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Bioinformatics and Medicine: Bringing Data to the Bedside

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Abstract

From being an empirical and experience-based practice, modern medicine has transformed into a codified and research-based discipline, known as Evidence-Based Medicine (EBM). Though EBM has greatly enhanced the quality of medical practice through population-scale clinical trials, it still has limitations in managing biologically diverse patient populations, especially when dealing with clinical outliers who respond in an unusual way to standard treatments. With the rapid progress in genomics, proteomics, and high-throughput technologies, there has been an unprecedented surge in biological and medical data, providing new avenues for further refinement in diagnosis, prognosis, and therapy selection in individual patients. Clinical bioinformatics has emerged as an important translational discipline to fill the gap between bench and bedside, and its application in conjunction with genomic variations, biomarkers, pharmacogenomics, and Electronic Health Records (EHRs) has paved the way for the transition to Personalized Precision Medicine (PPM), in which preventive and therapeutic approaches are tailored to individual biological characteristics. The increasing trend in big data from high-throughput technologies, medical images, and wearable devices has further enhanced predictive modeling and patient stratification, although concerns regarding data quality, compatibility, and governance issues still need to be addressed. Large-scale collaborative projects in genomics and infrastructure development are beginning to address these issues through large-scale analysis without compromising patient confidentiality. Clinical translation also requires infrastructure development, guidelines, and a clinical bioinformatician workforce proficient in interpreting complex genomic data and able to communicate this to clinicians. This article will highlight the evolution of EBM to PPM, how bioinformatics facilitates this evolution, current challenges in its implementation, and future requirements in developing a predictive data-driven framework in clinical practice.

Keywords: Modern medicine, evidence-based medicine, personalized precision medicine, clinical bioinformatics

"The art of medicine is founded on conjecture and improved by murder".
– Sir Astley Cooper, Guys Hospital

"Cured yesterday of my disease, I died last night of my physician."
– Matthew Prior, The Remedy Worse than the Disease (1714)

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INTRODUCTION

The field of medicine, from its roots in sorcery and witchcraft, to modern medicine, has undergone humongous transformation over the past millennia, with the most significant advances taking place over the previous two centuries [1]. Surgical techniques have been refined, reliable diagnostic tools discovered, and breakthroughs became a norm in the field of pharmacology, and scientific temper, and not the physician's reputation became the

primary determinant of the clinical outcome [2]. Infectious diseases were conquered, albeit with collateral damage in form of antibiotic resistance, and modern medicine seemed to rest on its laurels, till the hydra-headed monster called cancer crept in as people started living longer, and began to collect deleterious genetic changes, often purchasing them over the counter [3].

CLINICAL TRIALS

Starting with the 1980s, the availability of rich and vast medical literature documenting the results of multitudes of population-based trials of therapeutic interventions heralded the advent of Evidence Based Medicine (EBM) [4]. All treatment protocols were updated and underwent periodic revision as more peer-reviewed information poured in, reassuring physicians that they were doing right by their patients. The practice was laborious for the doctors where an average doctor would have to peruse a whopping 6000 articles per day to keep himself/herself abreast of all the recent advances [5]. However, this labor would sometimes prove to be infructuous, in case the patient corresponded to the one in the outlier group of the trial, responding either minimally to the treatment protocol, or reacting abnormally to it, with hazardous outcomes in both cases, proving that the ominous words of the stalwarts, quoted above, from centuries ago continue to ring true [6].

The outliers in clinical trials by virtue of constituting an infinitesimal fraction of the overall population under study have not been served well by the concept and practice of Evidence Based Medicine [7]. They respond differently to the established conventions because they happen to be different. However, these differences could not be ascertained easily except for a few established tests that were caried out on clinical samples before administration of certain drugs, e.g., testing a patient suffering from malaria for glucose-6-phosphatase enzyme deficiency before the administration of primaquine [8]. This is probably one of the first examples of personalized precision medicine (PPM), even before the acronym established itself in the last decade or so.

GENOMICS

The study of the human genome and proteome, consequent to the standardization of modern analytical techniques to delineate the genome and refinement of microarrays to analyze the transcriptome and the proteome has enabled an information explosion in modern biology [9]. Single nucleotide polymorphisms have been discovered to be the hallmarks of a plethora of diseases, and their timely detection with parental counseling has resulted in considerable savings to affected families and society [10]. Biomarkers and drugs have been developed to address specific diseases based on genetic information. Risk stratification for multifactorial diseases, including childhood malignancies using biomarkers and facilitated targeted therapy with specific therapeutic agents [11–13]. An entirely new field of pharmacogenomics has come to the fore that considers the genetic variations among individuals to customize the treatment guidelines as if tailor-made for the patient [14] and the flow is depicted in Figure 1.

Figure 1. Bioinformatics analysis of genomic data favors personalized clinical care.

These developments have lagged just a few steps behind the exponential growth in the computational ability to process data, structured, unstructured, as well as the seemingly overwhelming entity called the big data, the quantum of which only increases as wearable devices – like exercise monitoring watches, biosensors, and their data recorders – have become a part of routine lifestyle among the rich and affluent [15–17]. By definition, big data is characterized by its high volume, variability, generated at a rapid velocity, and holds enormous value, but its veracity must also be verified [18]. Combined with researchers' expertise, these information concealed within may prove to be instrumental in transforming the approach to patient care by means of predictive modeling built on the solid foundation of mechanistic understanding of disease [19], as shown in Figure 2. Molecular biomarkers for several

malignancies are already included in management protocols, and many are under investigation [20]. Addressing the inter-patient and intra-tumoral heterogeneity would ideally be the next step [21].

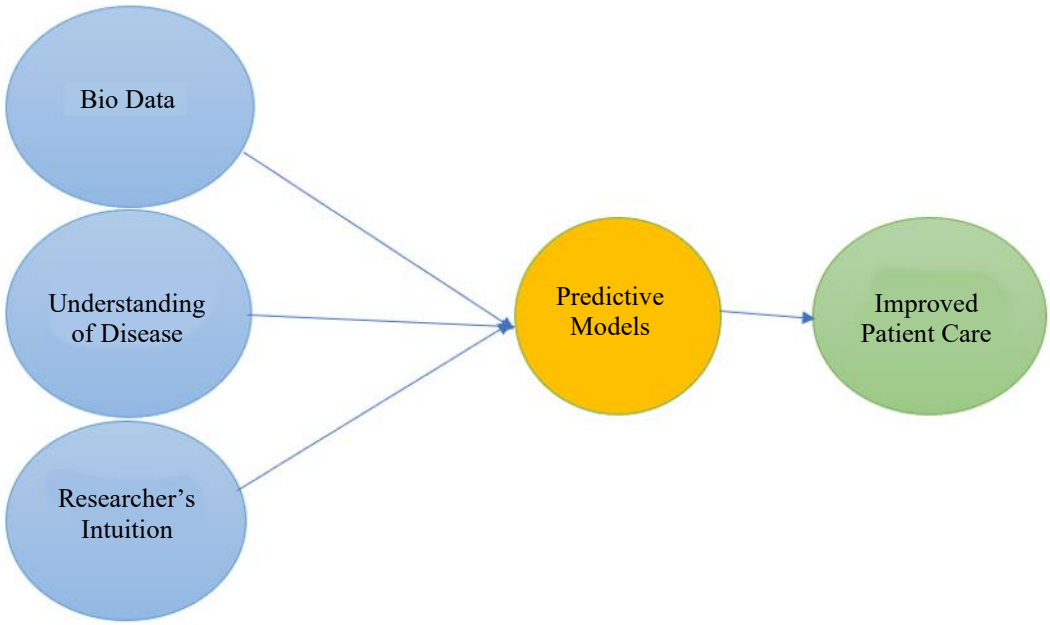

Figure 2. Building predictive models and its application.

Health Record

Consequent to these salutary developments, and improvement in the understanding of the underlying causes and pathogenesis of diseases, there is a strong conviction in scientific community that this information can be utilized to augment the Electronic Health Records (EHR) of individual patients, and arrive at accurate, precise, and hopefully infallible decisions in treatment strategy [22].

Limitations

The above lofty ideas are limited by the current difficulties in processing the huge volume of big data, because of unavailability of suitable analytical tools, lack of trained personnel, and the tendency of medical and bioinformatics personnel to restrict themselves to their respective silos [23]. The lack of clarity regarding the ownership of data further limits the formulation of a practical implementation strategy [24]. While Electronic Health Records are owned by the patient, and held in trust by the hospital, however, the clarity on ownership of genomic and proteomic data is yet to be delineated by several countries. Most of the annotated, generic DNA and protein sequence data are in public domain, and so are the various analytical tools making it easy for researchers to conduct their studies on them. Nevertheless, the critical issue of confidentiality of these data needs to be looked into, without compromising the safety of the patient, as well as the quality of ongoing research [25].

AVAILABLE RESOURCES AND THE NEED FOR PREDICTIVE MODELS

Necessity being the mother of invention, there have been large national and international collaborations that bring anonymized patient's genomic and proteomic data to common platforms to make it easily accessible to researchers aim to collate, annotate and ultimately integrate it into meaningful information [26]. Platforms – like All of Us, Genome Asia 100K, and 100000 Genomes Project – are some such examples. These have led to the evolution of a distinct branch of Bioinformatics, called Precision Bioinformatics that relies on the inputs from translational studies with the sole objective towards development of dedicated computational infrastructure, analytical tools, and methodologies required for clinical studies for adoption of PPM [27]. These would include dedicated databases, software tools, training materials, cloud storage, and supercomputing infrastructures. The net result expected is the introduction of predictive models for PPM to predict therapy response, clinical risk predictions and

refinement of in silico drug prescription tools. Successful models would rely upon AI-based tools to obtain useful information from complex data, e.g., AI predictive models using medical imaging to provide a more accurate way to predict disease progression. These tools are expected to grow exponentially while increasing accessibility to quality data and improving compatibility with robust technology platforms [28].

Need for Clinical Bioinformaticians

Bioinformatics, the field that applies the knowledge of computational tools to biological information obtained from DNA and protein, is in nascent stage, and realization of its full potential will involve inclusion of all stakeholders from the scientific community as well as the society and require coordinated efforts and investment in setting up bioinformatics infrastructure, technologies and training centers, all backed by legislation [29, 30]. Resources are being prepared among the academia, research institutions, and biotech companies before they are seamlessly aligned with the health systems. Well trained human resource, with working knowledge of computer science, statistics, and understanding of molecular biology and cell systems would be the most valuable asset to transform the clinical practice from EBM to PPM [31]. This group of professionals would represent the first generation of Clinical Bioinformaticians equipped with the ability to perform data analysis on heterogeneous biomedical data sources, to generate intuitive reports and communicate clinically relevant findings to the treating physicians and guide them in informed clinical decision-making [27].

FUTURE DIRECTIONS AND IMPLEMENTATION FRAMEWORK

The translation of bioinformatics-driven practice into routine clinical practice will not only be dependent on the maturity level of the underlying technologies but also on the systematic framework developed for their implementation. One of the priorities in this context will be to develop standardized data formats and infrastructures that will facilitate seamless integration among different data types, including genomic data, proteomic data, metabolomic data, imaging data, and clinical data. Without interoperability, it is likely that good quality data will still remain fragmented and ineffective. There is an urgent need for international standards bodies and health informatics associations to collaborate toward the development of standardized schemas that can facilitate cross-institutional analytics while ensuring strict privacy regulations are followed.

Another major area for bioinformatics research in the future is going to be the validation and benchmarking of predictive models, which are constructed from multi-omics and clinical data. This is going to be very important before their integration into clinical decision support systems. Prospective validation studies, adaptive clinical trials, and real-world performance monitoring will need to become an integral part of the development process for AI-driven medical tools. Moreover, predictive tools will need to be designed with the capability to augment, not replace, the judgment of skilled clinicians.

Capacity building is an equally important pillar in this case. With the advent of clinical bioinformatics, there is a need to develop a hybrid workforce that understands molecular biology, statistics, data science, and clinical practice. This calls for academic institutions to promote interdisciplinary curricula and training modules that train professionals to work at the interface of computation and medicine. There is also a need to develop continuous professional training modules to train practicing clinicians on how to interpret genomic data and risk predictions in an appropriate manner.

In addition to this, the ethical and legal issues also need to be updated in accordance with the technological advancements. There are many ethical issues, like data ownership, data storage, renewal of consent, secondary data usage, and bias in the algorithm, which need to be addressed through policy guidelines. With the help of federated learning, it is possible to perform training without the need to centralize data storage, thus reducing the risks while still retaining the analytical power.

In the future, the combination of high-resolution molecular profiling, high-performance computing, and intelligent analytics is likely to revolutionize the classification, targeting, and prevention of diseases. With the development of predictive models, the focus of healthcare is likely to change from

treating diseases to preventive precision medicine. This will depend on strategic investments, collaboration, and support.

CONCLUSION

To summarize, medical science is on the cusp of another revolution, consequent to our improved understanding of the molecular mechanisms underlying a disease, digitization of individual health records, explosion in the volume of available data, and technological breakthroughs in our ability to process, integrate and analyze these data using bioinformatic tools to enrich the novel field of personalized precision medicine. Investment in infrastructure, technology and training shall enable mankind to get there at a pace commensurate with the background developments.

Conflict of Interest

The authors report there are no competing interests to declare.

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