

Recombinant Full Length Human BEST1 Protein, GST-tagged

Cat. No. BEST1-1573HF **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Human BEST1 full-length ORF (AAH41664.1, 1 a.a. - 604 a.a.) recombinant protein with GST-tag at N-terminal.
Species	Human
Source	In Vitro Cell Free System
ProteinLength	604 amino acids
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
Form	50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.
Molecular Mass	95.5 kDa
AA Sequence	MFEKLTLYCD SYIQLIPISF VLGFYVTLVV TRWWNQYENL PWPDRMLSLV

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SGFVEGKDEQ GRLLRRTLIR YANLGNVLIL RSVSTAVYKR FPSAQHLVQA
GFMTPAEHKQ LEKLSLPHNM FWVPWVWFAN LSMKAWLGGR IRDPILLQSL
LNEMNTLRTQ CGHLYAYDWI SIPLVYTQVV TVAVYSFFLT CLVGRQFLNP
AKAYPGHELD LVVPVFTFLQ FFFYVGWLKV AEQLINPFGE DDDDFETNWI
VDRNLQVSL AVDEMHQDLP RMEPDMYWNK PEPQPPYTAA SAQFRRVSFM
GSTFNISLNK EEMEFQPNQE DEEDAHAGII GRFLGLQSHD HHPPRANSRT
KLLWPKRESL LHEGLPKNHK AAKQNVRGQE DNKAWKLKAV DAFKSAPLYQ
RPGYYAPQT PLSPTPMFFP LEPSAPSKLH SVTGIDTKDK SLKTVSSGAK
KSFELLSESD GALMEHPEVS QVRRKTVEFN LTDMPEIPEN HLKEPLEQSP
TNIHTTLKDH MDPYWALENR SVLHLNQGHC IALCPTPASL ALSLPFLHNF
LGFHHCQSTL DLRPALAWGI YLATFTGILG KCSGPFLTSP WYHPEDFLGP GEGR

Storage Store at -80 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 BEST1 bestrophin 1 [Homo sapiens]

Official Symbol BEST1

Synonyms BEST1; bestrophin 1; vitelliform macular dystrophy 2 , VMD2; bestrophin-1; BEST; Best disease; BMD; vitelliform macular dystrophy protein 2; ARB; RP50; VMD2; TU15B

Gene ID 7439

mRNA Refseq NM_001139443

Protein Refseq NP_001132915

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MIM	607854
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UniProt ID	O76090
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