The Promising Vision of Precision Medicine to Soon Revolutionize the Global Healthcare System

By Kacper Ścibski

While researchers and clinicians focus on understanding the reasons behind every disease and their possible treatments, we shall remember that just as every patient is individual, so are the symptoms, disease reasons and prevention methods unique for everybody. Have you ever had acne during your puberty? Do you remember your friend recommending you an amazing balsam that helped them? It did not do the trick with you, did it? This trivial example demonstrates that the *one fits all* model does not always apply.

This is why work on new Precision Medicine (PM) has just began in the recent years. PM, about which this article is about, uses a method of patient subcategorization, allowing to compare the individual medical conditions to other patients. Such approach narrows down the required treatment to a specific group of patients, rather than applying one, often ineffective method for all.[1]

A New Compromise? How Does the Precision Medicine differ from the Personalized Medicine?

Since the field of medicine emerged, it has always relied on observation and analysis of various health conditions. So far, all the medical knowledge has been scattered in forms of research papers, publications, articles, books, etc. From a very long time ago, people have always realized that, for example, walking around without a hat and a warm coat in the winter can lead to flu. However, over the years, this knowledge, based on observations of thousands of patients, had been accumulating to be used by the next generations of clinicians. Nowadays, (in addition to the continuous new research) humanity has already obtained a wide scientific understanding of diseases treatment and preventing many of them.

In the last few decades, there has been a great movement towards *Personalized* Medicine. As the name suggests, every clinical case here is investigated

individually. However, in real life, this approach is often found to be limited by the tremendous amount of testing (usually based on patient's genetics which is quite expensive, to say the least). If we add the time-management and lack of resources that could allow for a proper medical diagnosis, *Personalized* Medicine becomes rather a futuristic concept and Precision Medicine is a step towards it.[2]

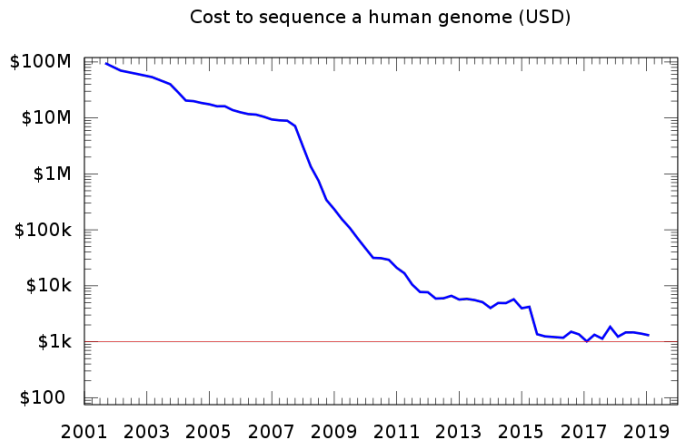
Healthcare providers do realize a similar problem, which is why a compromise has been suggested. While Precision Medicine is a relatively new approach in global healthcare system, for which the first step would be a development of a database, including disease classification, affected by lifestyle, medical conditions and susceptibility to diseases, followed by a more effective treatment. The development of such a medical data analysis tool could significantly reduce the waiting times to see the doctors, help to choose the individualized set of drugs, or even predict and prevent multiple diseases.

The movement towards Precision Medicine has been accelerated in 2015, when President of the United States, Barack Obama, decided to fund 200 million USD to the "Precision Medicine Initiative." After just a year, over 10,000 American specialists were already involved in the project.[7]

Examples of Already Existent and Potential Uses of Precision Medicine

The most common example of practice of Precision Medicine is blood group classification. In the early XX century, an Austrian Physician Karl Landsteiner noticed the blood serum of certain individuals would agglutinate when mixed together. Yet, he later discovered this was not always a rule and blood agglutination was dependent on the antigens present on the surface of the red blood cells. For categorizing humans into three groups (later named as A, B and O) his work was awarded with a Nobel Prize.[8]

Another huge milestone in PM was a huge development in the genome sequencing. When "The Human Genome" Project launched in 1990, it was estimated to last for 15 years and cost almost 3 billion USD. High cost and time necessary for this project pushed the scientists to start manufacturing other sequencing methods.



The initial price went down to millions, then thousands, ending up in a "race" of reaching a baseline of 1000 USD. Nowadays, the term 'Next-Generation Sequencing' is used to collectively refer to the most recent technologies. Those are not only becoming cheaper, but also more time-efficient while reaching a great accuracy.

Ability of sequencing the human genome has already been proved to have a massive potential in Precision Medicine. In 2017, "Nature" published an article claiming the Next-Generation Sequencing might allow for an effective cancer treatment. An example of leukemia was given, where after recognizing the mutation site, the abnormality could be targeted with a specific drug. For example, with Gleevec - a medicine being effective only in a treatment of particular genetic mutations, yet, passive in others. Clinical tests have already proven for this method to be efficient in curing cancer among patients - most astonishingly, with a use of a single drug only, essentially excluding the need for chemotherapy.[9-10]

In addition, NGS can revolutionize oncology by providing a fresh look on tumor formation. So far, the clinicians were mainly focusing on the location where cancer begins to develop. Surprisingly, according to other recent findings, "The genetics of some breast cancers, for instance, may be more like stomach tumors than other breast cancers."[9] This new approach is often referred to as a 'basket trial' - which characterizes cancer based on its genetic background and not only the location in the body. Specifically for the tumor, which originates from a random mutation, the conclusions can be made that sequencing the genomes of multiple patients and collecting them as a one database, would allow for a better understanding of cancer.

Yet, cheap and accessible genome sequencing may be the most important factor to enable Precision Medicine to develop, another important aspect, on which it

is heavily reliant, is the lifestyle. This broad term includes the diet, amount of physical activity or psychological health. PM is especially promising in the fight with diabetes - an arising problem in the developed countries. However, it is being observed that the increased tendency of obese people to develop diabetes at some point in their lifes, is not always a rule. Researchers are currently working on relating the different biomarkers with Type II Diabetes susceptibility. PM, if applied in the future, along with Artificial Intelligence, could categorize the patients and "use biomarkers to personalize lifestyle recommendations, intended to lower type 2 diabetes risk or to slow its progression."[12]

Another target of the modern Precision Medicine are the commonly-known "diseases of affluence." Those afflictions include conditions like heart-stroke, type II diabetes or obesity.[3] As long as everybody knows lack of sport activity or diet based on the junk food often leads to various complications, predicting the disease susceptibility might often rely on genetic factors too. The easiest way to determine that would be by comparison with data other patients' data. Such classification would be based on individuals' health histories, genetic susceptibilities or even the environmental factors they are exposed to, in order to decide on the best actions strategies for the patients[4]

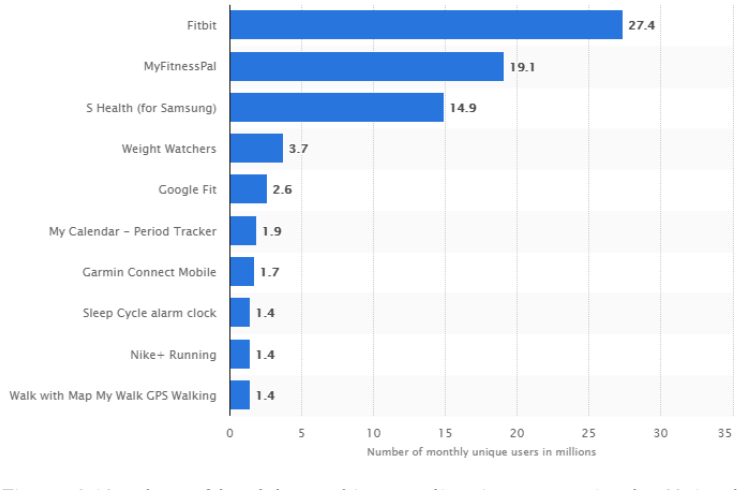
Another medical condition with a potential to be solved by Precision Medicine application is asthma. This disease includes symptoms like difficulty to breathe, wheeze and cough - all due to inflamed airways producing mucus. Asthma can be caused by number of factors, including allergic response to pollen and dust, too intensive physical activity, or respiratory infections. Because of that, the broad term 'asthma' is used to describe a group of diseases. Yet, caused by different medical conditions, the diagnosis is usually exclusively based on symptoms assessment rather than application of personalized medicine. The most common - Inhaled corticosteroid (ICS) therapy is prescribed for almost all the patients. However, according to the recent findings, therapeutic responses "to ICS are seen only in […] 50% of all asthmatics.[11] Remaining 50% is misdiagnosed. In the future, it is believed biomarkers analysis might soon enable a better treatment of asthma. However, because such a

strongly personalized approach goes along with a high cost, Precision Medicine might provide the clinicians with a more affordable alternative.

Need for Database and How to Create One?

As proven above, genome sequencing might soon become a pillar of the Precision Medicine. While technologies, allowing to do so for less than a thousand dollars already exist, one could expect genome sequencing to popularize among the society thanks to commercial companies. Apparently, this is already happening. A good example for that might be "23&Me," which for as little as 99 USD will analyze the sample of a person's genetic material to check for their ancestry, family tree or even the anatomical traits' origin. We can then speculate genome sequencing will very soon get popularized and become available to everybody. In addition, many researchers are focusing on making use of Artificial Intelligence to recognize common diseases. If this happens, the software will be automatically able to analyze the results from various patients, which is why creation of a dataset is the first necessity.

At the same time, with its potential, PM would then look at the environmental factors and their influence as well. Fortunately, measuring lifestyle is easily possible thanks to mobile health-tracking applications. In the US only, over 70 million of citizens (above 18 years old) regularly use them. The three most common ones include: Fitbit, MyFitnessPal and S Health (Figure 2.). After registering, those apps can automatically record any kind of fitness activity, the heart rate, blood oxygenation, etc. Sometimes, we can even connect other portable devices to our smartphone, like a bathroom scale to record our weight, or a smart



watch to automatically measure the pulse during the training. The preinstalled Artificial Intelligence is then able to come up with tasks for us to enhance our training or sometimes even tell us off for gaining a few kilograms after Holidays… All this precious data could then become an important part of our health history and be used in Precision Medicine - to prevent certain diseases of affluence.

Unarguably, fitness-tracking applications could serve as a precious source of information for the PM. Yet, they are capable of taking multiple measurements, automatization plays a huge role here, thanks to which, apps are very user-friendly and work in the background. However, many users complain about the feature of tracking the calories and micronutrients consumed - supposedly helpful for people who are on a diet. Currently, in order to add a meal eaten, application would ask about a type of food, followed by its weight. Due to limited recipes library, and difficulty in determining the weight of the food, many people do abandon this feature. One idea to improve it could be brought by Artificial Intelligence, so less effort is needed to input the data. Looking at the most recent research, two technologies might become extraordinarily useful.

The first one, being at a very early stage of development, proposes using AI to recognize different kinds of dishes from images taken by the user. Based on food recognition and placing it into subcategories (Vegetables, Fruits, Dairy, Oils, Grains and Protein), the learning algorithm could be implied to calculate calories and microelements uptake.[16] Furthermore, AI was already proven to successfully distinguish between complex dishes, such as those of Thai cuisine, achieving a great accuracy.[16]

Another research suggests substituting the traditional barcodes with QR codes on our daily shopping products. This is because the QR code is capable of storing large amounts of data. The new feature could be then extended to contain nutrition metrics. When scanned by our diet-tracking application, data would be added to the smartphone in order to process the nutrient components.[17]

If both of those solutions were to soon become implied in the health-tracking technologies, users could gain a much better image of their health. Furthermore, AI

could be programmed to analyze this information, provide tips on possible diet alteration, or advise an increase in particular nutrients consumption.

The new feature could become a part of Precision Medicine, directly advantageous to the users by recommending balanced meals and hence - preventing many vitamin deficiency conditions. Finally, all the data collected by the health-tracking applications, along with our sequenced genome, as well as health history could then be uploaded and stored in a programmed cloud. At the same time, AI would constantly analyze our results. This method could provide the clinicians with all the medical information about us - available from any point in the world.

Summary and Conclusion

Currently, Precision Medicine stays more as a futuristic, utopian concept - still at an early stage of development. However, it might not be that distant as it sounds. Humanity is already in possession of all the required tools to be able to create it and, most importantly – has a good scientific understanding of multiple diseases and their symptoms. In addition, PM's possible applications were already well defined, highlighting its necessity. Furthermore, PM could enable new ways in the research and be a self-learning platform. As described above, the most important stage of software and dataset formation, requires an effort from clinicians, Data Analysts and Computer Science specialists. This might become a milestone in PM development - promising more efficient treatments and prevention methods in the near future.

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I am currently a Biology (Pre-Med) undergraduate at NYU Abu Dhabi. My greatest passion is low-cost travel, followed by an inborn curiosity for Science, as well as playing Baroque music on a saxophone. My two life goals are - to help my future patients as a doctor and visit all the countries in the world!

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Figure 1 The cost of human genome sequencing change over time; source: https://commons.wikimedia.org/wiki/File:Historic_cost_of_sequencing_a_human_genome.svg [Accessed: 18th March 2020]

Figure 2 Number of health-tracking applications users in the United States; source: https://www.statista.com/statistics/650748/health-fitness-app-usage-usa/ [Accessed: 14th March 2020]