

令和7年8月5日実施

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問題1 次の文章を読んで、以下の問1～問4に答えなさい。

A Patient-in-the-Loop Approach to Artificial Intelligence in Medicine

The rapid development of artificial intelligence (AI) in medicine has far outpaced its clinical practice implementation. This gap stems largely from the high safety and trustworthiness standards expected and required by health care practitioners. Patient acceptance remains a substantial hurdle, as many lack sufficient information to form opinions on AI; in some cases, this uncertainty even leads to resistance against the integration of AI in their care.

A large-scale survey by Busch et al examined patient perceptions of AI in health care, gathering responses from 13,806 patients across 43 countries in 2023. The study found that while approximately half of respondents held favorable views of AI, acceptance was higher among those familiar with AI or those with higher technological literacy as measured by the number of technology devices used weekly.

Current research on AI in medicine predominantly focuses on evaluating model performance in specific tasks, such as interpreting imaging studies, summarizing clinical notes, or diagnosing from vignettes. While these studies sometimes demonstrate the superiority of AI over physicians in isolated tasks, more realistic scenarios, such as patient history-taking, reveal substantial limitations. These mixed results suggest that the role of AI is best envisioned as augmenting, rather than replacing, physicians' care processes. This notion aligns with the survey findings, in which only 4.4% of patients favored the use of fully autonomous AI systems without physician oversight.

Furthermore, the survey findings by Busch et al revealed that patients prefer less accurate AI systems that are explainable over highly accurate but opaque “closed box” models. This preference underscores the importance of transparency and the ability to audit AI-assisted processes, reinforcing the notion that patients value augmented health care processes in which physicians maintain oversight. Interestingly, the emphasis of patients on explainable AI aligns with the perspective of physicians, as many express a strong intention to integrate AI into their practice but stress the need for transparency, trust, and explainability in these tools.

Involving patients in the design and development of AI systems can play a pivotal role in making

these technologies more acceptable and aligned with patient values. This approach reduces the risk of a mismatch between the capabilities of deployed solutions and the expectations of patients, ensuring that the systems are tailored to clinical practice needs. Moreover, active patient involvement fosters trust, a critical factor for the adoption of AI in health care. Combining this with a physician-in-the-loop strategy could provide a robust framework for AI integration, with both patients and clinicians shaping the development and use of these tools. Rather than aiming to outperform clinicians on narrowly defined tasks, this collaborative approach emphasizes augmenting care processes in ways that resonate with all stakeholders involved.

出典:

Griot MF, Walker GA. JAMA Netw Open. 2025 Jun 2;8(6):e2514460.

問 1 医療における AI の開発と臨床現場での実装の間に、どのような状態が生まれていると述べられていますか？

問 2 Busch et al. の大規模調査によると、AI に対する患者の受容度が高いのはどのような人々ですか？

問 3 本文中で、AI の役割は医師を「置き換える」のではなく「補強する」ものとして捉えるのが最適であると示唆されている主な根拠は何ですか？

問 4 患者が「説明可能な AI」を「ブラックボックス (closed box)」モデルよりも好むという調査結果は、どのような重要性を強調していますか？

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問題2 次の文章を読んで、以下の問1～問5に答えなさい。

Genetics has long been used in specific areas of medicine, such as for the diagnosis of rare diseases caused by pathogenic mutations or in the context of prenatal genetic testing; however, it is now diffusing across many domains of clinical practice. In the UK, for example, all children with suspected or diagnosed cancer are now offered whole-genome sequencing by the National Health Service to inform the use of targeted treatments. And in the USA, ongoing clinical trials are returning information on genetic risk scores to study participants to assess if this information can be used to aid prevention and clinical decision-making for cardiometabolic diseases. In addition, the US Food and Drug Administration is anticipated to approve the first CRISPR-Cas9-based gene therapy in the next few months, which will pave the way for other gene-editing therapies currently in clinical trials for hemoglobinopathies, vision loss and cancer.

The interpretation of clinical genetic information and its communication to patients are by no means trivial. In the case of genetic tests, for example, the ideal scenario is that the test provides information on whether the patient carries an actionable pathogenic variant. The real-life situation, however, is often more complex. Detection of a germline pathogenic variant in a patient could have serious repercussions for other family members or may represent an incidental finding that requires careful counseling. The evidence in support of the pathogenicity of a variant may be weak as is the case of 'variants of uncertain significance', which further complicates the interpretation and subsequent clinical decision-making. In the case of delivering polygenic disease risk score results to patients, clinicians need to understand how to best communicate and act on the results, which are typically broad-range risk estimates. Clinical geneticists and genetic counselors play essential roles in the implementation of genomics in medicine, but their number is limited. In the USA, for example, the number of medical residents entering clinical genetics training programs has not increased substantially over the past 4 years, despite increases in demand.

The increasingly central role of genomics in healthcare means that not only are more genetic counselors needed, but also multidisciplinary teams are essential for utilizing genomic technologies in the clinical setting. Genomic tests (such as those based on whole-exome or whole-genome sequencing) generate an enormous amount of highly complex data, which requires professionals with specialized bioinformatic skills and the know-how to operate within clinically accredited frameworks. In addition, although genomics is currently the most common '-omic' used in the clinic, transcriptomics and proteomics are also being incorporated into algorithms to inform

clinical practice. Globally, clinical bioinformaticians are scarce. In a 2020 report, 'Genome UK: The Future of Healthcare', the UK government predicted a considerable increase in demand over the next 10 years and developed recommendations on how to scale up training of and how to retain computational experts in the clinical setting. The generation of large genomic datasets also poses challenges for data protection and informed consent for reuse, which will require specialized input from bioethics experts.

In addition to the need for scaling up the training and retention of bioinformaticians, clinical geneticists and counselors, physicians themselves will need to be better prepared to work confidently with genetic information. Although basic genetics classes are typically offered in medical school curricula, reports from Australia and the USA show that the genetic literacy of physicians varies widely across disciplines, and even within fields it is often inconsistent and dependent on the personal interest of residents and supervisors. Genomic technologies evolve rapidly, and there is a pressing need to update genetics teaching curricula and training in medical education. Some universities have already started to address this gap with initiatives aimed at enhancing undergraduate medical education in genomics and recruiting medical graduates into genetics residency training programs, but more formal education programs for medical students and current clinicians are urgently needed.

Genomics has also introduced new challenges to health equity. Sequencing remains an expensive clinical tool and gene therapies are typically very costly. These expenses risk exacerbating already existing inequities in access to healthcare. Such inequities to access are also widened by the scarcity of genomic data generated from people of diverse ancestries. For the most part, currently available disease polygenic risk scores have been trained on data from people mainly of European ancestries, and the resulting models are consequently less accurate when applied to understudied populations. If this research practice is not corrected, genomic medicine will continue to benefit only the few. Additional funding to diversify sequencing efforts and to increase awareness of these issues is needed to ensure that the promise of genomic medicine is available to all.

A rapidly evolving genomic revolution is poised to shape the future of healthcare, but its full clinical potential can be realized only with the development of a multidisciplinary healthcare workforce capable of evolving to stay abreast of rapidly developing genetic technologies.

出典： Nat Med. 2023 Aug;29(8):1877-1878.

問 1 実際の医療現場において、臨床遺伝情報の解釈が困難である理由を挙げなさい。

問 2 イギリスまたはアメリカにおけるゲノミクスの臨床応用の具体例を 2 つ挙げなさい。

問 3 ゲノム医療の導入に際して直面する主な障壁を 2 つ挙げなさい。

問 4 なぜ医師に対してゲノミクスに関する教育が重要なのか説明しなさい。

問 5 ゲノミクスが健康の公平性に課題をもたらす理由を挙げなさい。