



Human DCLRE1C peptide (DAG-P0171)

This product is for research use only and is not intended for diagnostic use.

PRODUCT INFORMATION

Antigen Description	This gene encodes a nuclear protein that is involved in V(D)J recombination and DNA repair. The encoded protein has single-strand-specific 5-3 exonuclease activity; it also exhibits endonuclease activity on 5 and 3 overhangs and hairpins. The protein also functions in the regulation of the cell cycle in response to DNA damage. Mutations in this gene can cause Athabascan-type severe combined immunodeficiency (SCIDA) and Omenn syndrome. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Jan 2014]
Specificity	Ubiquitously expressed, with highest levels in the kidney, lung, pancreas and placenta (at the mRNA level). Expression is not increased in thymus or bone marrow, sites of V(D)J recombination.
Conjugate	Unconjugated
Sequence Similarities	Belongs to the DNA repair metallo-beta-lactamase (DRMBL) family.
Format	Liquid
Preservative	None
Storage	Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles. Information available upon request.

GENE INFORMATION

Gene Name	DCLRE1C DNA cross-link repair 1C [Homo sapiens (human)]
Official Symbol	DCLRE1C
Synonyms	DCLRE1C; DNA cross-link repair 1C; SCIDA; SNM1C; A-SCID; RS-SCID; DCLREC1C; protein artemis; SNM1 homolog C; SNM1-like protein; severe combined immunodeficiency, type a (Athabascan); DNA cross-link repair 1C (PSO2 homolog, S. cerevisiae);

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Entrez Gene ID	<u>64421</u>
mRNA Refseq	NM_001033855.2
Protein Refseq	NP_001029027.1
UniProt ID	Q96SD1
Chromosome Location	10p13
Pathway	Non-homologous end-joining, organism-specific biosystem; Non-homologous end-joining, conserved biosystem; Primary immunodeficiency, organism-specific biosystem; Primary immunodeficiency, conserved biosystem;
Function	5-3 exonuclease activity; single-stranded DNA endodeoxyribonuclease activity;