

Recombinant Human C16orf57, GST-tagged

Cat. No. C16orf57-10410H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Human C16orf57 protein, fused to GST-tag, was expressed in E.coli and purified by GSH-sepharose.
Species	Human
Source	E.coli
ProteinLength	1-265a.a.
Description	This gene encodes a protein with several conserved domains, however, its exact function is not known. Mutations in this gene are associated with poikiloderma with neutropenia (PN), which shows phenotypic overlap with Rothmund-Thomson syndrome (RTS) caused by mutations in the RECQL4 gene. It is believed that this gene product interacts with RECQL4 protein via SMAD4 proteins, explaining the partial clinical overlap between PN and RTS. Alternatively spliced transcript variants encoding different isoforms have been noted for this gene.
Storage	The protein is stored in PBS buffer at -20°C. Avoid repeated freezing and thawing cycles.
Storage Buffer	1M PBS (58mM Na ₂ HPO ₄ , 17mM NaH ₂ PO ₄ , 68mM NaCl, pH8.) added with 100mM GSH and 1% Triton X-100, 15% glycerol.

GENE INFORMATION

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Gene Name	C16orf57 chromosome 16 open reading frame 57 [Homo sapiens]
Official Symbol	C16orf57
Synonyms	C16ORF57; chromosome 16 open reading frame 57; UPF0406 protein C16orf57; FLJ13154; HVSL motif containing 1; HVSL1; poikiloderma with neutropenia; PN;
Gene ID	79650
mRNA Refseq	NM_001195302
Protein Refseq	NP_001182231
MIM	613276
UniProt ID	Q9BQ65
Chromosome Location	16q13
Function	molecular_function;