DATA SCIENCE IN BIOLOGY: PERSONALIZED MEDICINE

The key idea of personalized medicine is to base medicinal decisions on individual patients’ characteristics rather than on the population average. Personalized medicine, also known as precision medicine, is using genetics or other biomarker information to make decisions about patients. Personalized medicine is a medical approach in which patients are classified based on their disease subtype, risk prognosis, or treatment response using a specialized diagnostic test. The key idea is to make medical decisions based on individual patient characteristics, including biomarkers, rather than on averages over the whole population. Biomarkers are termed for any measurable quantity or score which can be used as a basis to classify patients (e.g., molecular markers, genomic alterations, disease severity scores, lifestyle characteristics, etc). The main advantages of personalized medicine could be better medication effectiveness, reduction of adverse events, early disease diagnosis and prevention.

       Personalized medicine is closely related to data science and specifically machine learning. At present personalized medicine is an emerging reality. To date, FDA (US Food and Drug Administration) has listed more than 160 biomarkers and biomarker signatures that have been approved for classifying patients for drug response. For example, the anti-cancer drug trastuzumab can only be administered if HER2/neu receptor is overexpressed because the drug interferes with this receptor. In many cases, it is impossible to identify a single biomarker for the patient population because many diseases are complex and affect a multitude of the biological subsystem. Identifying biomarkers is difficult and requires advanced approaches offered by data science. Specifically, multivariate classification algorithms using techniques from the area of machine learning play an increasingly important role. Machine learning can be unsupervised or supervised. In supervised learning, we train a model based on a dataset with many observations, each containing a large number of features, coupled with known clinical outcomes (e.g. drug response). Based on the established model predictions for patients that were not part of training data can be made. Machine learning models can make accurate predictions even if there is no single biomarker that discriminates patient groups i.e. we can combine the outcome of more than one feature that together produce a stratifying biomarker. Unsupervised learning aims at inferring patterns from data without having access to a label.

One of the most prominent examples is MammaPrint, a prognostic test for breast cancer based on 70 gene-signature, which was approved in 2007 by FDA. MammaPrint produces a score from the weighted average of 70 measured genes, which is predictive of the development of distance metaset.

Another example is *Geno2pheno a machine learning based toolbox used*to estimate the resistance of HIV to an individual drug.

Personalized medicine is tightly connected with genomics. However, genomics and other biological high throughput data isn’t the handiest supply of records hired withinside the personalized medicine field. Other applicable records encompass records from wearable sensors and cell fitness applications, bio-images (e.g., MRT and CT scans), fitness claims records from coverage companies, and electronic medical records (EMRs).

Many methodological articles focus on yes/no decision tasks e.g. disease progression / no disease progression or clinical trial endpoint met / not met. Models with binary outcomes can be appropriate in certain situations but in most cases, a more comprehensive outcome is more helpful. For example, DAS28 disease score is a commonly used response criterion for rheumatoid arthritis. Which ranges on a continuous scale from 0 to 10 and is often discretised on three levels that are low, medium, or high disease activity.

While in recent times there has been a lot of enthusiasm about data science and machine learning but there are only a few examples that affect the current medical practices. The lack of impact on the clinical practices can be attributed to the insufficient performance of the predictive models, difficulties in implementing complex models, lack of validation via prospective clinical trials that demonstrate the clear benefit compared to standard care. In addition, general concerns regarding data privacy, as well as ethical and legal factors, cannot be overlooked.

In parallel computational algorithms must advance to provide direct benefit to clinical practices. Current machine learning models are far from being able to recommend the right treatment at the right time and dose for each patient. The following steps could bring us closer to the goal 1) innovative software tools that better link knowledge with machine learning based predictions from multi-scale, multi-model and longitudinal data, 2) innovative modelling approaches which go beyond typical state-of-the-art machine learning, 3) new computational modelling approaches that allow us to identify critical transitions in a patient’s medical trajectory.

Overall, the ambition of research towards personalized medicine should be to move from a system analysis perspective to a system control view that allows for the planning of optimal medical interventions at the right time and dose on an individual basis. New computational modelling approaches that go beyond the current machine learning methodology may play an increasing role for that purpose. It must be noted that no algorithm is meant to replace a physician, rather the idea is to provide them with a tool at the hand, which support their decisions based on objective, data-driven criteria and the wealth of available biomedical knowledge.

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