Mentor
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Project Title
Identification of causative genetic variants in pharmacogenomic-associated genes

Background and significance
Pharmacogenomics research aims to identify genomic variations that can be used to predict which individuals will benefit from taking an established or emerging medication, those predisposed to developing an adverse reaction, or to determine the optimal drug dosage for an individual. While > 1100 genes have been correlated with variable dosage or drug response, only 15 genes have corresponding guidelines for implementation in clinical practice, known as the Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines. One reason for the poor rate of clinical implementation is that the majority of the identified pharmacogenomics variants are not causal but in linkage disequilibrium (co-transmitted) with the casual variant(s). Without the causal variants, clinical tests using the correlated variants to predict drug response or dosage lack in accuracy, and is especially variable by ethnicity.

Project objective
The aim of this project is to identify potentially causal variants in well established pharmacogenomic genes.

Skills developed
The student will learn to navigate next-generation sequencing data (e.g. from the 1000 Genomes Project, UK2K Project) to identify all genetic variants (single nucleotide variants, SNVs) in known Pharmacogenomic genes, use annotation software to determine which are coding for an amino acid change or splice variant and which are more likely to have detrimental effects on the resultant protein, and calculate the linkage disequilibrium or correlation with the initial associated pharmacogenomic variants.

Pre-requisites
Fluency in UNIX, R, Biomedical computing/informatics

Project type
Undergraduate project
Masters project