Short Communication

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Precision Medicine in Healthcare: A Personalized Healthcare Framework that may have Wider Public Health Utility

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ABSTRACT

Abbreviations: SDH: Social Determinants of Health; NGS: Next-Generation Sequencing; SDH: Social Determinants of Health; AI: Artificial Intelligence; ADR: Adverse Drug Reaction; CVD: Cardiovascular Diseases

Introduction

Precision medicine is the use of a collection of large scale data like biomedical data (including genomics, transcriptomics, proteomics, metabolomics, lipidomics, epigenetic modulation, nutrigenomics and microbiomes studies), as well as data on the patient's environmental, travel, occupational, medical, lifestyle and family history, to provide a personalized context for the prevention and treatment strategies for individual patients [1]. The use of combined factors of the patient's environment and socioeconomic factors known as Social Determinants of Health (SDH), has been poised to have the potential to revolutionize healthcare delivery in an era where holistic, individual care has become more standardized. The overall aim of precision medicine is to allow healthcare practitioners and researchers to predict more accurately which treatment and prevention strategies would work best for a particular patient with a particular spectrum of signs and symptoms [2]. The conceptual framework of precision medicine, its technological advancements, and its potential to revolutionize healthcare delivery in an era where healthcare is rooted in socioeconomic inequity and inequality, especially in Sub-Saharan African Countries, possesses much potential.

Investing in precision medicine has the potential to not only optimize patient outcomes but extending long, happy patient life expectancy through value-based quality of care. Precision medical care, thus aims to efficiently assess or predict patients' risk profiles early, and to benefit the healthcare sector too. It could potentially result in high pay-off results by improving workflows, just in time guidelines achieve best practice standards, by enhancing operational indicators such as: decreasing patient length of stay, lowering complication rates, decreasing re-admissions and rehabilitation rates, lessening the need for step-down facilities and lowering the cost of patient per day equivalent, as well as enabling overall cost-effectiveness by decreasing mortality and early mortality rates [3]. In the last ten years, the advancements in Next-Generation Sequencing (NGS) technologies and target enrichment methods advancements in NGS technologies have resulted in the identification of genes responsible for more than 40 rare disorders, and currently has been reported that at least 10% of human diseases have genetic causes with the remaining 90% of diseases are linked to genomes (even as far back as the embryonic stage) and influenced by Social Determinants of Health (SDH) [4-6].

The predictive ability of precision medicine could allow more accurately prospective prevention and treatment strategies to deliver "the right treatment at the right time to the right person", rather than operating as reactive medical care. Since the mapping of the human genome in 2003, enormous progress has been made in understanding molecular and genetic pathways, underpinning human health and disease. With precision medicine, this genomic information has been poised to be able to unveil disease predisposition and onset in individuals much earlier and more accurately, by targeting disease-specific molecules and biological pathways [7]. Some of the disease areas that has gained huge strides with the use of precision medicine and has gathered a lot of attention, includes:

(A) The Epilepsies: single gene variant discovery in this area, has pushed new ideas through work on several gene identification, in order to assist more patients in predicting risk of disease earlier, which was not available before, is showing great potential [8].

(B) Oncology Research: here the availability of high dimensionality datasets coupled with advances in high performance computing, together with innovative deep learning architectures in oncology research, has become central to individualized treatment gains. Analyses on cancer tissues have identified germline and somatic mutations of several classical tumour suppressor genes, spurring on the opportunity to halt the regeneration of certain cancer cells [9].

(C) Sepsis Management: during sepsis, a dysregulated host immune response occurs as a result of the infection. Immunological biomarkers currently play an important role in facilitating predictive enrichment in treatment scenarios. Through the use of precision medicine, multiple immune and non-immune related parameters could potentially be integrated into diagnostic and predictive combitypes with the aid of machine learning and Artificial Intelligence (AI). This has tremendous potential to impact the health outcomes of critically ill patients suffering from sepsis [10].

(D) Diabetes mellitus: diabetes prevention, classification and management, through precision medicine, has increased our understanding especially in the genomic architecture of diabetes, and its complications [11]. It is this understanding of the genetic basis of diabetes that has the potential to enable healthcare workers to prevent or even reverse diabetes complications due to the utility of genomic medicine, as well as using this genetic information to guide management of monogenic forms of diabetes, which represents the best-known examples, of genomic medicine for diabetes. The biggest strides made in genetic research for diabetes, is in identifying its complications and pharmacogenetics, to assist in risk prediction models for prioritization of treatment strategies, as well as the use of multiomic analyses to discover novel drug targets with companion diagnostics [11].

(E) Cardiovascular Diseases (CVD): CVD's are complex and heterogenous in nature, as they are caused by multiple genetic, environmental (e.g. air pollution) and lifestyle factors like diet, exercise and gut biome. Traditionally risk score algorithms have been used to predict myocardial infarction outcomes in the possible future. Precision medicine and several deep learning and AI technology are under investigation to evaluate optimizing and overcoming the challenges of risk prediction of myocardial infarction and management of CVD's [12].

(F) Myeloid leukaemia: currently sufferers of myeloid leukaemia benefit by drugs prescribed to a sub-population, with, e.g., Gefitinib for patients who carry activating EGFR mutations, and certain drug prescriptions are avoided for those who are likely to develop a serious Adverse Drug Reaction (ADR), e.g., Azathioprine, an immunosuppressive medication, that can cause severe neutropenia in those patients who have mutated TPMT_12. It is for this reason that extended efforts currently put in place in a country like Qatar, to develop a policy for the implementation of precision medicine at a greater level within the subcontinent [13]. It would be remiss not to comment on the specific use of AI techniques in precision medicine. Some benefits of AI has been applied in cardiovascular medicine to explore novel genotypes and phenotypes in existing diseases, to improve the quality of patient care [14-16]. The field of AI in healthcare is on the cusp of significantly transforming and even disrupting the system. AI is certainly rapidly reshaping areas like e-health record keeping and sharing; improving the cloud infrastructure; providing streamlined workflow plans; simplifying and increasing the rate and sharing of records, decreasing duplication; standardizing patient care; decreasing blind decision-making and creating smarter logistics, funds, drug and equipment management.

It is thus easy to see why more stricter policy recommendations could benefit healthcare management as well as groups of patients presenting from similar symptoms and similar backgrounds, improving their health and smoothing out the process of patient management. Therefore, paradoxically, precision medicine designed as a "non-one-size-fits all" system to treat individuals, could be compiled in a way to treat patients at a population level and thus have major implications for public health [17,18]. This process of using precision medicine and including data related to a patient's SDH compiled to treat groups of patients, has been coined as precision public health. Unfortunately, although precision medicine is so unique and filled with huge possibilities, it does contain numerous pitfalls, challenges and obstacles, which has prevented the transition of its practice from the laboratories and literature pages to the patient's bedside.

These include: Absence of supporting information technology and infrastructure, lack of data standards and interoperability of systems that are currently used in silos, insufficient supporting technology and appropriate funding, lack of corresponding appropriate phar-

maceuticals, lack of user trust, poor governance policies that protects the patients and their data, leading to unfortunate ethical issues [4]. Never the less, the future of the use of precision medicine is bright.

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