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# MOVING BEYOND POPULATION AVERAGES

A Patient-Centered Research Agenda  
Advancing Personalized Medicine

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# Introduction

Personalized medicine presents opportunities to understand how differences in an individual's biology affect a person's health so that prevention strategies and treatments are guided to those patients who will benefit. However, there are fundamental gaps in awareness and access that impact the speed at which personalized medicine treatments and strategies are integrated into health care.

Through a two-year Eugene Washington Engagement Award from the Patient-Centered Outcomes Research Institute (PCORI) (Contract No. 10438-PMC), the Personalized Medicine Coalition (PMC), a coalition of over 230 organizations from across the health care system, convened patients and other health care stakeholders to develop a research agenda advancing personalized medicine that is informed by early successes in personalized medicine adoption and builds on principles defined by patients. By developing this research agenda, PMC has also sought to establish partnerships among patients, health care professionals and researchers to build a community that will work together to advance patient-centered research.

The research agenda that follows identifies 45 research topics to improve the delivery of personalized medicine in ways that are most meaningful to patients.

## What is Personalized Medicine?

Personalized medicine, often referred to as precision medicine, is an evolving field in which physicians use molecular diagnostic tests to determine which medical treatments and strategies will work best for their patients. By combining the data from those tests with an individual's medical history, circumstances, and values, health care providers and patients can develop targeted treatment and prevention plans.

### **Personalized medicine can benefit patients in a number of ways, including by**

- shifting the emphasis in medicine from reaction to prevention;
- increasing treatment effectiveness by directing targeted therapy and reducing trial-and-error prescribing;
- reducing adverse drug reactions;
- using cell-based or gene therapy to replace or circumvent molecular pathways associated with disease;
- revealing additional targeted uses for medicines and drug candidates;
- increasing patient adherence to treatment;
- reducing high-risk invasive testing procedures;
- helping to shift physician-patient engagement towards patient-centered care; and
- helping to control the overall cost of health care and reduce low-value care.<sup>1</sup>

As with patient-centered outcomes research (PCOR), personalized medicine aims to shift health care from a one-size-fits-all, trial-and-error approach toward a targeted approach that utilizes patients' characteristics and preferences to inform health care decisions.

## Project Overview

Patients, caregivers,<sup>2</sup> and patient advocates have participated in every aspect of this project. From the beginning, PMC prioritized patient input to determine the project's outcomes and engagement with other stakeholders and invited new voices to participate as gaps in perspectives were revealed.

PMC leveraged its network of patient advocacy organizations to invite patients from across the United States to participate. The project consisted of a series of four web forums to identify guiding principles that underpin potential research questions, an online collaboration platform<sup>3</sup> to brainstorm related research questions, virtual roundtables with patients and other stakeholders to finalize the research questions, and an advisory committee consisting largely of patient representatives who guided this process.

Patients, patient advocates, caregivers, patient advocacy organization representatives, health care professionals, researchers, and other stakeholders representing diverse backgrounds, disease areas, health needs, and with varying levels of experience with personalized medicine participated in project activities.

Throughout these activities, PMC focused on building a shared understanding among participants of how a research agenda could improve patients' experiences with personalized medicine. As a result of their engagement, participants reported an increased understanding of personalized medicine's benefits and the challenges it can present for some patients and health care providers.

This white paper summarizes patient-centered guiding principles defined by the project participants, the discussions that took place during the roundtable sessions, and the research agenda that emerged reflecting patients' priorities for additional research based on their experiences with personalized medicine.

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1 [http://www.personalizedmedicinecoalition.org/Userfiles/PMC-Corporate/file/The\\_PM\\_Report.pdf](http://www.personalizedmedicinecoalition.org/Userfiles/PMC-Corporate/file/The_PM_Report.pdf)

2 In the following paper, "caregiver" refers to family or friend caregivers. The *RAISE Family Caregivers Act* defines a family caregiver as "an adult family member or other individual who has a significant relationship with, and who provides a broad range of assistance to, an individual with a chronic or other health condition, disability, or functional limitation." [*RAISE Family Caregivers Act of 2018* (P.L. No: 115-119), available at <https://www.congress.gov/bill/115th-congress/house-bill/3759>.] In research and in advocacy, "caregiver" may be described as: informal caregiver, care partner, care taker, and related terminology.

3 <https://codigital.com>

# Patient-Centered Guiding Principles

At the beginning of the agenda-setting process, PMC led a series of online web forums to engage patients, patient advocates, and caregivers, as well as health care professionals and researchers, to develop a set of patient-centered principles to inform the research agenda. Over 120 participated, including more than 90 representatives from the patient, caregiver and patient advocacy communities. Participants had varying levels of familiarity with personalized medicine and patient-centered outcomes research, and they represented an array of disease areas and backgrounds.

The following guiding principles define aspects of patients and caregivers' values and circumstances that patients think should be considered in the treatment decision-making process; highlight areas for improving patient-provider communication, improving the education of patients and providers, and addressing patients' access challenges; and indicate how patients think a research agenda advancing personalized medicine could be most impactful and representative of diverse patient needs, diseases, backgrounds and experiences.

## Guiding Principles

1. Patient/caregiver values include personal priorities, religious/spiritual values, societal and cultural values (including family involvement in care decisions), views around quality of life, privacy concerns, desired level of access to and understanding of personal genetic information, beliefs about health and personal responsibility, and attitudes and preferences toward end of life.
2. Patient/caregiver circumstances include emotional state, socioeconomic situation, race/ethnicity, sex/gender, language, health literacy, ability to work, access to care and health insurance, access or lack thereof to a caregiver, social support, cognitive abilities, attitude toward illness (e.g., acceptance of diagnosis, willingness to accept help), personality, symptom burden, health-related quality of life, ability to consent and choose, relationship with the health care provider, the role of patient as caretaker, preferences of family members, treatment setting (e.g., community, academic, other), familiarity with personalized medicine and its benefits, other social determinants of health, and other expressed needs or barriers.

3. The research agenda would be most helpful if it focused on priorities that help providers (a) identify and communicate treatment options based on biological differences in the context of patient/caregiver values and (b) understand patient/caregiver circumstances and present treatment options in ways that highlight how a treatment aligns/does not align with those circumstances.
  - › The potential for shared decision-making tools and resources to facilitate these conversations should be explored.
4. The research agenda should address the education of patients directly by considering (a) the education of patients/caregivers as a continuing process over time and not just at diagnosis, including basic education defining personalized medicine and its benefits, (b) how both patients/caregivers and their providers can improve communication with each other, and (c) how to close the gap between patient/caregiver expectations and the reality of how/when test results and treatment options are delivered.
  - › Educational tools and resources, including those available online, should address different learning styles and levels of health literacy.
5. Opportunities to educate providers in genomics should be considered, along with providers' varying levels of understanding of genomics and varying levels of access to new and existing resources, such as translational tools, depending on their health setting (e.g., community, academic, other), health specialization, and health discipline (e.g., physicians, pharmacists, physician assistant, genetic counselors, nurse).
  - › This includes opportunities to educate health care providers and professionals in training.
6. The agenda should consider how to use oncology as a prototype for patient and provider education in disease areas beyond oncology.
7. Access challenges beyond those created by a lack of communication and education should also be considered.
  - › This includes timely access to novel and/or off-label therapies as a result of affordability, insurance coverage and provider availability.
  - › This includes strategies for mitigating inequity in the proportion of significant variants reported out for individuals of non-Western European ancestry/ethnicity compared with those of Western European ancestry/ethnicity.

8. The research agenda should be representative of diverse patient needs, diseases, backgrounds, and experiences (e.g., stage of life, disease trajectory, socioeconomic status and health literacy level), including patients underrepresented in medical research.
  - › Specifically, the research agenda should apply across disease areas.
  - › This includes considering how the research agenda can apply to additional disease areas as new personalized medicine treatments come to market.
  - › This includes potential research topics related to the management of comorbidities.
9. The advisory committee for developing the research agenda should consider areas the agenda must include or exclude in order to have the greatest impact, including the feasibility of proposed research topics.
  - › This includes potential research topics related to artificial intelligence, data collection, data integration and interoperability, informed consent, and patient concerns related to data privacy and access.

The guiding principles define aspects of patients and caregivers' values and circumstances that patients think should be considered in the treatment decision-making process; highlight areas for improving patient-provider communication; and indicate how patients think a research agenda advancing personalized medicine could be most impactful and representative of diverse patient needs, diseases, backgrounds and experiences.

## ROUNDTABLE SUMMARY

# Defining A Patient-Centered Research Agenda

To develop patient-centered research questions based on the guiding principles, PMC first invited patients and other members of the health care community who had been participating in PMC's web forum series to brainstorm research questions for discussion during the roundtables. Through an online collaboration platform, participants were asked, "If you could choose one research question to improve the care or treatment of people through personalized medicine, what would that be?" There were 40 contributors, including patients/caregivers (44%), representatives of patient advocacy organizations (26%), health care professionals (18%), researchers (10%) and other types of stakeholders (3%)<sup>4</sup> (see Figure 1).

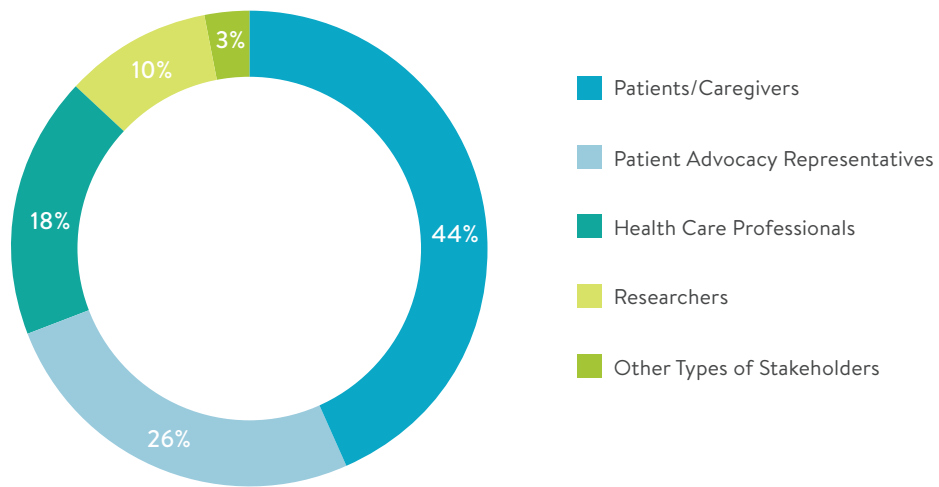
PMC then convened 55 participants for three virtual roundtable sessions in June 2020 to refine and build upon these research questions. Participants had varying levels of familiarity and experience with personalized medicine, ranging from those who have heard of personalized medicine but do not have a treatment available yet for their disease to those who have been treated with personalized medicine or use personalized medicine in their profession. Some participants had already been participating in the project, and many provided new perspectives.

The patient experience grounded these discussions, with various stakeholders represented. Roundtable session participants included patients and caregivers (27%), representatives of patient advocacy organizations (27%), health care professionals (25%), researchers (11%), and other types of stakeholders (9%)<sup>5</sup> (see Figure 2). Disease areas represented included Alzheimer's disease, cancer (breast, lung, multiple myeloma, colorectal, lymphoma, rare blood, genetic risk), COPD, cystic fibrosis, food allergy, HIV, kidney disease and rare diseases. Participants also had expertise in caregiving, community health, diagnostics, family medicine, genetic counseling, genomics education, health communication, health disparities, health insurance, health information technology, hospital and program administration, nursing and nurse education, outcomes research, patient education, pharmacogenomics, and practicing physicians/researchers from various specialties. Industry representatives, a payer representative, and a government representative from the U.S. National Institutes of Health also participated.

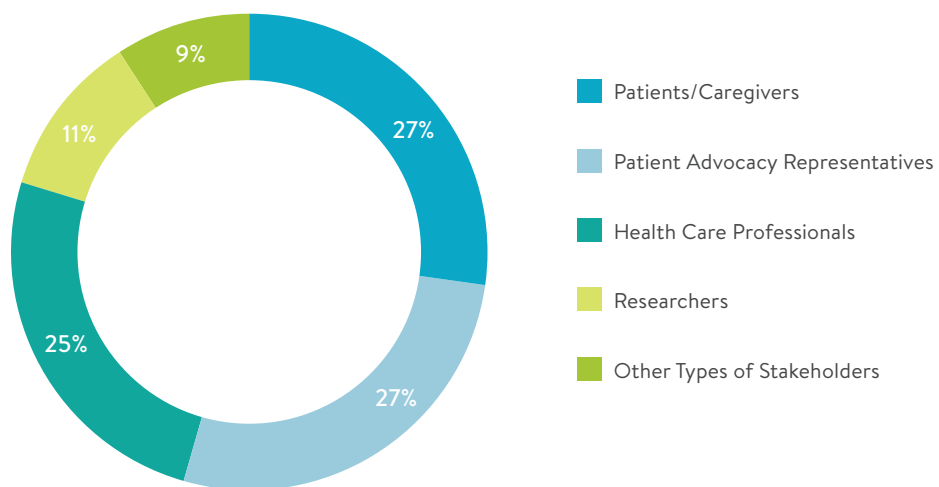


During the roundtables, summarized below, PMC utilized the patient-centered guiding principles and focused the discussions on building a shared understanding among patients and other health stakeholders about research questions important for improving patients' experiences with personalized medicine and about the varying challenges facing both patients and health care professionals.

**FIGURE 1: A Breakdown of the Contributors to the Preliminary Roundtable Questions**



**FIGURE 2: A Breakdown of the Roundtable Participants by Stakeholder Category**



## ● **ROUNDTABLE SESSION I**

### **Improving the Patient Experience with Personalized Medicine**

The first session included a panel of patient representatives who shared their experiences with personalized medicine and reflected on the patient-centered guiding principles. During the meeting participants shared their experiences with stage IV inflammatory breast cancer, cystic fibrosis, a rare genetic epilepsy, caregiving and health disparities.

Whether an individual patient is able to access and benefit from a personalized medicine approach for his or her condition depends on multiple factors including the specific genetic mutation present, the availability of a targeted therapy for that mutation, and access to a clinical trial or practice where the therapy can be obtained. Although the vast majority of patients are treated in community settings, in some cases personalized medicine may only be available in academic settings, and community physicians may not have conversations with patients about personalized medicine due to their lack of awareness about personalized medicine treatments and strategies. Physicians practicing in academic settings may also not be fully educated about personalized medicine. As a result, the potential benefits of a personalized medicine approach may not be realized unless the patient is aware that this option is available and is fortunate enough to have a caregiver and provider who will advocate for them. Good fortune, however, should not play a central role in one's ability to access personalized medicine. Broad public educational programs to increase health literacy around personalized medicine, as well as provider-focused education programs, can help take luck out of the equation.

Partnering with advocacy organizations may help accelerate the development of patient education programs. To reach a diverse spectrum of patients and caregivers with varying educational, language, and literacy levels, multiple types of educational materials will be needed, ranging from animated videos to three-page briefs. The development of educational programs that target minority populations should include not only disease group organizations and advocacy groups but also cultural and religious organizations, such as faith-based health ministries, in order to address historical, cultural, and religious experiences, fears, and taboos that may engender mistrust and present obstacles to participation in research, clinical trials, and treatment with novel therapies.

Educating health care professionals about personalized medicine is also needed. This education is especially important for primary care providers and specialists in fields like pediatrics who may need to facilitate family health communication about a hereditary condition and what that means for a patient's children and other family members. Non-physician specialists, patient ambassadors, peer educators and navigators,<sup>6</sup> and telemedicine could help to address barriers to patient education and access to personalized medicine that may be caused by a shortage of expertise at the patient's local health care institution.

The complexity of personalized medicine and the rapid pace at which it is evolving further complicate the development of adequate and appropriate educational programs, not only for patients but also for health care providers. For example, some personalized medicine treatments target common genetic mutations but are unavailable to those with rarer mutations due to a lack of research on the treatment's efficacy in people with rarer mutations. Other gene-targeting personalized medicine approaches target modulators rather than specific mutations and thus may be available to patients regardless of genotype. Genetic testing and personalized medicine might also be used to identify the best candidates for promising new drug therapies, and thus enable effective treatment of non-genetically based disorders.

Yet even with adequate educational resources, patients and caregivers may feel overwhelmed by the amount and complexity of the information they receive at a time when they are still processing and grieving about their diagnosis with a health condition that could be life-threatening. They need adequate time to consider options before making a final decision, as well as behavioral health support to manage their feelings of grief, anger, etc. Patients should also be educated and have resources available to learn about personalized medicine treatment strategies and testing options throughout their care journey, including pre-diagnosis, at diagnosis, during treatment decision-making, and during follow-up visits.

Multidisciplinary teams are essential to ensure comprehensive care, support and education for patients embarking on personalized medicine treatment approaches. As personalized medicine becomes more widely available, care coordination becomes more challenging, particularly for family and friend caregivers and caregivers to children. Care coordination initiatives should be expanded or developed to include caregivers on the treatment and decision-making team(s).

Ensuring equity requires attention to the cost of genetic testing and the financial burden associated with travel, time away from work, and childcare, all of which can pose substantial barriers to patients and caregivers hoping to access personalized medicine. The active engagement of underrepresented populations is essential across all aspects of personalized medicine research and treatment delivery to ensure equity across diverse population groups and avoid exacerbating systemic biases that lead to further health inequities.

## ● **ROUNDTABLE SESSION II**

### **Improving the Delivery of Personalized Medicine in the Clinic**

The second session featured a panel comprising an expert in health communication and shared decision-making who is also a parent and advocate for a child with a food allergy; a genetic counselor working in the field of cardiovascular genetics; and the director of a community cancer center and research institute. Discussion participants included patients, providers, and advocates with other diseases that may be targeted by personalized medicine approaches, such as rare neurological disorders and sickle cell disease.

One of the major functions of a health care provider is to suggest the optimal treatment for a patient and communicate the rationale, benefits and risks of that treatment to the patient and caregiver. The explosion of personalized medicine testing and treatment technologies has made this increasingly challenging for providers, particularly those in community settings where they may see relatively few patients with disorders that may be eligible for personalized medicine treatment options.

Added to that complexity is the fact that many new targeted agents are very specific to small populations of people with a particular disease. This requires subdividing patients into small subgroups through genetic testing. However, some genetic testing results are indeterminate, in that while they identify a probability of a therapy being effective for a patient, the clinical implication of that test result remains uncertain.

Another barrier to identifying appropriate treatment is the lack of receptivity among some health care professionals to listen carefully to patients and consider, for example, what the patient has experienced as helpful in treating their condition. These challenges make the dialogue between provider and patient both increasingly important but also increasingly fraught. To engage in shared decision-making when discussing personalized medicine treatment options, the provider must understand considerations important to the patient and caregiver, including their values, circumstances, and preferences, while also educating the patient and caregiver about what factors to consider.

The roles of other health care professionals, including genetic counselors, pharmacists, nurse navigators<sup>7</sup> and social workers, should be maximized to help alleviate some of the burdens for expertise and patient education that can fall on physicians. Genetic counselors, for example, can play an essential role in these conversations with patients due to their knowledge across the broad array of genetic and hereditary conditions. They also have access to the tools that can help assess a family history and results of genetic testing, as well as training in how to communicate that knowledge to patients and caregivers, who may be reluctant to discuss their family history. While genetic testing done in a research setting may not require reimbursement by health insurance, once the test becomes available in clinical settings, issues arise not only in navigating criteria for reimbursement but in selecting one from a number of competing tests available from different laboratories and/or companies. There are often strategies for financing genetic testing through supplementary sources, but providers may not know about all the resources available. Maximizing the roles of other health care professionals, continuing education, and clinical decision support tools could all help address barriers health care providers face in delivering personalized medicine to their patients.

There is also a need to develop a path forward that explains the limitations and purpose of any particular genetic test. Genetic tests are only available for those mutations that are known, to-date, to be associated with a particular condition. Negative genetic test results do not necessarily mean the patient does not have genes that may affect their condition, but rather any genetic associations related to their disease have not yet been identified and

validated for testing. Receiving a negative test result can cause frustration for patients trying to understand their condition or searching for an accurate diagnosis. Furthermore, based on the circumstances in which they are used and the biological marker being evaluated, different genetic tests also provide different information for different purposes. Without adequate counseling, patients may conflate testing for inherited genetic risk with testing to inform the selection of a treatment option.

### ● **ROUNDTABLE SESSION III**

#### **Improving the Health Care System for Personalized Medicine**

Sessions I and II explored issues around the importance of education and communication between patients and health care providers and how access issues and disparities may limit the delivery of personalized medicine in the existing health care system. Session III considered the health system as a whole and how it must change to improve the delivery of personalized medicine for patients. Panelists represented a patient-focused research foundation generating large data sets to advance new personalized medicine treatments in cancer, a foundation focused on data privacy, and a nonprofit health insurance provider.

The COVID-19 pandemic has been disruptive to health care systems worldwide and many of the changes, such as an increased use of telemedicine, may become permanently entrenched in the way patients are cared for. Telemedicine, in particular, may be a critical part of personalized health care in the future.

To advance the development of new treatments for their disease, including personalized medicine treatment approaches, patient-led research foundations have identified obstacles to identifying and developing effective treatments for their disease and then adopted business models resembling virtual biotech companies to try to overcome these obstacles. Such efforts have included generating their own large, representative, longitudinal data sets with clinical, genomic, and immune system information to provide a robust understanding of the diversity of phenotypes and genotypes for their disease. These data have enabled, in some cases, the launching of clinical networks and platform trials.

Health care data are currently organized around many different entities and stored differently by health care systems, payers, research and clinical laboratories, and patients' personal devices. Since patients are particularly concerned about the collection and use of their own personalized health information, data should ideally be person-centered (i.e., reorganized around individual people and their relationships, with patients in charge of assigning access to different entities).

Health care systems and payers increasingly must address the clinical and economic value of personalized medicine. This requires determining for which patients a treatment is safe and effective, as well as the clinical and economic value of a treatment to patients and the health system. Patient perspectives and preferences should also have a role in determining

this value. Developing this evidence requires gathering data from a diverse set of patients that represents all populations affected by a disease, including minorities and women. Data systems should be improved so that this information can be shared among researchers, health care providers, payers and other health care decision-makers.

Moreover, ensuring the value and cost-effectiveness of any personalized treatment approach will only be possible if providers and patients are educated about available treatments as well as the testing that is required to select the optimal treatment for a patient. Patient advocates, advocacy organizations, and peer and patient navigators offer potential cost-effective strategies to help patients utilize and comprehend as fully as possible the vast amount of information they will encounter and to identify resources that can help them manage costs not covered by insurance. While physician expertise is essential, peer navigators may help patients gain access to these experts.

Peer navigators are especially important for marginalized communities that may not have access to the same resources available to others. Meanwhile, payers working with marginalized communities may be able to work with disease-specific advocacy organizations and peer navigators to ensure that patients have access to resources, systems, and treatment options that will lead to better health outcomes. For example, peer navigators may be able to help patients access not only genetic counseling and testing, but also social services, transportation services, and other assistance with out-of-pocket expenses.

Genetic testing can be expensive, although the costs are coming down considerably as the technology evolves. Strategies to ensure that genetic testing is cost-effective include reducing unnecessary testing and identifying technologies that can provide the most valuable information. For example, a test that assesses multiple genes may provide much more useful information at a cost that is the same as one that tests for a single gene. Genetic counselors are critical team members for interpreting these results, identifying and implementing strategies that ensure the use of high-value technologies, and educating patients and caregivers.

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4 In this case, “other” refers to a health insurance representative.

5 In this case, “other” refers to representatives from diagnostic, health insurance, and health information technology companies as well as a representative from the U.S. National Institutes of Health.

6 “Peer navigators” are individuals from a community who are often patients themselves and are trained to assist other patients diagnosed with the same health condition. Peer navigators can help educate newly diagnosed patients, connect patients with needed services, promote treatment adherence, foster trust in the individual’s health care, and provide interpersonal support. For example, see <https://www.cancommunityhealth.org/peer-navigators/>.

7 According to the Academy of Oncology Nurse and Patient Navigators, “nurse navigators” are clinically trained individuals responsible for guiding the patient through the care continuum including diagnosis and treatment. They can act as a central point of contact for a patient and coordinate components of care that involve physicians, social workers, patient education, community support, financial and insurance assistance, and others. See <https://aonnonline.org/faq>.

# Patient-Centered Research Agenda Advancing Personalized Medicine

The following research agenda identifies research topics that could improve the delivery of personalized medicine in ways that are most meaningful to patients.

## PATIENT-PROVIDER COMMUNICATION

1. How can education, communication, and health care delivery strategies **close the existing gap between a patient/caregiver's expectations** about personalized medicine and the reality of how and when test results and treatment options are currently delivered?
2. How can patients and caregivers be **encouraged to more actively participate** in their personalized treatment and in clinical research when they may prefer not to think about it, want to rely on their trusted medical team, or feel inadequate to participate?
3. How can conversations between a patient/caregiver and health care professional about personalized medicine treatment options best **address issues related to access and affordability of care**?
4. How can health care professionals and patients/caregivers **navigate language and cultural differences** when discussing personalized medicine treatment options or delivering/receiving care?
5. How can health care providers understand and engage in conversations about the unique concerns of patients/caregivers with diverse racial and ethnic backgrounds, especially **concerns stemming from historical underrepresentation or mistreatment** in medical research, when discussing personalized medicine treatment options?
6. What resources (e.g., checklists, decision aids or conversation scripts) and processes are needed to **facilitate shared decision-making between a patient/caregiver** and provider about personalized medicine treatment options?
7. How can shared decision-making facilitate conversations between patients and providers about **how personalized medicine treatment options align/do not align with a patient/caregiver's values and circumstances**?

## PATIENT EDUCATION

8. What kinds of **new and existing educational tools and resources**, including those using digital platforms, would empower patients/caregivers with varying levels of understanding about personalized medicine and its benefits to discuss personalized medicine treatment options with their health care provider?
9. How can the **coordination between patient advocacy organizations and providers** be improved to provide optimal resources for patients and caregivers?
10. How does educational material about personalized medicine, including general education materials and disease-specific **education materials, need to be reframed (not simply repackaged or translated)** for audiences with different cultural backgrounds, languages, and health literacy levels?
11. What kinds of **information do patients and their caregivers want to receive at different stages of their care pathway**, including pre-diagnosis, so that they are appropriately informed, but not over-burdened, by information about personalized medicine and potential treatment options?

## CAREGIVER, PEDIATRIC AND FAMILY CONSIDERATIONS

12. How should conversations between a health care provider and patient/caregiver **address health transitions throughout the patient's lifetime and disease progression**, including transitions between pediatric, adolescent, and adult care and eligibility for new personalized medicine treatment options that may emerge, so that the patient understands his/her evolving health needs and begins to independently navigate his/her own care?
13. How **can family and friend caregivers be better integrated** into the care team as partners in delivering personalized medicine and be better prepared to support a patient's decision-making process?
14. How can health care providers best **communicate with patients/caregivers the level of health risk** associated with a genetic test result and what this result does or does not mean for other family members, including children?
15. How can health care providers, other health professionals, peer educators, and patient advocacy organizations better **facilitate family communication about health history** and address family members' potential concerns, reservations, or taboos about discussing family health information?
16. How can patient education programs best address a family's concerns with **understanding new health information**, including the impact of a positive genetic test result, on other family members' potential genetic health risk and decisions to test further within the family?



## PROVIDER EDUCATION, RESOURCES AND COLLABORATION

17. What kinds of **educational and clinical decision-support tools, technologies, and resources would empower providers** in varying health settings, health disciplines, and health specialties, who may be under-educated about personalized medicine and its benefits, to discuss personalized medicine treatment and prevention strategies with patients/caregivers?
18. How can the **roles of other health care professionals beyond a patient's physician**, such as nurses, nurse navigators, genetic counselors, pharmacists, dietitians, physical/occupational therapists, psychologists, and social workers, be maximized to improve the delivery of personalized medicine?
19. How can **barriers that providers and their medical teams face in discussing and sharing information** on personalized medicine with patients and caregivers, including testing options, patients' results, treatment options, and enrollment in clinical trials, be addressed?
20. What strategies should be used to **educate primary care providers, physicians practicing in non-academic settings, and community health workers** on personalized medicine and how to integrate valid personalized medicine approaches into clinical care?
21. How can health care professionals, including genetic counselors, **select the most appropriate genetic test for a patient from among a number of competing tests** available from different laboratories/companies? How can providers best communicate with patients/caregivers limitations of these tests, including how their condition could still have a genetic driver even if they receive a negative test result?

## ACCESS, AFFORDABILITY AND UTILIZATION

22. How do a **patient's needs and access barriers, including those related to cost and affordability, change** depending on the purpose of a genetic test (e.g., to identify risk of developing a disease vs. to identify an appropriate treatment)?
23. How does **affordability and insurance coverage impact a patient's timely access** to novel and/or off-label personalized treatments?
24. What factors contribute to **underutilization of personalized therapies**? How must the health care system, including health insurance and coverage policies, change to address these factors as well as promote utilization in underserved populations?
25. How can **expanded coverage for patients/caregivers' basic needs**, such as transportation and childcare costs, promote utilization of personalized medicine in underserved populations?
26. What mitigating strategies are needed to **deliver personalized medicine treatment options to individuals with non-Western European ancestry/ethnicity**, where underrepresentation in research has created challenges in interpreting their genomic information?

## COVERAGE AND REIMBURSEMENT

27. How can personalized medicine **help employers and payers design benefit structures or create utilization management strategies** to allocate resources that are more streamlined, patient appropriate and based more on patients' heterogeneity?
28. How can the **use of billing codes and other payment incentives**, including those in the Medicare and Medicaid programs, be improved to encourage additional education and training of patients, caregivers, and family members about genetic testing, the meaning of the results, and the relevance of potential treatment options based on that information?
29. How would **coverage and reimbursement for genetic consultations** with trained genetic counselors and/or pharmacists about personalized treatment options and strategies improve the utilization of personalized medicine?
30. What **clinical utility studies, cost-effectiveness studies, and budget impact studies are needed** to inform payers about the value of covering the cost of a genetic test or genomic panels?
31. What data are needed to **establish consistent reimbursement policies** for genetic testing and to guide the selection of the most appropriate genetic test for a patient?
32. What evidence is needed, and where is there already adequate evidence, to support payers and other members of the health care community in collaborating to **develop value-based contracts for personalized treatments** that may have higher up-front costs?
33. How can **patient/caregiver perspectives**, including their preferences regarding personalized medicine treatment options, their decision-making criteria, and their successful adherence to treatment, factor into the "value" defined in value-based contracts?

## CLINICAL TRIALS

34. What changes are needed in the design and execution of clinical trials (including outreach, recruitment and funding) to **ensure representation of all populations** affected by a disease, including minority populations and women?
35. How can health care providers **build trust with patients/caregivers** and patient communities from diverse backgrounds to address their concerns and fear of mistreatment, including misuse of their health information, when deciding on personalized medicine treatment options, including whether or not to enroll in a clinical trial?

## NEW TECHNOLOGIES AND DATA MANAGEMENT

36. How can the **informed consent process for research participation** be improved to account for evolutions in technology, such as artificial intelligence and digital health technologies, and establish trust with patients/caregivers over the use of their data in health research?
37. How can **data sharing between repositories of real-world data** (e.g., electronic health records, registries, etc.) be improved to increase transparency and facilitate shared, dynamic learning about personalized medicine utilization and treatment outcomes?
38. How can **real-world data be leveraged to support health care providers** in selecting the most appropriate personalized treatment option for an individual patient?
39. How can **telemedicine be effectively used** for the continuing education of patients/caregivers about their genetic information, personalized medicine and relevant treatment options?

## OUTCOMES RESEARCH

40. How can outcomes research in personalized medicine **measure the impact of treatment on a patient's financial health, psychosocial well-being, and experience** receiving care?
41. How does a patient/caregiver's and a health care provider's **understanding of genomics**, including the patient's understanding of his/her own genetic information, impact the integration of personalized medicine into clinical care?
42. How can outcomes research on genetic testing improve patients/caregivers' **understanding of the relative risk associated with their diagnosis** and the potential benefit of pursuing a more aggressive treatment?
43. How does **involving peer navigators, nurse navigators and/or community health workers in care pathways** impact patient outcomes with personalized medicine? How can they be trained and supported, particularly in under-served communities, to improve the delivery of personalized medicine?
44. How does **family and friend caregiver involvement in decision-making and long-term care** impact patients' outcomes with personalized medicine? Which activities should caregivers be involved in to improve care, and how can caregivers be prepared to participate in these activities?
45. How can **longitudinal outcomes research** track the value of personalized medicine in preventing the onset of a disease or condition and in optimizing patient outcomes?

# Conclusion

PMC's interactions with patients over the past two years revealed that many patients and caregivers simply do not know what to ask, when, and to whom during the diagnosis and treatment decision-making process in order to receive personalized medicine. Barriers to provider education and training, to integrating personalized medicine across clinical settings and health provider specialties, and other access challenges related to affordability and health insurance coverage compound the education and awareness challenges for patients.

Through the PCORI-funded web forums and roundtable sessions, patients, caregivers, health care professionals and researchers learned about the challenges facing each other and ultimately developed a shared understanding of meaningful, patient-centered research questions that could improve the delivery of personalized medicine. When discussing personalized medicine treatment options, patients want the decision-making process to be informed by their values and circumstances as much as their genetic information, and they want family and caregiver considerations, financial concerns, and reservations about participating in clinical research to be a part of that conversation as well. All of these considerations will differ depending on the disease area, treatment options available and type of genetic information being used to inform the treatment decision. Health care professionals must listen to their patients and have more nuanced conversations, but they are facing their own barriers as well.

The health care community must shift away from a one-size-fits all paradigm to a system accounting for a patient's unique needs and priorities. In order to improve the quality and efficiency of care, the health care system, including billing and reimbursement structures and data systems, must improve to create opportunities for continued patient and caregiver education about genomics and personalized medicine treatment options in varying health care settings. Health care professionals and researchers must also focus on being transparent and building trust to address biases and inequities facing minority populations. These changes will help empower patients to make informed decisions about their care and ultimately benefit from personalized medicine.

This patient-centered research agenda should inform future studies that will provide patients, caregivers, and health care professionals with the information they need to make more informed health care decisions and, ultimately, improve the delivery of personalized medicine to patients in ways most meaningful to them. PCORI, public-private partnerships, private foundations and other entities interested in all patients receiving personalized medicine should fund these studies.

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Finally, PMC is grateful for the engagement and continued support of the project's **Advisory Committee**, as well as the numerous patients, patient advocates, caregivers, health care professionals, researchers and other stakeholders who have contributed to developing this research agenda over the past two years, including the **roundtable participants** (see list on opposite page). We are proud to be able to convene so many different voices and to build a shared understanding of patients' priorities for improving the delivery of personalized medicine.

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# About Us

## **Personalized Medicine Coalition (PMC)**

The Personalized Medicine Coalition convenes over 230 organizations representing innovators, scientists, patients, providers and payers to promote the understanding and adoption of personalized medicine concepts, services, and products to benefit patients and the health system. For more information about PMC, please visit [www.personalizedmedicinecoalition.org](http://www.personalizedmedicinecoalition.org).

## **Patient-Centered Outcomes Research Institute (PCORI)**

The Patient-Centered Outcomes Research Institute® (PCORI®) is an independent, nonprofit organization authorized by Congress in 2010. Its mission is to fund research that will provide patients, their caregivers, and clinicians with the evidence-based information needed to make better informed health care decisions. PCORI is committed to continually seeking input from a broad range of stakeholders to guide its work.





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