bicarbonate ion transport in retinal tissue. Mutations in this gene are responsible for juvenile-onset vitelliform macular dystrophy (VMD2), also known as Best macular dystrophy, in addition to adult-onset vitelliform macular dystrophy (AVMD) and other retinopathies. Alternative splicing results in multiple variants encoding distinct isoforms.

**Storage** The protein is stored in PBS buffer at -20°C. Avoid repeated freezing and thawing cycles.

**Storage Buffer** 1M PBS (58mM Na<sub>2</sub>HPO<sub>4</sub>, 17mM NaH<sub>2</sub>PO<sub>4</sub>, 68mM NaCl, pH8. ) added with 100mM GSH and 1% Triton X-100, 15% glycerol.

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## GENE INFORMATION

<b>Gene Name</b>	BEST1 bestrophin 1 [ Homo sapiens ]
<b>Official Symbol</b>	BEST1
<b>Synonyms</b>	BEST1; bestrophin 1; vitelliform macular dystrophy 2 , VMD2; bestrophin-1; BEST; Best disease; BMD; vitelliform macular dystrophy protein 2; ARB; RP50; VMD2; TU15B;
<b>Gene ID</b>	7439
<b>mRNA Refseq</b>	NM_001139443
<b>Protein Refseq</b>	NP_001132915
<b>MIM</b>	607854
<b>UniProt ID</b>	O76090
<b>Chromosome Location</b>	11q12
<b>Function</b>	chloride channel activity; contributes_to chloride channel activity; ion channel activity;

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