Democratizing Pharmacogenomics: Bridging NGS, AI, and Clinical Practice

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Pharmacogenomics (PGx) is a cornerstone of precision medicine, yet its clinical adoption is often hindered by the limitations of standard SNP arrays and the complexity of interpreting genetic data for drug response. In this presentation, we introduce the new SeqOne PGx Module, a transformative addition to our GermVar workset designed to unlock the full potential of Next-Generation Sequencing (NGS) data—from Capture panels to WES and WGS.
We will demonstrate how this module moves beyond the "known variant" limitations of SNP arrays by performing accurate Star Allele calling and identifying novel variants of interest. A key focus will be our integration of DiagAI, which allows for the identification of rare or novel deleterious variants (e.g., Loss of Function) in PGx genes that could significantly alter patient metabolism but are missed by traditional screening methods.
Finally, we will showcase our new AI Copilot, powered by an MCP (Model Context Protocol) grounded in ClinPGX guidelines. This feature bridges the gap between complex genomic data and clinical utility, allowing clinicians and pharmacists—regardless of their PGx expertise—to ask natural language questions regarding drug-gene interactions and posology adjustments.