Recombinant Human BEST1 cell lysate

Cat. No. BEST1-1910HCL **Lot. No.** (See product label)

SPECIFICATION

Species	Human
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
Size	100 ul
Storage Buffer	1X Sample Buffer (50 mM Tris-HCl, 2% SDS, 10% glycerol, 300 mM 2-mercaptoethanol, 0.01% Bromophenol blue)
Applications	Western Blot;

GENE INFORMATION

Gene Name	BEST1 bestrophin 1 [Homo sapiens]
Official Symbol	BEST1

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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
MIM	607854
UniProt ID	O76090
Chromosome Location	11q12
Function	chloride channel activity; contributes_to chloride channel activity; ion channel activity;

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