

Justin St. Cloud

Glenn Leadership Award

Biology Major, Public Health Minor

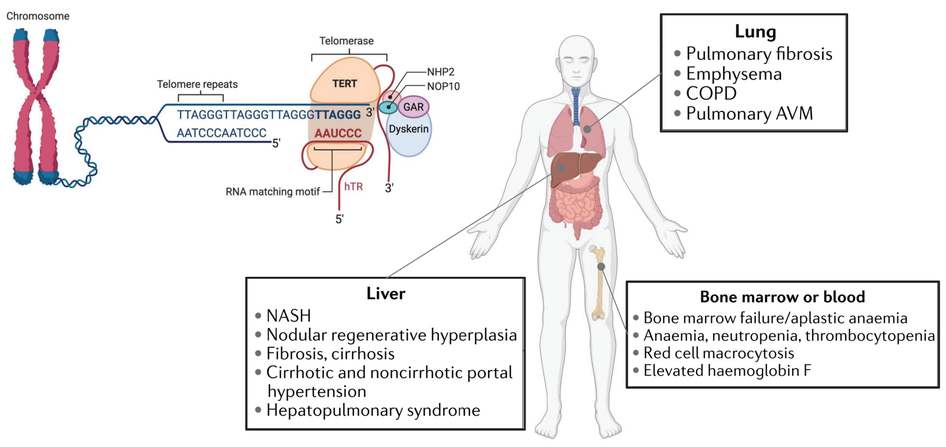
Second Year ARCS Scholar

MOREHOUSE COLLEGE

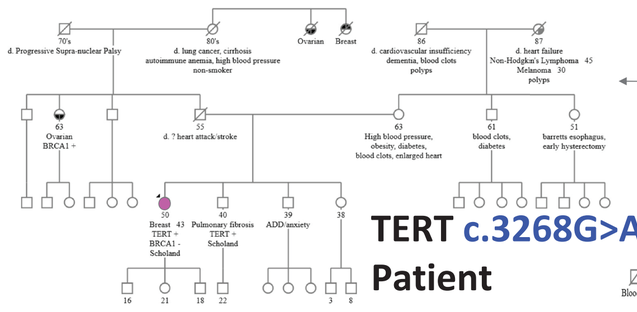
Functional Characterization of hTERT Variants in Telomere Biology Disorders

This project evaluates how the patient-derived hTERT variants G172E and V1090M affect telomerase function. Functional assays were used to help determine whether these variants may contribute to telomere biology disorders.

Telomere Biology Disorders (TBDs)



TERT c.515A>G p.G172E Patient



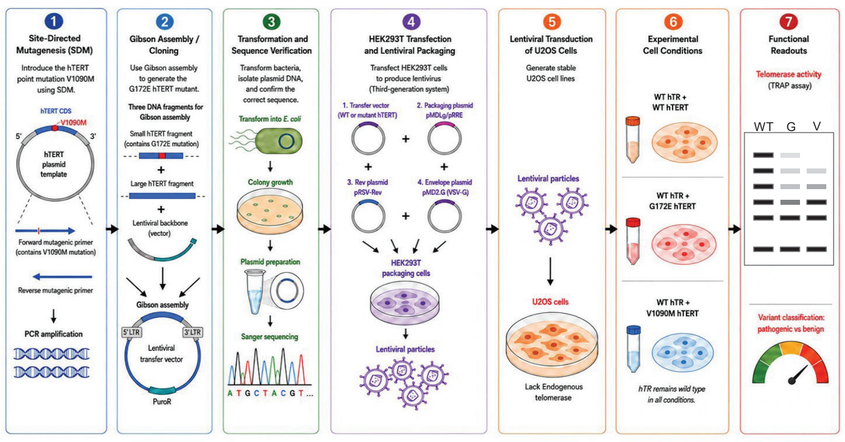
History notable for breast cancer, hematologic abnormalities, liver steatosis/fibrosis, mild pulmonary changes, and critically short telomeres (<1st percentile)

TERT c.3268G>A p.V1090M Patient

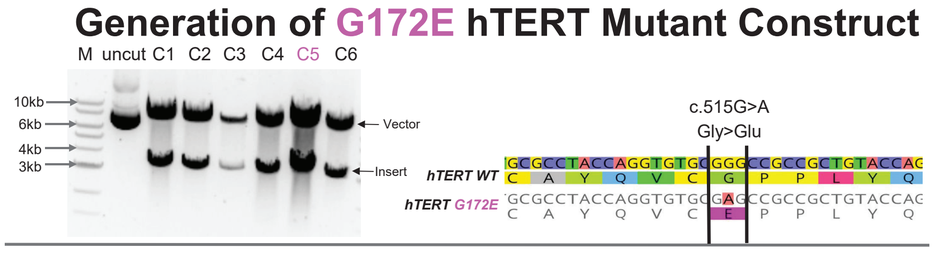
Presented with AML at age 29, later found to carry the TERT p.V1090M VUS, with markedly shortened telomeres, ongoing oncology follow-up, and referral for interstitial lung disease screening



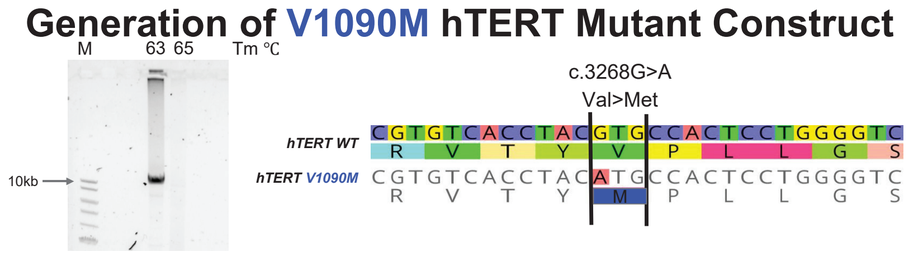
Experimental Workflow for Functional Characterization of hTERT Variants



Generation of G172E hTERT Mutant Construct



Generation of V1090M hTERT Mutant Construct



Expression of different hTERT variants in U2OS cells

G172E hTERT but not V0190M shows reduced telomerase activity *in vitro* (TRAP Assay)

SCHOLAR AWARDS CELEBRATION

250 Years of American Science: Honoring Those Who Advance It

ARCS ADVANCING SCIENCE IN AMERICA®