Developing an Integrated Pharmacogenomics Platform to Enhance Medication Safety in Chronic Disease Management

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Background:

Pharmacogenomics (PGx) has become an integral part in precision medicine by identifying genetic variants that influence drug response. However, accurate detection of genetic variants requires a robust platform which can analyze, interpret and report the genetic data in line with clinical guidelines. This study focuses on developing a comprehensive PGx analysis platform (PGx-Core) handling the output of various sequencers, covering quality control, variant calling, haplotype phasing, star-allele annotation, and clinical reporting.

Methods:

Since the platform is developed following nf-core guidelines, it ensures reproducible workflows, portability and broad compatibility with high-performing clusters and cloud-based systems. PGx-Core has three modules that can analyse long-read as well as short-read WGS, WES, and panel data sequenced using Illumina, PacBio and ONT.

Results:

PGx-Core will be utilised to call star-alleles on the UK Biobank as well as the AllofUs cohort to create a landscape of star-allele among different ethnic populations and compare novel and risk-associated star-alleles, which can help to reduce the adverse drug response.

Conclusions:

This platform will help to create a clinical impact by producing a pharmacogenomics report implementing pharmacogenomics guidelines.