Interdisciplinary training to build an informatics workforce for precision medicine

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1. Introduction

In 2015, the federal government announced its intention to devote $215 million in funding to the Precision Medicine Initiative in the President's 2016 budget (Collins and Varmus, 2015). The translation of discoveries made through this initiative into healthcare practice will require a multidisciplinary approach to implementation into the healthcare delivery system. At the core of such efforts will be the participation of biomedical informaticians and data scientists that possess broad and diverse expertise relative to the application areas of such innovations, spanning a spectrum from bio-molecule to patients to large-scale populations. At the same time, most biomedical informatics and data science training tend to be organized into disciplinary subdomains (bioinformatics, clinical informatics, public health informatics, etc.), with minimal opportunity for trainees to gain exposure to or experience working across and between disciplines and application areas (Hersh, 2008). The preceding challenges and opportunities are further magnified, given the movement in the field of Biomedical Informatics (BMI) and Biomedical Data Science (BDS) to align workforce development and training efforts with community-defined and common competencies (Kulikowski et al., 2012). Via such efforts, it has become clear that access to diverse and representative practical environments for trainees at all levels is needed in order to ensure that experiential learning is well grounded in “real world” contexts. In particular, and as it relates to the broad precision medicine problems space, there is a need to expose trainees to opportunities where they can demonstrate mastery of: 1) heterogeneous data, information, and knowledge representation, analysis, and manipulation across the aforementioned spectrum from bio-molecules to patients to populations; 2) the assessment of the effects, value, and cost of healthcare information technology (HIT) platforms and informatics or data science based interventions; and 3) the re-engineering and management of organizational and processes changes informed by the aforementioned areas. Of note, the pursuit of projects that exhibit dimensions of such competencies is highly influenced by environmental factors, thus necessitating their pursuit in multiple settings and contexts (including academic health centers, integrated delivery networks, and industry-based research and development efforts). Furthermore, the preceding needs and requirements are being amplified by the evolving landscape in which BMI and BDS innovators and practitioners are expected to make substantive contributions, again, such as recent foci on precision medicine (Collins and Varmus, 2015) and the concomitant goal of leveraging of HIT to achieve a triple aim of reduce costs and increased quality and safety of healthcare delivery (Sheikh et al., 2015).

2. Perspective of the healthcare delivery system

As an exemplar of the type of environments in which precision medicine must ultimately be operationalized, Geisinger Health System (GHS) represents a large integrated delivery system caring for a large
predominately rural population in central and northeast Pennsylvania. GHS was an early adopter of electronic health record systems (EHR) across the inpatient and outpatient setting and led initiatives to develop Health Information Exchanges (HIE) to gather clinical information from providers and facilities affiliated with but not part of GHS. As an integrated system approximately 40% of the patients treated in GHS facilities are covered by Geisinger Health Plan (GHP) which includes a variety of commercial, Medicaid and Medicare products. Thus claims data are available for a substantial fraction of the patient population. These rich data assets provide an exceptional opportunity to drive quality improvement activities and health services research. Use of standardized and generalizable data models has allowed GHS to participate in multisite research networks such as the Health Maintenance Organization Research Network (HMORN soon to become the Health Care System Research Network). GHS has taken a lead role in the collection, storage and use of patient entered data to improve patient care and experience. This has supported participation in one of the Patient Centered Outcomes Research Institute (PCORI)-funded Patient Powered Research Networks.

GHS has also participated in projects that are in the precision medicine space including the electronic Medical Records and Genomics (eMERGE) network (Gottesman et al., 2013) and the Clinical Genome Resource (ClinGen) project (Rehm et al., 2015). In 2014 GHS began a partnership with Regeneron Pharmaceuticals to perform whole exome sequencing and genome-wide genotyping on as many as 250,000 GHS patients over the course of 5 years. The availability of this sequencing data on patients with deep longitudinal EHR data available to derive clinical phenotypes can be used to support discovery research. The sequences can also be used to identify variants in clinically actionable genes that could be returned to patients who may be unaware of their risk due to a lack of a personal or family history of disease.

Efforts are now being taken to realize a strategic plan that emphasizes research and clinical implementation of precision medicine. This necessitates harmonization of current and future activities in health services, genomics and informatics research and clinical implementation to a degree not previously required. Evaluation of the strategic plan identified gaps in expertise that could impact the translation of the genomic data into improved health outcomes. Many of these exemplify the problems described in Section 1. As an example, participation in the eMERGE network led to the development of a data core with analysts that specialize in extracting phenotypes from the EHR data. Additional investment in the analytic team was spurred by the Regeneron project. However, only one bioinformaticist was available for the eMERGE network projects. At the initiation of the Regeneron project, only one bioinformaticist was available to analyze the sequence data. This was completely insufficient given the anticipated volume. The eMERGE pharmacogenomics project was designed to return actionable pharmacogenomics information to inform drug choice and dosing. Implementation of clinical decision support (CDS) in the EHR required interaction with the clinical informatics team. This team has limited contact with research. As a result several problems were encountered that delayed implementation of the CDS including prioritization of research against clinical requirements, novel workflows not previously encountered in the EHR, and developing reports necessary for the research project. These experiences made it clear that GHS would either have to invest significant resources to develop the multi-disciplinary capabilities internally, or develop collaborations with outside institutions to obtain access to needed expertise. The latter approach was pursued. To support the return of secondary findings from the Regeneron exomes GHS contracted with an outside laboratory to analyze the sequence through their existing informatics pipeline to identify pathogenic variants in 76 genes determined to be clinically actionable by Geisinger leadership. For the eMERGE-PGX project GHS had no research informaticists with clinical training, so a sub-contract with an informaticist at an academic medical center was needed.

Our experiences in the above endeavors also led to the determination that there was an increasing need for clinical informaticists to engage in such projects. In response to such a workforce development objective, GHS pursued and was recently approved to offer a clinical informatics fellowship training program. However, GHS does not have a biomedical informatics academic program that is positioned to offer the didactic content necessary for such fellows. As such, GHS has partnered with The Ohio State University Department of Biomedical Informatics (OSU-BMI) to utilize a series of online courses that will be made available to GHS clinical informatics fellows to fulfill such curricular content needs. Of note, the GHS mission emphasizes includes both outstanding clinical care and research. Therefore, the clinical informatics fellowship will include engagement with research activities at all stages of training. Fellows will be expected to complete a capstone project as part of their training. This project, while clinically focused, will have a scholarly component such that the fellow will not only solve a clinical problem but will be expected to contribute to generalizable knowledge in informatics. Such projects will be conducted with multi-institutional mentoring teams, building upon the preceding OSU-BMI partnering agreement. Realizing the ideal of a learning healthcare system will require a softening of the distinction between clinical care and research necessitating a solid grounding in both disciplines to be a successful leader. Further, doing so will also require the demonstration of collaborative and team based approaches to research and training that employ and leverage unique and complementary competencies and expertise, such as that afforded by the partnering model spanning GHS and OSU-BME.

The end result of the experiences noted in this section was a realization that a collaborative and multi-institutional approach, as opposed to a traditional single institution model provided many more opportunities and benefits and has been adopted as the preferred model going forward. Of course for this to work there should be value to the collaborators that goes beyond simple financial remuneration for services.

3. Perspective of the academic institution

3.1. The Ohio State University

The training programs within OSU-BMI are designed and conducted in concordance with competency-based curricular models and heavily emphasize experiential and project-based learning so as to demonstrate mastery of those competencies. These educational initiatives span a spectrum from practice-oriented certificate and master's degrees to pre- and post-doctoral degrees that focus on rigorous research and evaluative training in the field. A challenge to achieving this initiative is the relative lack of opportunities for the students to work in clinical settings and a lack of clinical data that would support adequate analytic experience to meet the identified competencies. While the OSU Wexner Medical Center (OSUWMC) provides access to clinical and operational data as well as experiential learning opportunities, by virtue of its roles as a tertiary care provider, it does not provide trainees with a full spectrum of options spanning areas such as primary care and population level health. As such, the opportunity to partner with a large integrated healthcare delivery system such as GHS with extensive and multi-level data assets provides a substantial benefit to the department's workforce development programs, enabling trainees to access a truly comprehensive "living laboratory."

3.2. Pennsylvania state university

The Pennsylvania State University (PSU) training program in Bioinformatics and Genomics (BG), with an NIH funded predoctoral training program complement "Computation, Bioinformatics, and Statistics (CBIOS), is designed to prepare scientists for a cross-disciplinary career in genomics with research spanning life sciences, computer science, bioinformatics, and statistics. These programs are well suited to prepare
scientists for the challenges emerging in high-throughput genomics including whole genome sequencing and genome-wide genotyping. In addition to the BG and CBIOS including whole genome sequencing and genome-wide genotyping, scientists for the challenges emerging in high-throughput genomics.

As precision medicine propels into the future, where genetic and genomic data will be tied to clinical information in the form of electronic health records, the skills to manipulate, analyze, and interpret these data will be critical. While PSU has great strength in bioinformatics, statistics, and genomics, there is limited availability and expertise in accessing electronic health records. Therefore, partnering with a large integrated healthcare delivery system will enable scientists in these programs an opportunity for cross-training in clinical data integration with the genomic data.

3.3. The case for collaboration

Cross disciplinary training is essential if the potential of precision medicine is to be achieved. The synergy between academic institutions and a healthcare delivery system provides outstanding opportunities for each discipline’s respective strengths to elevate the partnering institution in complementary areas. Through strong collaborative training environments, researchers and clinicians will be provided the best of both worlds. In light of the evolving landscape that such trainees will be expected to work in, partnerships between GHS, OSU-BMI and PSU can expand the scope and scale of opportunities for these types of experiential learning. The opportunities for clinical informatics fellows as well as master’s and doctoral level BMI students to work together will broaden the perspective of these trainees positioning them to lead the movement to learning health care systems, especially in an era of increasing focus upon interdisciplinary education and practice. Of note, this approach also necessitates developing adjunct faculty positions, as well as an infrastructure for collaborative research and systematic study of the best models to foster fruitful partnerships between BMI students and clinical informatics fellows.

By applying these goals we have created a “living laboratory” by which our trainees will gain hands-on and application level knowledge in both an academic health center and an expansive integrated delivery network. We anticipate that individuals will gain critical and comparative experience with regards to areas such as (but not limited to): 1) mechanisms to integrate and reason upon comprehensive collections of clinical phenotype, bio-molecular phenotype, and payer data, so as to support explorations of linkages between precision diagnostics or therapeutic and the costs or outcomes of healthcare delivery; 2) systems engineering and operations research methods that can be utilized to optimize the use of EHR platforms so as to improve care delivery given a set of “triple aim objectives” and enhanced biological understandings of health and wellness; and 3) implementation science issues surrounding the translation of biological discoveries into precisely targeted diagnostics and therapeutics into heterogeneous care delivery environments and settings. In doing so, we believe that our trainees will be well prepared to make major contributions to biomedical research and healthcare delivery in what is a rapidly evolving and extremely complex environment. As such, we envision the training partnership between OSU-BMI, PSU and GHS as an exemplar of the settings in which BMI training will increasingly be conducted in the near to long term time horizon and will represent a model approach to such interdisciplinary research, training and implementation of precision medicine.

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