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Implementation and Cost-Effectiveness of Whole-Genome Sequencing for Acute Leukemias in Latin America

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Abstract

Background

Leukemia outcomes in Latin America remain poor despite major therapeutic advances in high-income countries. Despite precision treatments being available, modern genomic diagnostics to inform these treatments are not available across the region. In 2020, approximately 38,000 people in Latin America were diagnosed with acute leukemias, and nearly 27,000 died from their disease. Although genomic profiling has transformed risk stratification, treatment selection, and transplantation decisions in the United States and Europe, many Latin American centers lack access to cytogenetics, fluorescence in situ hybridization, and molecular testing. Consequently, patients are frequently treated without the genomic information required for precision care. The Genomics of Leukemia in Latin America (GEMA) study was developed to address this disparity.

Methods

GEMA is an international, multi-institutional, prospective cohort study embedded within the Leukemia in Latin America Mapping and Sequencing initiative (LLAMAS; llamas.lat). The study will collect bone marrow aspirate or blood, perform on-site WGS with local partners, and transfer raw sequencing data for analysis using the ChromoSeq workflow developed at Washington University (Duncavege, NEJM 2022). The specific aims of this study are to: 1) characterize the clinico-demographic and cytomolecular features of leukemia in participating centers; 2) evaluate the detection of clinically relevant variants by on-site WGS, and 3) perform a contextualized healthcare cost-effectiveness analysis comparing WGS with existing workflows.

Results

The study is enrolling in two countries. It has established a collaborative framework for prospective leukemia enrollment, standardized biospecimen collection, local WGS implementation, and bioinformatic analytical validation. The initial network includes participating institutions in Paraguay and Peru and provides an infrastructure for planned regional expansion. The study is designed to quantify the diagnostic yield of WGS, its effects on disease classification and clinical decision-making, and its cost-effectiveness contextualized to each health system.

Conclusion

This study in progress will provide a scalable framework for integrating comprehensive genomic diagnostics into leukemia care in Latin America. This initiative has the potential to improve access to genomically informed treatment, increase the representation of historically understudied populations, and establish a durable regional platform for biologic discovery, outcomes research, health-economic evaluation, and therapeutic trial development.

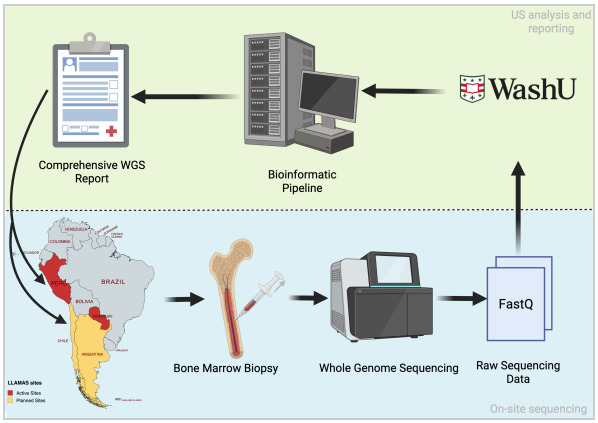


Figure 1.

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