

2<sup>nd</sup> International Conference & Exhibition on

# Tissue preservation and Bio-banking

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## Biospecimen repository genomic annotations in the precision medicine era

Biorepositories play a crucial role in biomedical research on human diseases. In the current climate of precision diagnostics and personalized therapies, it is critical to annotate biospecimens, which frequently undergo molecular testing, with available genomic data. Molecular assays used for clinical testing are in evolution, ranging from analysis of single genes, a panel of genes, to whole exome mutation profiling. For clinical diagnosis and research, genomic annotation includes the type of analysis performed, whether or not sequence variants were found, and the clinical significance of the detected variants. Curated diagnostic genomic data using standardized nomenclature is directly transferred from our CLIA-certified and CAP-accredited Molecular Pathology Laboratory. A software bridge for specimen and data management was created in house. Mutation results with annotation are populated in specific gene fields in our i2b2 research data warehouse, as approved by our IRB. The genomic annotation for research may include more extensive information and may extend to the scope of whole genome analysis. Therefore Fastq and BAM files are also stored to enable re-analysis of whole exome and whole genome sequencing. The creation of a framework for a data warehouse that assembles annotation with clinical, pathologic, and genomic profiles provides a powerful tool for biomarker discovery and the design of clinical trials for personalized medicine. The accessibility of these data results in “democratization” of information. The sharing of data will drive clinical, translational, and basic research that will enhance patient care in the future.

## Biography

Stephen Peiper is the Peter A Herbut Professor and Chairman of the Department of Pathology, Anatomy and Cell Biology at Thomas Jefferson University/Jefferson Medical College. Previously, he was a Professor of Pathology and Chairman of the Department of Pathology in the School of Medicine at the Medical College of Georgia (MCG). He was also a Distinguished Scholar of the Georgia Cancer Coalition, an active member of the executive board of the Georgia Center for Oncology Research & Education, and was appointed as the inaugural Senior Associate Dean for Translational Research at MCG. He has published over 160 peer-reviewed publications and authored over 30 book chapters and symposia. In addition, he served as a Section Editor for the *Journal of Immunology* and is currently on the editorial boards of *Human Pathology* and *Biotechnology Healthcare*. His research interests include the molecular characterization of hematopoietic cells and their neoplastic counterparts and the application of emerging molecular technologies to diagnostic pathology.

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