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Decoding telomere gene variation through saturation genome editing: from functional genomics to clinical interpretation

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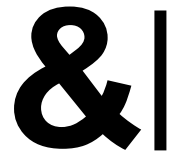
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List of Abbreviations

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List of Abbreviations

ABE Adenine Base Editor

ACMG/AMP American College of Medical Genetics and Genomics / Association for Molecular Pathology

ALT Alternative Lengthening of Telomeres

AML Acute Myeloid Leukaemia

A-NHEJ Alternative Non-homologues End Joining

ATM Ataxia Telangiectasia Mutated kinase

ATR Ataxia Telangiectasia and Rad3-related kinase

BH Bonferroni-Hochberg

bp Base pairs

BFB Breakage Fusion Bridge

CADD Combined Annotation Dependent Depletion

Cas9 CRISPR Associated Protein 9

CBE Cytosine Base Editor

CDS Coding DNA Sequence

CLL Chronic Lymphocytic Leukaemia

ClinGen Clinical Genome Resource

ClinVar Clinical Variant Database (NCBI)

CPLT Cancer Predisposition with Long Telomeres

CRISPR Clustered Regularly Interspaced Short Palindromic Repeats

CST CTC1-STN1-TEN1 complex

CTCL Cutaneous T-cell Lymphoma

DC Dyskeratosis Congenita

DDR DNA Damage Response

DSB Double Strand Break

dsDNA Double-Stranded DNA

eQTL Expression Quantitative Trait Locus

FACS Fluorescence Activated Cell Sorting

FDR False Discovery Rate

FLOW-FISH Flow Cytometry Fluorescence In Situ Hybridisation

gnomAD Genome Aggregation Database

GoF Gain-of-Function

GWAS Genome Wide Association Study

HAP1 Human near-haploid HAP1 cell line

HDR Homology-Directed Repair

HR Homologous Recombination

IF-FISH Immunofluorescence Fluorescence In Situ Hybridisation

IPF Idiopathic Pulmonary Fibrosis

iPSC Induced Pluripotent Stem Cell

KO Knockout

LFC Log2-Fold Change

LoF Loss-of-Function

MAF Minor Allele Frequency

MANE Matched Annotation from NCBI and EBI

MAVE(s) Multiplexed Assay(s) of Variant Effect

MDS Myelodysplastic Syndrome

ML Machine Learning

MPRA(s) Massively Parallel Reporter Assay(s)

NGS Next-Generation Sequencing

NMD Nonsense-Mediated mRNA Decay
NMTC Non-Medullary Thyroid Carcinoma
OB fold Oligonucleotide Oligosaccharide Binding fold
OR Odds Ratio
PAM Protospacer Adjacent Motif
PCA Principal Component Analysis
PE Prime Editing
pegRNA Prime Editing Guide RNA
(q)PCR Quantitative Polymerase Chain Reaction
PPEs Protospacer Protection Edits
RCC Renal Cell Carcinoma
scRNA seq Single Cell RNA Sequencing
SGE Saturation Genome Editing
sgRNA Single Guide RNA

SNP Single Nucleotide Polymorphism
SNV Single Nucleotide Variant
ssDNA Single-Stranded DNA
SVI Sequence Variant Interpretation Working Group
TBD Telomere Biology Disorder
TIFs Telomere Dysfunction Induced Foci
UKBB UK BioBank
UTR Untranslated Region
VAMP seq Variant Abundance by Massively Parallel Sequencing
VEP Variant Effect Predictor
VUS Variant(s) of Uncertain Significance
WES Whole Exome Sequencing
WGS Whole Genome Sequencing