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

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Citation
Obolenski, S. (2026, September 24). *Decoding telomere gene variation through saturation genome editing: from functional genomics to clinical interpretation*. Retrieved from https://hdl.handle.net/1887/4310110

Version:	Publisher's Version
License:	Licence agreement concerning inclusion of doctoral thesis in the Institutional Repository of the University of Leiden
Downloaded from:	https://hdl.handle.net/1887/4310110

Note: To cite this publication please use the final published version (if applicable).

Appendix

List of Abbreviations

English Summary

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Резюме на русском языке

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Acknowledgements

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