Artificial Intelligence for Precision Medicine and Better Healthcare

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Abstract

Precision medicine is an important and growing area of research, development and healthcare for the diagnosis of diseases and patients precare. It involves analysis of a patient's personal data, genetic information, circumstances to diagnose and cure the disease. It allows researchers to design and develop the medication for prevention of specific viruses. It has the potential to improvise the traditional symptom driven retrospective practice of medicine, by allowing earlier interventions with advanced diagnostics, which can further be used for tailoring personalized treatments. Identification of the pathway for developing a personalized medicine involves analyzing comprehensive patient information along with broader aspects to monitor and distinguish between healthy and sick people, which will lead to a better understanding of biological indicators that can signal shifts in health. In order to positively impact the patient's health and to provide real time decision support, it is vital to leverage the power of electronic health records by integrating disparate data sources and discovering patient-specific patterns of disease progression. The goal of which is to use multiple types of data and classify patients into precise groups that will benefit from a given treatment approach. In this article, we will focus on various machine learning, deep learning models, and applications of AI which can pave the way for a new data-centric era of discovery in healthcare. In this article, we will focus on various machine learning, deep learning models, and applications of AI which can pave the way for a new data-centric era of discovery in healthcare.

Keywords: Precision medicine; Patients precare; Population medicine; Comprehensive patient information; Diagnostics.

Introduction

Precision medicine is a medical model, which proposes customization of the healthcare to a subgroup of patients, based on a genetics, lifestyle and environment. This technique allows doctors and researchers to prognosis treatment and prevention strategies for a specific disease which can work on a group of people. It is opposed to a one-size-fits-all approach, in which disease treatment and prevention techniques are advanced for the average individual with much less attention for the variations among individuals.

There is an overlap between the terms "precision medication" and "personalized medicine." As per the National Research Council (United States), "personalized medicine" is a traditional word with a meaning close to "precision medication." However the word "personalized" may be misinterpreted to suggest that treatments and preventions are being evolved uniquely for every person. In precision medicine, the focus relies on figuring out the methods which can be effective for group of patients.

Precision medicine approach leverages a patient's genetic history, location, environmental factors, lifestyle and habits to determine a plan of action for treatment. Artificial Intelligence has been successfully able to classify problems using different algorithms and solve precision medicine problems e.g. accurate disease diagnosis, disease detection and prediction, treatment optimization. The analysis of multidimensional datasets to capture variations can be learnt (trained) by using AI algorithms and identify cryptic phenotypic organotypic structures. It can further be used to predict the risk of a disease, identification of the disease response and outcomes on the individual patients based on their own characteristics. Recently, prediction algorithms utilizing artificial intelligence approaches for cancer and cardiovascular disease have shown promising results, predicting disease risk with a higher degree of precision [1]. Artificial Intelligence in healthcare has the potential to achieve the goals of providing real-time, better personalized and population medicine at lower costs [2]. The article reviews applications of Artificial Intelligence and various algorithms which can help in better healthcare for humankind.

Traditional Medicine versus Precision Medicine

In traditional medicine, as shown in Figure 1, the doctor uses their expertise and trial and based method. Based on assumptions from the symptoms given by patients, doctor suggests same medicine with equal dosage. This type of treatment may not work every time. Techniques that benefit any victims are weak for others and the corresponding injection may additionally produce floor consequences in only some cases [3].

Precision medicine is the orienting of clinical strategy to the particular characteristics of the individual patient. The technique depends on correct findings in our understanding of how a patient's unique outline gives them sensitive to some situations. The corresponding analysis is enhancing our information to divine which treatments be reliable and valid for any case. As shown in Figure 2, the doctor suggests medicine and dosage based on person DNA and personal health information [3].

Precision medicine approach can be considered as an extension of traditional approaches to treat the disease with greater precision. A profile of a patient's gene variations can guide the choice of drugs or remedy protocols that reduce side effects or ensure greater success outcomes.

Role of Artificial Intelligence in Precision Medicine

Artificial intelligence (AI) has been used for years in the field of healthcare and continue to grow tremendously each year with its ability to advance medicine and research. Even precision medicine is not completely possible without the addition of machine learning algorithms to assist in the process. Machine Learning (ML) is an application of artificial intelligence (AI) that can learn and upgrade from experiences and without being explicitly coded by programmer. ML specializes in the development of computer programs which can retrieve data and use it to learn for themselves. Most commonly used ML algorithms in medicine includes SVM, deep learning, logistic regression, DA, decision tree, random forest, linear regression, Naïve Bayes, K-nearest neighbor (KNN) and hidden Markov model (HMM) [2,4,5].

Figure 3 shows that ML algorithms are applied for clinical, genomics, metabolomics, imaging, claims, labs, nutrients and life style data fusion, integration and analysis. So, ML algorithms integrates multiple data sources to produce more consistent, accurate, and useful information than that provided by any individual data source.

Over the last few years, AI approaches has been used in neurodevelopmental disorders specifically in autism spectrum disorder, epileptic encephalopathy, intellectual disability, attention deficit hyperactivity disorder (ADHD) and rare genetic disorders [1]. AI algorithms can create an impact in 4 complex unresolved problems in neurodevelopmental disorders as shown in Figure 4.

Identifying causal genes

AI methods are crucial for identifying causal genes and locus. Bioinformatics prediction still not able to exactly classify the more common missense mutations as per pathogenicity. Even identifying causal genes from those 'variations of uncertain importance' (VUS) stays a major unresolved hassle that does lend itself to an AI solution. AI models have recently shown reasonable success for improving genetic diagnostics in Neurodevelopmental Disorders (NDDs). Two AI algorithms named Human Splicing Code, and

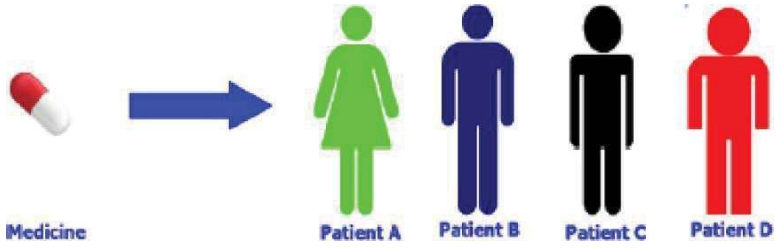

Figure 1: Traditional Medicine [3].

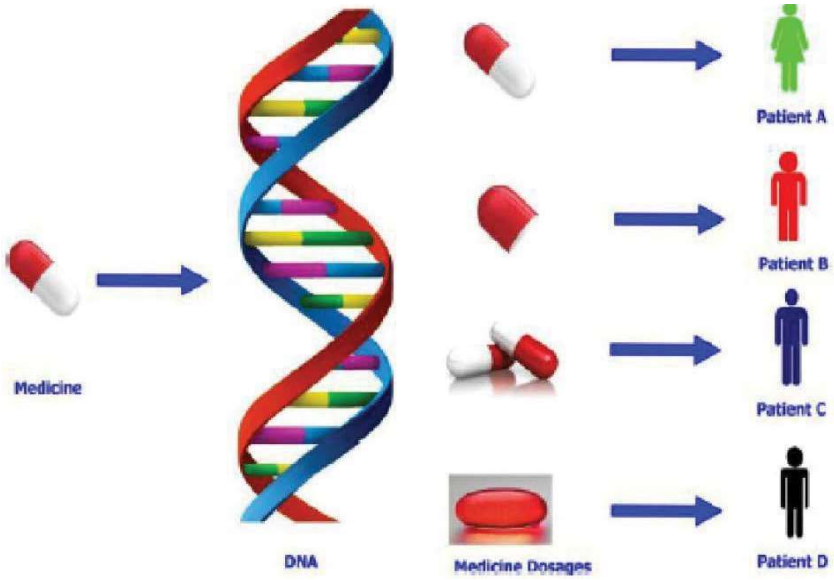

Figure 2: Precision Medicine [3].

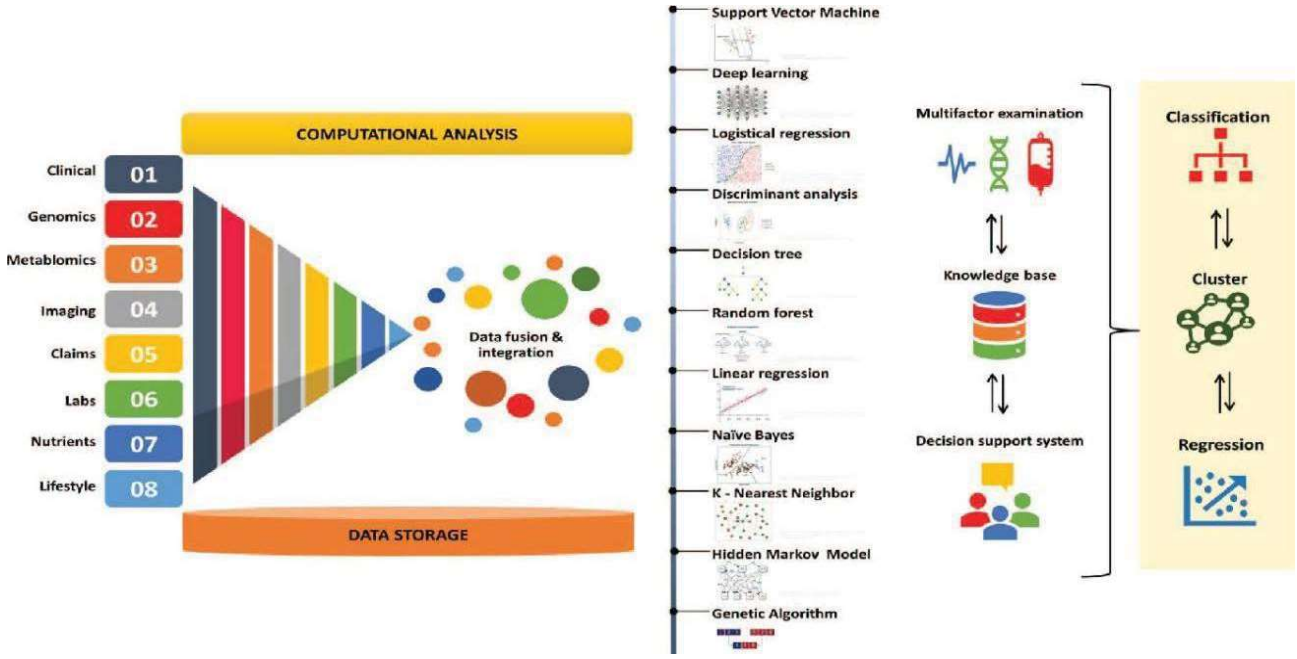

Figure 3: Role of ML Algorithms in healthcare [2].

DeepSEA showed very promising results in the challenging task of correct classification of missense variants.

Phenotypic and genetic heterogeneity

Despite the fact that NDDs are ordinarily genetic in etiology, environment will nonetheless effect on genetically driven brain patterning, and consequently have the capacity to influence disease severity. Multiple independent reports have shown an association among post zygotic mosaic mutations and autism spectrum disorders, intellectual disability, epilepsy and other NDDs. In the last few decades, digitization of medical health record added a large amount of data related to healthcare. The application of AI algorithms might be significantly benefitted from those digitization efforts that can help establish genotype phenotype relation for genetic diseases and have the capacity to conclude numerous phenotypic correlations and associations.

Polygenic risk score and gene-gene interactions

Gene-Gene interaction is a major contributor to the phenotypic variance of NDDs but there is currently no credible AI algorithm able to cope with data on this scale. There exist major complexities concerning deep phenotypic and large scale omics data. Unsupervised AI approaches may be applied

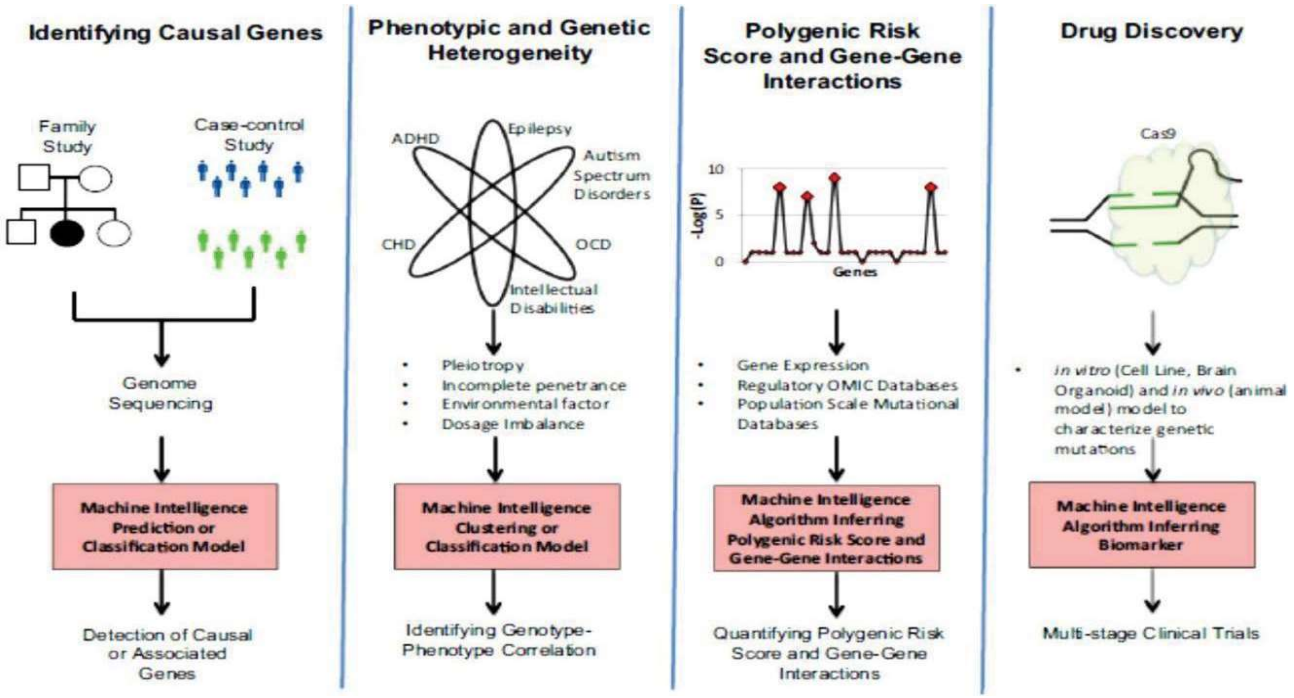

Figure 4: Complex Unresolved Problems in Neurodevelopmental Disorders [1].

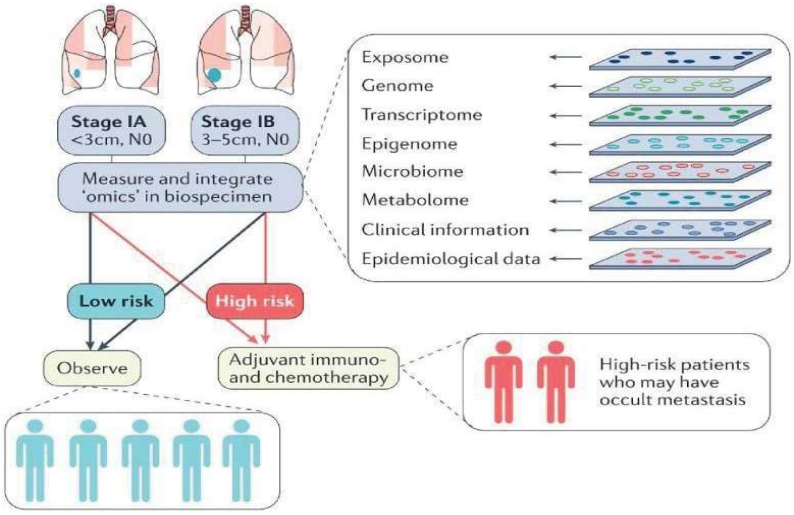

Figure 5: Classification of Early Stage (IA and IB) Lung Cancers by Biomarkers [6].

to perceive previously unknown sub-structures within NDD cases based on environmental factors, dosage balance etc. Even though none of the techniques have been carried out in a quantitative context. So the future work can be done in quantifying polygenetic score and gene – gene interactions using AI/ML algorithms.

Drug discovery

AI models are at the frontier for therapeutic intervention and drug design. Currently, there are 51 food and drug administration (FDA) approved targeted gene unique drugs for neurology and psychiatric situations. The advent of sequencing technology has basically been targeted on facilitating the implementation of early precision diagnostics. Recently the appearance of genome modifying technologies (i.e., CRISPR/cas9), and antisense oligonucleotide remedy has allowed scientists to mimic cellular phenotype, and help become aware of precise molecular objectives. Such drug design would require a primary push on AI algorithm implementation. Recently the idea of repurposing drug is turning into a prime area of research using AI algorithms.

Applications of artificial intelligence in precision medicine

An example is biomarker development in precision medicine for early-stage lung cancer. Biomarkers are characteristics of the body that we can measure. e.g. blood pressure or heart rate can be considered as a biomarker. Biomarkers are integral to drug development, because we need to measure the effects of investigational drugs on people during the clinical trials. It has shown use of precision medicine (biomarkers) to classify patients with early-stage lung cancer into subclasses to provide appropriate treatment. Figure 5 classifies early stage (IA and IB) lung cancers by biomarkers that predicts risk of recurrence generated using a precision medicine research strategy into low risk for recurrence and high risk for recurrence [6].

Conclusion

Artificial Intelligence takes precision medicine to the next level and increases the accuracy and prediction of outcome for patients. It can also make treatments more affordable and accessible to those who may not be able to receive those treatments due to cost and health insurance at this time. There are many challenges ahead for precision medicine to be perfect, but artificial intelligence can help drive us closer to those goals.

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