GUEST EDITORIAL

Big Data, Precision Medicine and Person-Centered Healthcare

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Keywords
Artificial intelligence (AI), big data, clinical observation, empathy, genomics, information overload, Karl Popper, person-centered healthcare, practical reasoning, precision medicine

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Accepted for publication: 30 July 2018

Introduction

Hardly a day goes by without hearing stories about the promises and perils of big data and artificial intelligence in healthcare [1]. As physicians at opposite ends of our careers, we are witnessing first hand the impact these innovations are having on patient care. We are both awed and alarmed. Precision medicine fuelled by big data, such as genomics, promises to change how physicians make diagnoses, determine prognoses and develop new treatments [2].

In some respects, however, big data in medicine is not new. On a daily basis, clinicians are confronted with data from multiple sources and face the challenge of integrating this complex information to improve care for patients. Nonetheless, despite our years of cumulative experience, this data deluge is making us both feel like novices. Is this sense of disorientation simply the result of rapid knowledge expansion in our field? Or is it because new technologies are altering the very meaning of person-centered healthcare [1]?

Conceptual frameworks and clinical observation

We have both encountered information overload at different stages of our careers. We can recall our first clinical encounters as medical students spending hours taking a history and performing a physical examination, fearful of missing some titbit of potentially useful information. We remember presenting these cases to our consultants, who (sometimes) listened patiently to the rambling series of facts - a disjointed review of systems - lacking any coherent narrative description of the patient's illness.

Early on as students, we lacked the necessary conceptual framework that the experienced clinician adds to formulate an assessment. We were reminded of the anecdote recounted by philosopher Karl Popper from when he taught a group of physics students in Vienna in the 1930s [3]. Popper began his lesson with the instructions: "Take pencil and paper; carefully observe, and write down what you have observed!" His students were perplexed - what did he want them to observe? As Popper demonstrated in this exercise, "Clearly the instruction, 'Observe!' is absurd … Observation is always selective. It needs a chosen object, a definite task, an interest, a point of view, a problem."

As novice medical students, data without theory or a conceptual framework was both uninterpretable and uncommunicable. Even though we had learned the medical semantics, we lacked the syntax and grammar to construct a coherent story. In medical education, the transitions from "data gatherer" to "sense maker" to "case manager" have been identified as key steps along the path from novice to medical expert [4].

Big data and precision medicine - the end of theory?

The rise of big data in medicine is giving us a sense of *déjà vu*. Consider the following case commonly seen by a haematologist: a 60-year-old woman referred with fatigue and pancytopenia. From our learned conceptual framework, we form a differential diagnosis which includes myelodysplasia, acute leukaemia and aplastic anaemia. With our patient's consent, we perform a bone marrow biopsy. In addition to microscopic examination of the marrow, we order molecular testing on the sample to screen for common mutations associated with

haematologic malignancies. The bone marrow biopsy shows aplastic anaemia, an uncommon non-malignant disease characterized by loss of stem cells, most often resulting from auto-reactive cytotoxic T-cells. Aplastic anaemia is treated with immunosuppressive drugs, such as anti-thymocyte globulin and cyclosporine, or stem cell transplantation.

Enter precision medicine, complicating the picture. The molecular tests show a number of mutations that have been associated with malignant diseases such as acute myeloid leukaemia. This is not an uncommon finding as up to 50% of patients with aplastic anaemia can have these mutations [5]. Does this information change our approach to treatment in favour of more aggressive therapies, such as stem cell transplantation? Perhaps, but consider the fact that these same mutations have also been identified incidentally in the normal aging population, a condition euphemistically referred to as "CHIP," Clonal Haematopoiesis of Indeterminate Potential [6].

How do we interpret this information? One of our teachers always emphasized that diagnoses are made in the clinic, not in the laboratory, emphasizing that all data must be interpreted within the clinical context. Researchers have developed genetic screening panels based on existing knowledge of genes with cancer potential. Nonetheless, the sheer number of genes analysed, along with the range of possible mutations within each gene (many with unknown functional consequence, so-called "variants of uncertain significance" [7]), make interpreting the potential clinical effects a daunting task. Some hope that machine learning algorithms will help cut through this complexity - that we can hand over this vast body of information to a computer and answers will emerge. While these algorithms may identify associations in data, like our novice student, they lack the conceptual framework necessary to identify causes and effect, both essential for clinical reasoning and scientific advancement [8].

Some pundits have declared "the end of theory" in the era of big data [9]. But as Popper reminded us, data cannot do without theory - all inquiry presupposes interests and points of view [3]. For example, the latest trend in precision medicine brings with it a focus on genomics, which has been criticized for downplaying the importance of other factors, such as social determinants of health. This is not to deny the importance of genes in human diseases, but simply to point out that if we understand disease solely in these terms we will inevitably constrain how we view problems and find solutions. As the common saying goes, if you only have a hammer, everything looks like a nail.

Clinical judgement and person-centered care

So how do we advise our patient with aplastic anaemia, regarding the best treatment option? Like her doctors, she is overwhelmed by the information and struggles to understand the meaning of these results for her as an individual. To help her interpret the data, we discuss the potential future risk of leukaemia weighed against the up-front risks associated with stem cell transplantation. In our dialogue, we also explore her values, review what different treatment approaches might entail and consider how they might align with her personal goals.

In the end, our patient chose treatment with immunosuppressive drugs rather than transplantation. She explained that her decision was based on concern about prolonged hospitalization, being the primary caregiver for her partner who was disabled after having suffered a stroke. This information, which some may consider secondary to her genetic risk profile, is in practice no less germane to clinical decision-making, reminding us of the limits of our data in capturing all the relevant factors.

Conclusion

Amid the data deluge, we risk losing sight of the patient - we risk forgetting the fact that any particular genetic marker exists within a unique individual, often with multiple medical conditions, as well as a complex social and cultural life-world [10]. New technologies threaten to shift the focus of medical knowledge further from the bedside, devaluing first-person knowledge and reducing the patient to data, complete with analytic precision, but lacking meaningful relation to their lived experience. In our rush to embrace big data and artificial intelligence we must not leave behind experiential learning, practical reasoning and empathy, all among the many marvels of human intelligence.

Big data and precision medicine bring both uncertainties and new opportunities. Senior clinicians must remain open to new ideas, while younger trainees must maintain awareness of the limitations of new technologies. Both must not surrender their clinical judgement and responsibility as physicians to their patients in the pursuit of person-centered healthcare.

Conflicts of Interest

The authors declare no conflicts of interest.

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