

Table S1. Primers for *RECQL4* variants

Target	Forward	Reverse
G3014A	CTGCTGGCGACCACCTATAC	CTGTGAAAGGCCTGGAAGGT
T274C/G801C	CGGGAGATTCGCTGGACGAT	GGCCAGCCGAGGGAAGATGT

Fig.S1

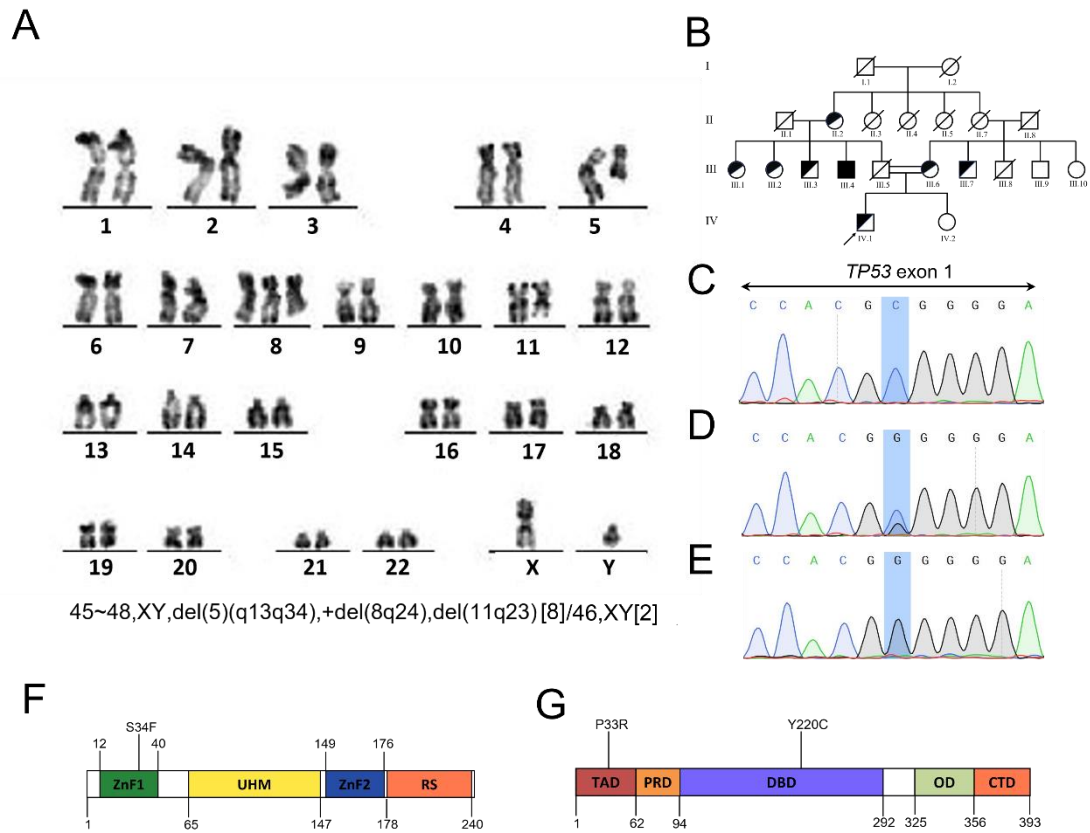


Fig.S1 Chromosomal karyotype and *TP53* germline mutation of the proband. (A) Chromosomal karyotype of bone marrow cells from the proband. (B) Pedigree of the family with eight members affected by *TP53* c.C98G: p.P33R germline mutation. Carriers marked in black, and the proband indicated by an arrow. (C-E) The presence of *TP53* c.C98G: p.P33R in homozygous state (C), heterozygous state (D), and wild type (E). (F) Somatic missense mutation was detected in codon 34 of *U2AF1*. The ZnF1 (zinc finger 1), UHM (U2AF homology motif), ZnF2 (zinc finger 2) and RS (arginine-serine rich) domains are shown. (G) Germline missense mutation in codon 33 and somatic missense mutation in codon 220 of *TP53* were detected. The TAD (transcription activation domain), PRD (proline rich domain), DBD (DNA-binding core domain), OD (tetramerization domain) and CTD (C-terminal regulatory domain) are shown.

Fig.S2

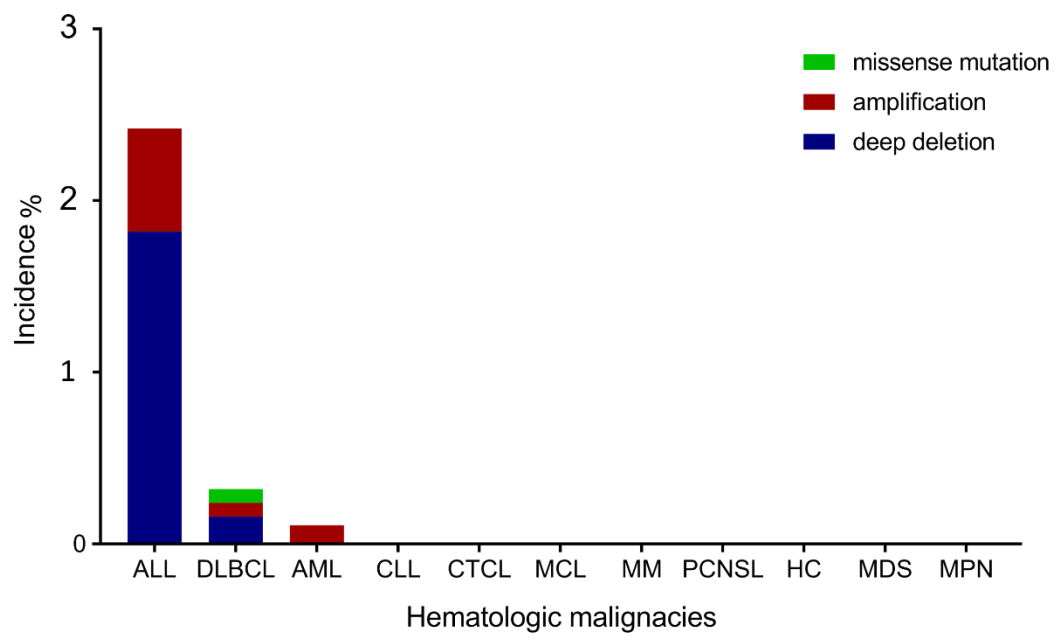


Fig.S2 Incidence of somatic *RECQL4* mutation across different hematologic malignancies. Mutational data originated from Cbioportal. ALL, Acute Lymphoblastic Leukemia; DLBCL, Diffuse Large B-cell Lymphoma; AML, Acute Myeloid Leukemia; CLL, Chronic Lymphocytic Leukemia; CTCL, Cutaneous T-cell Lymphoma; MCL, Mantle Cell Lymphoma; MM, Multiple Myeloma; PCNSL, Primary Central Nervous System Lymphoma; HC, Histiocytosis Cobimetinib; MDS, Myelodysplastic Syndromes; MPN, Myeloproliferative Neoplasms.

Fig.S3

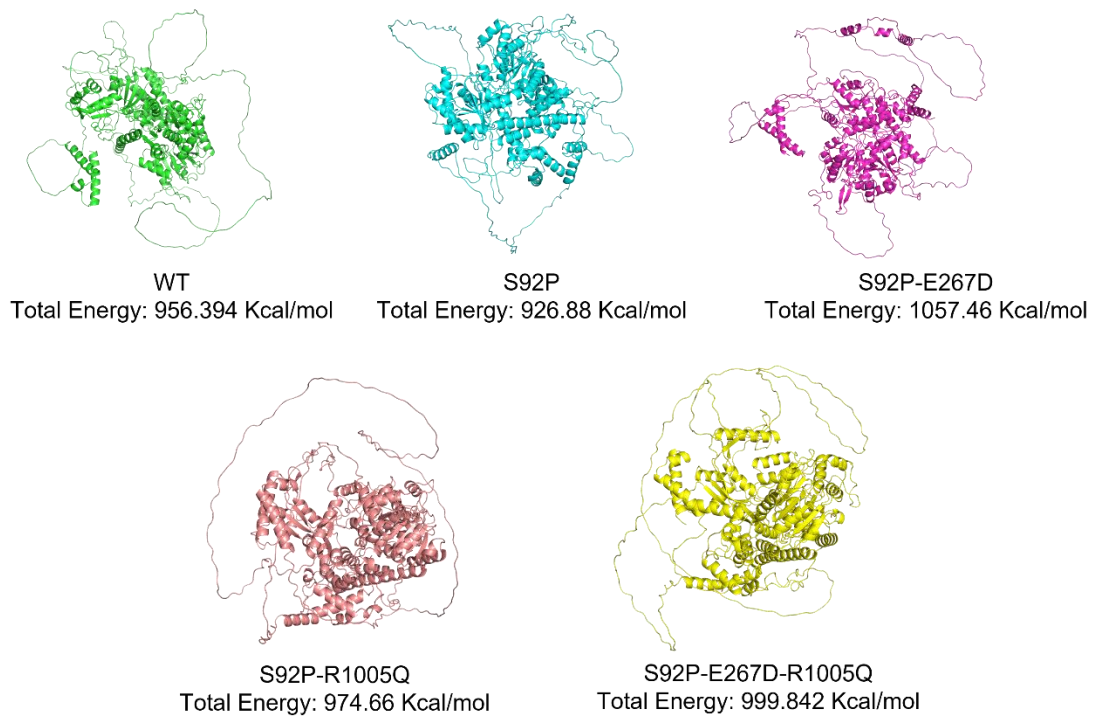


Figure S3. Structure and energy prediction of wild-type RECQL4 and RECQL4 mutants.

Higher energy of protein indicates unstable. The most unstable S92P-E267D mutant could be seen in the proband (Homozygous c.T274C: p.S92P, heterozygous c.G3014A: p.R1005Q and heterozygous c.G801C: p.E267D) and his mother (Homozygous c.T274C: p.S92P and heterozygous c.G801C: p.E267D). WT, wild-type.