My First Medical Writing

SECTION EDITOR

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Editorial

We all have been at the first stages of our careers, knowing what we want to do but without much experience. This is the case of many aspiring medical writers who come out of academia with a burning desire to become medical writers but without much knowledge of the industry and, sometimes, not confident of whether they have what it takes.

This new section serves two main purposes: to offer an opportunity for those who want to showcase their first medical writing work and, most importantly, to receive feedback on the quality of their writing and have an opportunity for improvement.

As the section editor, I'm committed to providing comprehensive feedback for all submissions and to working together with the author on improving quality while also training writing skills, dealing with feedback, and gaining experience.

We welcome all submissions from aspiring medical writers about the topic of their choice. However, we encourage authors to explore making scientific topics accessible to the general public. You can write about the topic of your research in academia, a health issue that you are passionate about, or a recently published paper.

For the debut of this section, we would like to thank Bhavana Achary, a PhD in biochemistry and molecular biology from Singapore, who is passionate about bringing science closer to the lay public.

If you are an aspiring medical writer, don't miss this opportunity of having your work read by experienced medical writers all over Europe (and ever further away, I would dare say). If you know someone who could benefit from this opportunity, spread the word… and the love!

Evguenia

Understanding precision medicine: Bringing the bench closer to the bedside

Five years ago in 2013, the actress Angelina Jolie underwent an elective double mastectomy. Her decision was based on her family history of cancer and a mutation that increases her risk of developing breast cancer by 87%.[1] Her courageous account brought the words "BRCA1 mutation" out of the doctor's office into our everyday conversation. In recent times, we have learnt how genes influence our health. These advances in our understanding of the underpinning causes of disease are resulting in more precisely targeted treatments, hence the term *precision medicine*. Precision medicine can help treat rare conditions as well as improve the currently available treatments for common diseases. There are a number of challenges in this evolving field. They include the need for a regulatory framework that protects consumer data while ensuring that the information is accessible to all. There is also a need to balance the costs of developing new lines of treatments that beneficial to all.

Precision medicine is a move away from the "one size fits all" principle, which suggests that most drugs work similarly in all individuals. We all share experiences that contradict the above-mentioned principle. Precision medicine is tailored to a specific group of individuals sharing certain characteristics such as genetic makeup, family history, and environment. While the term *precision medicine* might be new, the basic tenet of *personalising* the treatment to the individual is not. Historically as we learnt more about the differences in physiologies amongst individuals, knowledge of these differences have dictated *personalised* treatments. Examples of these are blood typing routinely performed prior to transfusions or how drug allergies influence treatment options.

In the past two decades, there has been an explosion of information regarding the genetic and molecular causes of diseases, resulting in clinicians thinking differently about these diseases starting with how they classify them.[2] Traditionally, most diseases are classified based on where in the body they originated. For example, cancers are classified as breast cancer, lung cancer, etc. However, these different cancers might share similarities in their molecular markers and genetic mutations suggesting a need for new classifications based on common root causes that can be targeted more effectively.

Cancer is a key focus area of precision medicine.[3] Currently, doctors have more knowledge about the genetic mutations and molecular markers associated with specific types of cancer. These markers can predict disease and indicate the treatment prognosis. Lung cancer treatment is hailed as an exemplary model of precision medicine. Using next-generation sequencing platforms to profile for most common mutations associated with lung cancer, the choice of drug treatments are tailored to those specific mutations. Significantly, most of these genetic variants are considered *actionable,* indicating that treatments are available targeting

these mutations.[4] The genetic profile of the cancer cells allows physicians to identify the drugs that will or won't be effective in an individual patient; thereby reducing the severity of side effects and possibly the cost of treatment. Similar progress has recently been reported in the treatment of pancreatic and breast cancer.[5,6]

Precision medicine is not limited to "matching" mutations to known drug treatments. It also helps to treat conditions with fewer drugs or lower doses. For instance, psychotropic medications for mental health conditions have different levels of efficacy for different people. Often, due to the low efficacy or the side effects of the drug, many patients discontinue their prescribed medication. Genetic testing combined with information on the drug metabolism predicts how individuals with different genetic makeup respond to various psychotropic medications. Patients who metabolise the prescribed drug faster are more likely to respond, albeit at a higher dose and suffer from lesser side effects. Poor metabolisers might not respond as effectively and are more likely to suffer from side effects.[7] These genetic tests such as GeneSight, CNSDose, and Millenium PGT help doctors to tailor the chosen drug and dosage to the individual.

Regenerative medicine is another example of how precision medicine is revolutionising the field. The substantial progress in biomaterial development, cell biology, and tissue engineering allows one to foresee a future where replacements for defective or damaged organs are designed from cells harvested from the same individual. Such replacements can circumvent the potential complications such as rejection of donor tissue and result in improved function of the repaired organ over prolonged periods.[8]

As in any evolving field, there are a number of challenges to overcome. Identifying patients with similar genetic mutations to participate in clinical studies is a challenge as genetic testing is not yet a common procedure. It is difficult to show the effectiveness of a treatment in clinical studies with a small number of patients as such studies do not always arrive at statistically significant results. This poses a problem as insurance companies are reluctant to cover the costs of treatment unless the drug is shown to be effective in a clinical study.[9] Additionally, the costs associated with developing drugs that benefit only a small percentage of people with a disease can be high. For instance, the drug ivacaftor is very effective in about 5% of cystic fibrosis patients with a specific mutation. However, the costs of developing the drug added up to 300,000 US dollars per patient and the drug is ineffective in the remaining 95% who do not have the same mutation.[10]

While there is tremendous value in genetic testing and contributing this data to research, the layperson needs to be aware of the limitations of genetic testing and analysis, especially those that are not stringently regulated. Direct-to-consumer genetic testing, like those offered by companies such as 23andMe, have regulations restricting what they can say about a consumer's health. However, consumers use other companies for data analysis, which are not subject to similar regulations, to report on genetic linkages to diseases. Sometimes these results are misleading due to false positives, either because the person does not carry the mutation or the mutation is not linked to the disease.[11] Many of the detected genetic mutations might be classified as variants of unknown or uncertain significance or if they are linked to disease, they might not be actionable.[12] False negatives or mutations that are not detected by the genetic tests are equally alarming to doctors, as people might not visit their doctor when they should and the opportunity to treat diseases at an early stage might be lost. These limitations associated with precision medicine are at times lost in the hype surrounding it.

The rapidly diminishing costs of genome sequencing herald a future when DNA sequencing is a routine part of one's health check-up. This is already happening in Pennsylvania, USA. Geisinger, a health care system recently announced that DNA sequencing report will be part of their routine clinical care.[13] In the near future, just like high cholesterol levels alerts one to the possibility of developing heart disease and prompts individuals to alter their lifestyle; knowledge of one's genetic makeup and its implications in health and lifestyle choices might be commonplace. Precision medicine is paving the way in reducing the gap from the lab bench to bedside.

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