Personalized Medicine

CHELSEA L. RATCLIFF
University of Georgia, USA

Personalized medicine and related terms precision medicine and precision health refer to a medical model based on the idea that medicine should not be one-size-fits-all but instead tailored to a person's unique biological, behavioral, and environmental factors (Collins & Varmus, 2015). Although the terms are often used interchangeably, *personalized medicine* is sometimes considered broader than *precision medicine*, the former referring to personalization across a wide range of health factors and the latter referring to a focus on genomic and physiological biomarkers (Hekler et al., 2020; Juengst et al., 2016). Further, personalized and precision medicine typically refer to the prevention, diagnosis, or treatment of disease within the context of the healthcare system, while *precision health* represents a broader framework that includes public health and consumer health management (Hekler et al., 2020). All of these concepts will be considered in the current entry.

Healthcare has always, to some degree, been personalized, or geared toward the individual patient. Yet thanks to advances in genomic medicine and biomedical informatics, avenues are emerging to support the complex, multifactorial personalization of medicine (Collins & Varmus, 2015; Hekler et al., 2020). For instance, information technologies enable the collection, analysis, and synthesis of broad swathes of health-relevant data, such as genomic and metabolomic data; electronic health records (EHRs) that can include patients' diagnostic history, family health history, and self-reported symptoms, behaviors, life events, and environmental exposures; and patient-generated health data (PGHD) from smart devices and wearables (Swan, 2012). Using machine learning or other algorithms, this confluence of diverse data streams will ideally make it possible to provide highly tailored health information and interventions customized to the individual (Swan, 2012).

Large-scale research programs have been launched around the world to facilitate discoveries that will support personalized medicine. Many of these programs leverage collaborations among government, healthcare, academia, industry, and community partners to recruit volunteers into large cohorts and make their data broadly available for research. These include the UK Biobank, the World Health Organization's International Agency for Research on Cancer Biobank, and the United States-based All of Us Research Program led by the National Institutes of Health. At the forefront of many of these research initiatives is a goal to develop better therapeutic approaches for the treatment of diseases, especially cancers (Collins & Varmus, 2015).

Despite the potential for using a wide spectrum of factors to generate tailored health guidance, personalized medicine research programs have focused primarily on

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genomic discoveries. Genome-wide association studies make it possible to search for markers of risk across all of a person's genes, as well as to examine how their genes interact with each other and with external influences. Also referred to as "personalized genomic medicine" (Juengst et al., 2016, p. 22), genomic testing in clinical and direct-to-consumer (DTC) settings is expected to hold the keys to more effective risk profiling and treatment of physical and mental health conditions. By taking into account a patient's genomic makeup, it is hoped that *pharmacogenomics* will enable "the provision of the right drug at the right dose to the right patient" (Collins & Varmus, 2015, p. 795). For instance, drugs targeting specific gene variants can be safer and more effective for cancer patients than standard therapies. Gene-targeted drugs are in development for numerous other health conditions, including cardiovascular diseases and mental health conditions such as Alzheimer's and major depression.

Some scholars argue that the label precision medicine is preferable to personalized medicine, as it shifts the responsibility for health away from the individual (Juengst et al., 2016). At the same time, the *precision* label may give an unrealistic picture, as people are typically being stratified into subgroups rather than receiving truly individualized information (Juengst et al., 2016). Furthermore, highly personalized health information does not always generate a precise or clear roadmap for health management. Scholars have also criticized the emphasis on genetics in personalized medicine over other factors, such as social and behavioral factors, which are known to be stronger determinants of health.

Communication challenges and opportunities

Health communication scholars have an important role to play in the realization of effective and equitable personalized medicine (Hekler et al., 2020; Scherr et al., 2017). Central areas for personalized medicine communication research are described below.

Research recruitment and informed consent

Personalized medicine is largely still in the discovery phase, with research programs recruiting sizable cohorts of patients and healthy people to drive discovery. These studies generate databases that contain participants' DNA, along with linked personal health information from EHRs, self-report questionnaires, and wearables (Collins & Varmus, 2015). Linkage to additional types of health-relevant data, such as credit card purchase histories, criminal records, and social media activity, has also been proposed.

Many of these programs will continuously collect data from participants in order to enable longitudinal research. Given that future use of participant data is undetermined at the time of enrollment, the traditional model of informed consent is considered untenable in personalized medicine research, and volunteers are instead asked to provide "broad" or "open" consent. Therefore maintaining trust is considered crucial to the successful recruitment and retention of research volunteers (Ratcliff et al., 2021; Scherr et al., 2017). This trust is expected to come through transparency, as well as through inclusion of medically underserved groups and those historically underrepresented in

biomedical research (Scherr et al., 2017). Fairness in personalized medicine will depend on building diverse research participant cohorts, in order to ensure the knowledge generated has broad and equitable population benefit.

Scholars highlight a need for evidence-based research to guide communication best practices in this area, including research on how to support informed decisions about participation, how to return research results to participants, and how to engage diverse populations (Kaphingst et al., 2019; Morgan et al., 2022; Ratcliff, Kaphingst, & Jensen, 2018; Scherr et al., 2017). There is also a need to understand how to engage research volunteers as *partners* (Collins & Varmus, 2015), such as by soliciting input from participants about how their data should be used and which types of results they would like to receive.

Healthcare delivery

Personalized medicine is closely aligned with the goals of patient-centered healthcare and shared decision making, empowering patients with personalized information to help them manage their own health (Hekler et al., 2020; Swan, 2012). It is also driven by the vision for *connected health*, or the use of information technology to enable the efficient collection, flow, and use of health information in the healthcare system (Collins & Varmus, 2015). In line with these goals, more research is needed to inform how healthcare providers should collect data from patients to support personalization and how they should communicate personalized health recommendations to patients (Ratcliff et al., 2018). Healthcare providers often feel underprepared to discuss genetic test results with patients, highlighting the importance of genetic counselors in personalized medicine delivery as well as the need for communication training for healthcare providers (Scherr et al., 2017).

With cancer genetics at the forefront of precision medicine, a sizable body of research has examined best practices for communicating with patients about the benefits and drawbacks of genetic testing, how to convey test results and support patient decision making, and how to lessen negative impacts of behavioral and psychosocial outcomes from testing (Kaphingst et al., 2019). However, this research has primarily focused on women, Caucasians, older adults, and individuals with higher socioeconomic status (Kaphingst et al., 2019), indicating a need to include diverse populations in future studies.

Research is also needed to understand how to support individuals who receive personalized health information outside the healthcare setting, such as those who undergo DTC genetic testing, as well as how individuals can discuss this information with their healthcare providers. Interpretation may be supported by the development of educational resources and interventions to improve genetic and other health and numeric literacy skills.

Conveying complex risk information

A primary aim of personalized medicine is to generate personalized disease risk profiles. These include polygenic risk scores, or sums of the estimated impact of multiple

gene variants on a person's risk for a disease. For example, a person with a greater number of genetic markers associated with depression is thought to be at higher risk of depression. The potential for gene–environment interactions, in which outside factors (e.g., life events, lifestyle behaviors, or environmental exposures) change the expression of a gene and can "awaken" genetic predispositions, may also figure into patients' risk profiles.

Compared to standard recommendations, personalized health guidance has the potential to increase individuals' engagement with their health (Swan, 2012). At the same time, there is also the potential for personalized risk information to be more disturbing or stigmatizing (Ratcliff et al., 2018). Personalized information could also trigger "personalization reactance" if patients find it to be an invasion of their privacy or to threaten their sense of self-determination (Ratcliff et al., 2018).

Although personalized health information is expected to be more precise and thereby reduce uncertainty, it can also be more complex or ambiguous than generic information, at times increasing uncertainty (Juengst et al., 2016; Ratcliff et al., 2021; Scherr et al., 2017). For example, a key challenge in the return of genetic test results is helping patients navigate information about gene variants of uncertain significance or utility (Scherr et al., 2017; Kaphingst et al., 2019). As personalized approaches evolve to incorporate a broad range of individual health factors, the interpretation and use of such data may become even more complex.

Personalized communication

Patients and healthcare consumers have different information preferences, literacy skills, background knowledge, and privacy needs. Scholars have suggested that if healthcare be personalized, so should its delivery, with communication tailored to an individual's preferences and values. For instance, patients should be able to opt out of receiving certain types of personalized information, such as information of uncertain meaning or utility or information of a stigmatized or sensitive nature (Morgan et al., 2022; Ratcliff et al., 2018; Scherr et al., 2017). Transparency about the types of personal data gathered from individuals may also be appropriate, as well as which variables are used in algorithms upon which personalized guidance is based. Digital platforms can be used to enable the delivery of tailored health information and health interventions, and communication research is needed to facilitate this process, such as identifying strategies to enhance patients' digital literacy.

The roles of digital health engagement and health information technologies

Digital health engagement plays a pivotal role in personalized medicine. Digital tools make it possible to collect a wide range of biological and behavioral data from individuals, both within and outside the healthcare system (Collins & Varmus, 2015; Hekler et al., 2020). Individuals can use digital tools like smart devices and wearables to collect PGHD, either for their own use or to share with healthcare providers in order to facilitate personalized care. For example, smart device apps make it possible to keep track of behaviors, moods, and physiological symptoms and to monitor progress toward health

goals (e.g., a smoking cessation app to track cravings and identify and manage triggers). Biosensors or wearables (e.g., Fitbit, Apple watch, blood pressure monitors) and implants (e.g., pacemakers) can also be used to generate data, as well as automatically transmit data to healthcare providers (Swan, 2012). Additionally, many personalized medicine research programs request PGHD from participants. Thus, facilitating access to and use of digital health tools among patients, consumers, and research participants is essential to the success of personalized medicine.

On the systems side, it will be equally essential that infrastructure is in place to use the insights generated from this ever-growing swathe of digital data (Collins & Varmus, 2015; Hekler et al., 2020). This includes integration of a broader range of patient data and sophisticated algorithms in clinical decision support systems. It also includes the development of dynamic, adaptive interventions that use these data – interventions grounded in health behavior theories (Scherr et al., 2017) and that continuously tune to the individual patient, rather than being based on prior participants' responses to interventions (Hekler et al., 2020). It will be necessary to develop standardized ways of coding the data generated from research participants and patients so that data can be effectively searched, organized, curated, and translated into clinically actionable knowledge to guide personalized care (Hekler et al., 2020). Healthcare providers will also require training in order to translate the insights generated from technology systems into patient guidance and care (Scherr et al., 2017). Thus, success on the systems side will depend upon transdisciplinary teamwork and development of a shared language within and across health-relevant sectors (Scherr et al., 2017). As Hekler et al. (2020, p. 817) observed: "Without innovative tools to support cross-sector and disciplinary access and curation, the vision of precision health may become a proverbial Tower of Babel that is largely inaccessible and, therefore, of questionable value." Technology developers, data scientists, and health communication researchers will all play pivotal roles in the creation of such tools.

Media portrayals

The media is considered a key partner in disseminating information about personalized medicine to the public, including opportunities to participate in research programs. However, media portrayals of personalized medicine are frequently hyped, exaggerating potential benefits and omitting mention of important caveats (Marcon, Bieber, & Caulfield, 2018). Future scientific discoveries are often presented as guaranteed, and achieved initial discoveries are frequently misrepresented as "revolutionary" with immediate implications for prevention, detection, and treatment. Further, there is often no mention of ethical, legal, and social implications of personalized medicine, such as those entailed by sharing DNA and other personal data for broad use (Marcon et al., 2018). Caveats such as unestablished validity of genetic markers and unknown generalizability of findings from European ancestry samples to those of non-European ancestry are rarely discussed. Recent work suggests that depicting these types of uncertainties in news coverage is unlikely to hurt public support and engagement in precision medicine research, and disclosure of scientific uncertainty may have a positive impact on perceived ethicality of the scientists (Ratcliff et al., 2021). Promoting

realistic expectations among patients and the broader public is essential in order to maintain trust in the research and medical enterprises, making media-based and other public communication about personalized medicine a critical area for further study.

Summary

Though showing great promise, personalized medicine is still in the early stages of research and implementation. Its success will depend not only upon continued biomedical and bioinformatics innovations, but also upon social and behavioral research – including communication research – to drive discovery and clinical translation. Research is needed to inform best practices for communication between patients and healthcare providers; between research participants and research institutions; and among scientists, the media, and the public.

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SEE ALSO: eHealth Literacy; Health Informatics; Genetic Communication; Digital Tailored Strategies; Customization; Electronic Health Records; eHealth; Message Tailoring; Clinical Decision Support Systems; Diffusion of Health Information Technology.

References

Collins, F. S., & Varmus, H. (2015). A new initiative on precision medicine. *New England Journal of Medicine, 372*, 793–795. https://doi.org/10.1056/NEJMp1500523

Hekler, E., Tiro, J. A., Hunter, C. M., & Nebeker, C. (2020). Precision health: The role of the social and behavioral sciences in advancing the vision. *Annals of Behavioral Medicine, 54*, 805–826. https://doi.org/10.1093/abm/kaaa018

Juengst, E., McGowan, M. L., Fishman, J. R., & Settersten, R. A., Jr. (2016). From "personalized" to "precision" medicine: The ethical and social implications of rhetorical reform in genomic medicine. *Hastings Center Report, 46*, 21–33. https://doi.org/10.1002/hast.614

Kaphingst, K. A., Peterson, E., Zhao, J., Gaysynsky, A., Elrick, A., Hong, S. J., … Chou, W.-Y. S. (2019). Cancer communication research in the era of genomics and precision medicine: A scoping review. *Genetics in Medicine, 21*, 1691–1698. https://doi.org/10.1038/s41436-018-0402-0

Marcon, A. R., Bieber, M., & Caulfield, T. (2018). Representing a "revolution": How the popular press has portrayed personalized medicine. *Genetics in Medicine, 20*, 950–956. https://doi.org/10.1038/gim.2017.217

Morgan, S. E., Peng, W., Occa, A., Mao, B., McFarlane, S., Grinfeder, G., … Byrne, M. M. (2022). Tailored messages about research participation: Using an interactive information aid to improve study recruitment. *Journal of Cancer Education, 37*(1), 16–22. https://doi.org/10.1007/s13187-020-01775-5

Ratcliff, C. L., Kaphingst, K. A., & Jensen, J. D. (2018). When personal feels invasive: Foreseeing challenges in precision medicine communication. *Journal of Health Communication, 23*, 144–152. https://doi.org/10.1080/10810730.2017.1417514

Ratcliff, C. L., Wong, B., Jensen, J., & Kaphingst, K. A. (2021). The impact of communicating uncertainty on public responses to precision medicine research. *Annals of Behavioral Medicine, 55*, 1048–1061. https://doi.org/10.1093/abm/kaab050

Scherr, C. L., Dean, M., Clayton, M. F., Hesse, B. W., Silk, K., Street, R. L., Jr., & Krieger, J. (2017). A research agenda for communication scholars in the precision medicine era. *Journal of Health Communication, 22*, 839–848. https://doi.org/10.1080/10810730.2017.1363324

Swan, M. (2012). Health 2050: The realization of personalized medicine through crowdsourcing, the quantified self, and the participatory biocitizen. *Journal of Personalized Medicine, 2*(3), 93–118. https://doi.org/10.3390/jpm2030093

Further reading

Chambers, D. A., Feero, W. G., & Khoury, M. J. (2016). Convergence of implementation science, precision medicine, and the learning health care system: A new model for biomedical research. *JAMA, 315*, 1941–1942. https://doi.org/10.1001/jama.2016.3867

Faulkner, E., Holtorf, A. P., Walton, S., Liu, C. Y., Lin, H., Biltaj, E., … Payne, K. (2020). Being precise about precision medicine: What should value frameworks incorporate to address precision medicine? A report of the Personalized Precision Medicine Special Interest Group. *Value in Health, 23*(5), 529–539. https://doi.org/10.1016/j.jval.2019.11.010

Ferryman, K., & Pitcan, M. (2018). Fairness in precision medicine report. Data & Society. https://datasociety.net/research/fairness-precision-medicine

Juengst, E. T., & McGowan, M. L. (2018). Why does the shift from "personalized medicine" to "precision health" and "wellness genomics" matter? *AMA Journal of Ethics, 20*, 881–890. https://doi.org/10.1001/amajethics.2018.881

Sankar, P. L., & Parker, L. S. (2017). The Precision Medicine Initiative's All of Us Research Program: An agenda for research on its ethical, legal, and social issues. *Genetics in Medicine*, *19*, 743–750. https://doi.org/10.1038/gim.2016.183

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ABSTRACT

"Personalized medicine" and the related concepts of "precision medicine" and "precision health" refer to the idea that healthcare approaches should not be one-size-fits-all but instead tailored to a person's unique biological, behavioral, and environmental factors. Personalized medicine largely relies on individuals' digital health engagement and on healthcare informatics to generate personalized guidance. A central focus of personalized medicine is genomic testing to generate more accurate disease risk profiles and to guide gene-targeted therapies (i.e., pharmacogenomics). Health communication scholars have an important role to play in the success of personalized medicine. From research to clinical implementation, effective communication is key to enabling and implementing the discoveries that support a personalized approach to health. Central areas for communication research include research participant recruitment, digital health engagement and bioinformatics, conveying complex risk information to patients and consumers, and the portrayal of personalized medicine in the media.

KEYWORDS

digital health engagement; genetics; genomics; health informatics; personalized medicine; precision health; precision medicine; risk profiling; tailoring