Summer Student Research Program
Project Description

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PROJECT TITLE (200 Characters max):

Structural Hypervariation at Subtelomeric Regions

HYPOTHESIS:

A **subtelomere** is the DNA region adjacent to the telomere, which acts as a protective cap at the ends of chromosomes. While telomeres are composed of repetitive sequences that help prevent chromosome degradation, subtelomeres contain more complex structures, including genes. These regions, rich in repetitive sequences, are often genetically unstable, making them prone to structural changes such as duplications, deletions, or rearrangements. This instability has been implicated in aging, disease, and genetic disorders. Additionally, the evolution of subtelomeric regions contributes to differences between species; for example, humans share approximately 98.5% of their DNA with chimpanzees, with the key differences often arising from the presence and arrangement of repetitive sequences, including subtelomeres.

The mechanisms behind subtelomeric instability remain unclear. One hypothesis suggests that repetitive sequences in subtelomeres serve as binding sites for transcription factors. We will test the hypothesis that DNA-binding proteins obstruct replication forks, causing DNA breaks and triggering subsequent structural changes in these regions.

PROJECT DESCRIPTION **(Include design, methodology, data collection, techniques, data analysis to be employed and evaluation and interpretation methodology)**

We have established several genetic systems in budding yeast cells. Additionally, we will generate various constructs using PCR and integrate them into subtelomeric regions. To confirm genetic variation, we will use PCR and Southern blotting techniques.

SPONSOR'S MOST RECENT PUBLICATIONS RELEVANT TO THIS RESEARCH:

PMID: 26263073

THIS PROJECT IS: laboratory

THIS PROJECT IS CANCER-RELATED
Chromosome rearrangements are often observed in cancer cells.

THIS PROJECT IS HEART, LUNG & BLOOD- RELATED
Pneumocystis remains a significant cause of disease in humans with immunodeficiencies, particularly those with HIV/AIDS, organ transplants, and increasingly in patients receiving immunodepleting monoclonal antibody treatments. As an atypical fungus, **Pneumocystis** has highly adapted to the mammalian lung environment. The genes responsible for its pathogenicity are located in the subtelomeric regions of its genome and are prone to acquiring mutations.

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THIS PROJECT INVOLVE RADIOISOTOPES? No

THIS PROJECT INVOLVES THE USE OF ANIMALS No

THIS PROJECT INVOLVES THE USE OF HUMAN SUBJECTS? No

THIS PROJECT IS SUITABLE FOR:
ALL STUDENTS

THIS PROJECT IS WORK-STUDY: **No**

THIS PROJECT WILL BE POSTED DURING ACADEMIC YEAR FOR INTERESTED VOLUNTEERS: **Yes**

WHAT WILL THE STUDENT LEARN FROM THIS EXPERIENCE?
Genetics, Concepts of genomic rearrangement and mutagenesis