EP18.010 Pharmacogenetic Profiling from Decentralized Whole Blood Collection and PCR-free Library Preparation

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Introduction

- Adverse drug events remain a significant clinical burden, often driven by variability in drug metabolism, transport and response.
- Pharmacogenetic profiling of ADME/Tox genes can identify variants linked to altered drug response and increased toxicity risk.
- Venous blood collection can be challenging to scale in decentralized studies due to phlebotomy, cold-chain shipping and storage requirements.
- This study evaluates a decentralized workflow combining truCOLLECT® Whole Blood Collection Kit and truCOVER® WGS PCR-free Library Prep Kit for pharmacogenetic profiling from finger-stick samples.

Materials and Methods

- Pediatric participants and family members were enrolled across decentralized clinics in Puerto Rico; matching venous and finger-stick samples were collected to compare conventional and decentralized methods.
- Finger-stick samples used the truCOLLECT device, stabilized by active desiccation, and shipped/stored at room temperature; venous samples required freezing and dry-ice shipment.
- DNA was extracted from truCOLLECT samples using the truCOLLECT DNA Extraction Kit (96).
- Libraries were prepared with the truCOVER WGS PCR-Free Library Prep Kit and compared to matching venipuncture samples on yield, fragment size, coverage, and pharmacogenetic concordance.

Results

Yield and Fragment Size

	truCOLLECT Samples (N=47)	Venipuncture Samples (N=15)
Average Library Yield (ng)	94.3 (17.6 %CV)	72.1 (14.7 %CV)
Average Library Fragment Size (bp)	751 (6.2 %CV)	681 (4.1 %CV)
Sample Number	15 affected siblings 32 family members	15 affected siblings (matched)

Figure 1. NGS library performance from truCOLLECT and venipuncture-derived DNA. Average library yield and library fragment size were compared between truCOLLECT whole blood samples and venipuncture samples. truCOLLECT-derived DNA generated PCR-free libraries with comparable or higher average yield and fragment size relative to venipuncture-derived DNA.

% of Reads Mapping to Genome

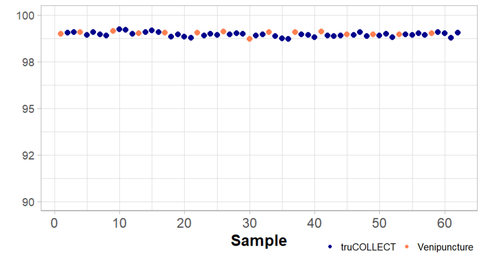

Figure 2. Consistent pharmacogenetic target coverage across truCOLLECT and venipuncture samples. Coverage performance across sequenced samples run on a NovaSeqX, showed consistently high values for both truCOLLECT and venipuncture-derived libraries, supporting the suitability of the decentralized sample collection workflow for pharmacogenetic profiling.

% GC Coverage Across Pharmacogenetic Target Loci

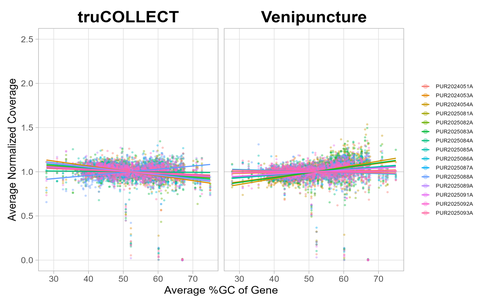

Figure 3. Coverage uniformity across pharmacogenetically relevant genes with variable GC content. Average normalized coverage was assessed across target genes with GC contents ranging from approximately 32–68%. truCOLLECT-derived libraries showed stable coverage across this GC range, comparable to venipuncture-derived libraries.

Concordance: truCOLLECT and Venipuncture

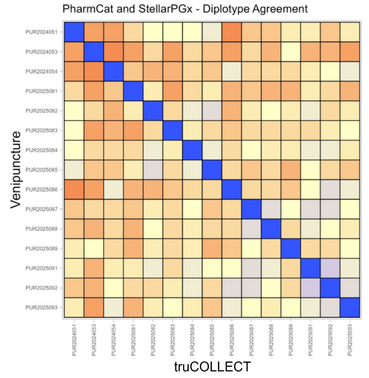

Figure 4. Diplotype agreement between truCOLLECT and matched venipuncture samples. Heatmap showing PharmCAT and StellarPGx diplotype agreement between matched truCOLLECT and venipuncture samples. 100% (95% CI: 99.9%-100%) agreement between sample types supports the use of truCOLLECT-derived DNA for pharmacogenetic profiling.

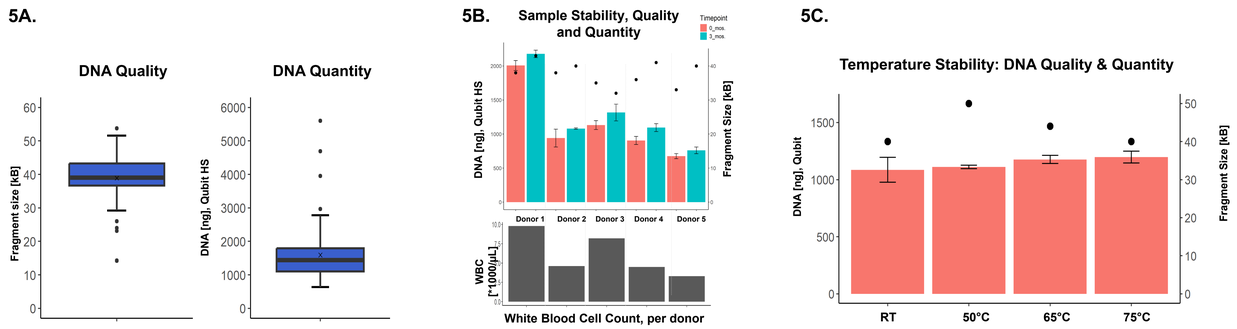

Figure 5A. DNA quality and quantity of truCOLLECT whole blood samples. DNA extracted from 94 truCOLLECT samples showed suitable fragment size and quantity for PCR-free library preparation. **Figure 5B and 5C: DNA sample and temperature stability:** DNA quantity and fragment size were maintained across donor samples, storage timepoints and temperature-stability conditions, supporting room-temperature sample collection and shipping.

Conclusions

- The truCOLLECT Solution paired with truCOVER WGS PCR-free library preparation provides a robust decentralized pharmacogenetic profiling workflow.
- DNA extraction and purification from up to 96 truCOLLECT samples can be completed in 2 hours, followed by library preparation in under 3 hours, supporting same-day sample-to-library processing.
- Room-temperature stabilization and standard shipping simplify logistics versus venous collection, which requires cold-chain handling.
- truCOLLECT-derived DNA yielded high-quality PCR-free libraries with performance comparable to matched venipuncture samples.
- These findings support decentralized finger-stick collection for scalable pharmacogenetic studies and broader genetic screening to reduce adverse drug events.

References

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