Disruptive Technology Empowering Precision Medicine (D-TECH) Task Force- 2024 Updates

Annual Research Retreat Summary

Mission: The overarching goal of the Disruptive Technology Empowering Precision Medicine (D-TECH) strategic area is to transform research and healthcare at UAB by defining new ways to integrate cutting- edge technologies into biomedical research and precision healthcare. The D-TECH strategic area encompasses an expansive mission, effectively covering any research at UAB that relies on the rapidly advancing infrastructure in informatics, artificial intelligence, Electronic Health Record (EHR), and high dimensional multimodal data management on one hand, and the expanding experimental, technical informatics, and computing infrastructure on the other hand.

Task Force: The D-TECH Task Force transitioned to Co-leads Ralph Zottola and Javier Neyra. Membership in the Task Force was updated. A challenge for the task force has been the broad scope of this domain. We believed that it would be more productive to focus our work within domain-specific task-focused subgroups: big data, AI, and Digital Health; Electronic Health Records; Imaging/Biomarkers; and Precision Medicine and Omics. We developed a consensus for the development of a platform-agnostic genomic clinical decision support (CDS) system.

DTECH: Genomic Clinical Decision Support Platform

Genomic sequencing is becoming cheaper, more reliable and more valuable – it is inevitable that patients' genomes will become a standard part of electronic health records (EHRs). Genomic data are already used in clinical workflow to support medical decision-making. Current EHRs, however, although effective in managing conventional CDS (e.g., best practice advisory alerts, drug allergy alerts), fall short on managing genomic-based CDSs. They are not designed for managing the level of computation needed for genomic-based risk-classification and actionable recommendations. Furthermore, if you have seen one EHR, you have seen one EHR, in other words, CDS systems built locally are not easily scalable to other EHRs and populations. Interoperability is still a problem in CDS design and research. Knowledge of actionable genetic variants is growing constantly, and the rate of effectiveness/implementation testing is suboptimal. The rate growth will become exponential as learning health systems (a) get better at learning and (b) have more genomic data to learn from. A new platform for scalable genomic CDC would be innovative and disruptive, allowing interdisciplinary collaborations, multimodal data pipeline development, AI-risk classification, and implementation testing.

We will develop and deploy a new standards-based, scalable, integrated genomic decision support platform that is EHR agnostic and scalable.

We acknowledged several requirements for such a system. There is a dire need for a new and scalable method for incorporating knowledge into CDS, which does not require new programming or increased maintenance with every new genomic discovery. It must include a new method for delivering the anticipated increase in knowledge to the point of care that does not disrupt workflow with, for example, alert fatigue. It must be EHR platform agnostic. The new method should also be able to deliver curated information to both clinicians and patients and be readily available for quantitative and qualitative analyses for implementation testing. Finally, it should scale for any prevalent complex disease (e.g., cancer, heart failure, diabetes, kidney disease, and others).

The approach we are developing considers a few assumptions. First, we recognize that integration into Cerner is tedious. Cerner will be replaced with EPIC, but that will take at least two years, with innovation activities delayed even further. EHR vendors and capabilities may change so this new platform needs to be EHR agnostic. There are standards for CDS that are external to any EHR but can be integrated with them (especially EPIC) that we will leverage. Examples include CDS standards based on Fast Healthcare Interoperability Resources (FHIR), such as OpenCDS and CDS Hooks. At UAB, we have many rich datasets available for a pilot project. We will develop the platform using AGHI data which includes patient genomic sequence data coupled with EHR data. Additionally, preliminary work on assembling genetic variants of interest and characterizing the types of CDS to which they can contribute has already been done. Thus, we already have many critical components.

There are many benefits if we are successful beyond just the potential for a novel and disruptive technology platform. Providing actionable information at the point of care will result in better care leading to improved patient outcomes. Adherence to an open-standard, platform-agnostic approach would facilitate integration with any EHR anywhere. This also allows for incremental enhancements independent of vendor production schedules. We plan to add a consumer facing (patient and clinician) interface that can elevate patient awareness, engagement, and empowerment with their own care. This could also increase opportunities for patient recruitment for clinical trials and has the potential to inform basic science that can leverage an automatically maintained repository of known gene-condition and gene-drug pairs.

Deliverables and Resources:

- AGHI genomic and clinical data to start. Other data sets as time and resource allow.
- Multimodal data pipeline development with standardized data harmonization methods
- A programmer to deploy and maintain, database of actionable variants and CDS technology.
- Effort from a clinical informatician/MD and a genomic medicine expert to provide the platform with actionable genomic information in the form of variants, necessary clinical attributes, types of recommended actions, and action messages.
- A clinical informatician to collaborate with the programmer to develop a CDS application that matches a patient's genomic data to the variants and clinical data to the clinical attributes to generate a list of recommendations.
- A clinical informatician/MD to assemble a report of recommendations for a patient, organized by recommendation type.
- Effort from a human factors expert to perform heuristics usability evaluation of the user interface of the CDS system to be presented to end users.
- An informatics/implementation team to evaluate the CDS function, maintain the knowledge base, assess usefulness, effectiveness, and usability.

In summary, current approaches are proprietary and inefficient. This proposed platform is designed to keep up with the rapidly increasing knowledge about actionable genes and their associated conditions. We are fortunate to have a rich dataset such as the AGHI to develop a platform that can scale to much larger patient populations. It will lead to better integration of genomic research with patient care leading to improved patient outcomes. It will position UAB as a leader in the national learning health system.

Current D- TECH Membership
Carlos Cardenas, James Cimino, James Cimino, Tiago Colicchio, Alexander Mackinnon, Merry Lynn McDonald, Matthew Might, Javier Neyra (co-lead), Ralph Zottola (co-lead)