A Research Program for Advancing Personalized Medicine in 2022

An Overview of Initiatives to Evaluate Progress in Health Care, Assess Clinical and Economic Value, and Examine Clinical Integration Strategies

PMC
PERSONALIZED MEDICINE COALITION

President/Chairman's Letter

Dear Colleague:

Personalized medicine — linking therapies to molecular diagnostic tests to ensure that the right treatments are targeted to the right patients — is still more aspiration than reality.

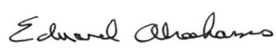
Although we have made enormous progress in developing a new scientific appreciation of human heterogeneity, as evidenced by the rise in the number of personalized treatments on the market from five in 2008 to more than 300 today, significant obstacles remain.

In brief, we need more evidence that personalized medicine works — that it can improve clinical outcomes while making health care more efficient and therefore less costly. We also need to understand the opportunities and challenges shaping the pace of our progress toward integrating personalized medicine into clinical workstreams.

This three-part *Research Program for Advancing Personalized Medicine in 2022* is designed to facilitate progress on each of these fronts. The projects presented in these pages focus on evaluating technological progress in personalized medicine, assessing its clinical and economic value, and examining clinical integration efforts. We have made considerable progress in advancing many of these studies, but others are not fully funded.

We therefore request your thoughtful consideration of these initiatives as we call on organizations from multiple sectors of the health care ecosystem to provide support, in addition to membership dues, for the Personalized Medicine Coalition's expanding research portfolio.

Sincerely yours,



Edward Abrahams
President

Jay G. Wohlgemuth, M.D.
Chairman, Board of Directors

Program Overview

AS OF JANUARY 2022

EVALUATING PROGRESS IN HEALTH CARE

ASSESSING CLINICAL AND ECONOMIC VALUE

EXAMINING CLINICAL INTEGRATION STRATEGIES

STUDIES 1–3

Evaluating Progress in Health Care

The first three studies included in this research program are designed to outline the trends and opportunities associated with personalized medicine. Focusing on genomic testing and pharmacogenomics, the first two studies will assess how the field is advancing and what more needs to be done. The third study will survey a representative sample of payer representatives to better understand payer decision-making and evaluate how coverage policies shape the development of personalized health care.

STUDY 1

Understanding Genomic Testing Utilization and Coverage in the US

BACKGROUND

Personalized medicine rests partly on the assumption that genomic testing can aid in treatment management decisions, yielding both clinical utility and economic value. Genomic testing has the potential to improve clinical care by providing important information that can speed diagnosis, inform treatment decisions, and improve the efficiency of health care delivery in several areas, such as prenatal screening, oncology, and rare and undiagnosed disease. However, genomic testing technologies are relatively new, and providers face several barriers to the adoption of policies and procedures that will lead to widespread access of genomic testing in clinical practice. A better understanding of the utilization of genomic testing across the U.S. health system and how it relates to patient access will provide insight into genomic testing implementation trends and barriers.

OBJECTIVE

This study will examine the patterns of utilization of genomic sequencing for noninvasive prenatal testing, tumor diagnosis, and rare and undiagnosed disease in the U.S. Utilization will likely be related to a patient's distance to testing facilities, insurance coverage, and social/environmental impacts on access to testing. The results of this study's analysis will be included in a report whose purpose will be to provide key personalized medicine stakeholders, including providers, pharmaceutical and diagnostic manufacturers, payers, patients, and lawmakers with insight into genomic testing implementation and barriers so as to inform efforts to integrate and deliver personalized medicine to all patients who can benefit.

PROJECT OUTLINE

Using a U.S. clinical genetic testing market database cross-referenced to claims data, the project will involve examination of current utilization patterns of genomic sequencing-based testing as related to access factors, including coverage, distance to genomic testing facility, and social and patient-level determinants of health care.

PMC, Illumina, Concert Genetics, and the Blue Cross Blue Shield Association will work together to coordinate data collection, analysis, and development of reports.

Results of the study will be published in three separate documents:

1. A two-page analysis preview;
2. A PMC white paper detailing the findings; and
3. A manuscript focusing on implications to the integration of personalized medicine in health care to be published in a peer-reviewed journal.

The report and manuscript will be disseminated through a robust communications plan to increase awareness and become a resource for health care decision-makers.

STUDY 2

Pharmacogenomics in Clinical Guidelines and at FDA

BACKGROUND

Pharmacogenomics (PGx) can play an important role in identifying responders and non-responders to medications, avoiding adverse events, and optimizing drug dosing. The use of diagnostic tests to detect PGx gene-drug interactions is a cornerstone of personalized medicine, providing a way to guide treatment and prevention strategies based on individual patient characteristics. Despite varying levels of evidence supporting the clinical utility of many PGx markers, only a select few are recommended for use in informing clinical decisions. Because of this variability in supporting evidence, PGx clinical practice guidelines formulated by panels of experts have a significant impact on the pace at which providers integrate new PGx tests and other novel technologies into their clinical work streams.

A better understanding of the current status of PGx gene-drug association inclusion in the clinical practice guidelines developed by major guideline development bodies will help clarify the extent to which personalized medicine technologies have been integrated into clinical care. An examination of biomarkers included in guidelines can also highlight areas where clinical evidence of utility is being developed but where widely recognized biomarker-based prevention and treatment strategies have not yet been included in clinical guidelines or within drug labels. This, in turn, can inform further efforts to advance personalized medicine implementation.

OBJECTIVES

PMC will catalogue the PGx-based gene-drug associations that are included in current guidelines for different health conditions and compare this to PGx associations included in drug labels. The results will be included in a report whose purpose will be to provide key personalized medicine stakeholders, including product developers, clinical laboratories, health care providers, payers, patients and policymakers with information about what PGx strategies are commonly used in practice and what strategies may require additional evidence development or provider outreach and education efforts.

PROJECT OUTLINE

The project will involve a comprehensive examination of the current clinical practice guidelines for PGx published by major guideline development organizations, including but not limited to those published by CPIC, and subsequent cataloging of included gene-drug interactions.

The clinical guideline PGx catalogue will be compared directly to PGx information listed in FDA's *Table of Biomarkers in Drug Labeling* and through a direct review of existing drug labels.

Drug-gene interactions for which there is inconsistency between inclusion in FDA labeling and clinical guidelines will be highlighted.

The results will be published in a white paper detailing the findings and policy implications.

PMC will engage with guideline development organizations and FDA to have them advise the research strategy and review results.

STUDY 3

Payer Perspectives and Policies on Personalized Medicine: A Landscape Analysis

BACKGROUND

If the health care system is to secure the full benefits of personalized medicine, it must provide appropriate access to technologies, products, and services that make getting the best treatment option to each patient possible based on individual characteristics and circumstances. To help ensure equitable access, the reimbursement system must have evidence-based coverage and payment policies that support the timely adoption of validated personalized medicine technologies, including both diagnostics and therapeutics.

Payers are increasingly considering coverage and reimbursement of personalized medicine products and services both in the U.S. and internationally. However, there remain significant challenges in establishing coverage policies and payment rates for diagnostic tests and targeted therapeutics that reflect the value of their care. As a result, many novel diagnostics are under-reimbursed or not reimbursed at all, and targeted therapies are often subject to utilization management practices such as step therapy or prior authorization. Such practices ultimately restrict patient access to needed tests and optimal care. Coverage and reimbursement policies vary widely among different payers and laboratory benefits managers, and decision-making processes are often inconsistent and not transparent. Thus, it is not clear whether the broader payer community supports the move towards personalized medicine.

A better understanding of how payers view personalized medicine will help clarify the extent to which it has informed payer policies and perspectives globally and help identify remaining needs, which, in turn, will help product developers understand the payer landscape and the challenges they face.

OBJECTIVE

This project will involve an examination of varying policies and perspectives to capture a global picture of the coverage and reimbursement of personalized medicine services and technologies by public and private payers. The landscape analysis will include a representative sample of public and private health insurers, as well as large employers and laboratory benefit managers, and will include both quantitative and qualitative results to ensure that a global health system-wide payment picture of personalized medicine is captured. The results will be included in a report whose purpose will be to provide key personalized medicine stakeholders, including product developers, clinical laboratories, health care providers, payers, patients, and policymakers, with information about access to personalized medicine technologies as related to the payment environment, thus informing payer-related strategies that may lead to improved access to personalized medicine.

PROJECT OUTLINE

The project, to be completed in six months, will involve the development of a framework to be used to measure personalized medicine coverage and reimbursement levels by government-run and private health insurers, as well as large employers.

The framework will be based on publicly available data showing payer coverage and utilization management policies, as well as payment details. It will also include survey data highlighting payer perspectives, partnerships, and practices related to personalized medicine.

To facilitate a holistic view of the landscape of personalized medicine coverage and reimbursement, the analysis will focus on a representative sample of public and private payers and payment manager organizations, including large and small private health insurers; national and state-run public health plans; large employers with managed employee health benefits; and laboratory benefit manager organizations.

A project steering committee consisting of PMC members across payer, provider, industry, and patient stakeholder groups will guide the project, review survey questions, track progress at various milestones, and ensure an appropriate sample of survey respondents.

CONCLUSION

Many organizations within the personalized medicine community have called for an analysis of the current landscape of payer perspectives and policies on personalized medicine in the United States and internationally. This proposal is PMC's answer to that call.

STUDIES 4–5

Assessing Clinical and Economic Value

Projects four and five are designed to assess the clinical and economic value of personalized medicine in clinical care. By examining the clinical and economic benefits of personalized medicine for patients with suspected rare diseases and by relating overall improvements in clinical care to the implementation of personalized medicine, these studies will advance peer-reviewed evidence necessary to help payers and providers make decisions about the circumstances in which they should adopt policies and programs that provide access to the tests and treatments underpinning personalized medicine.

STUDY 4

Evaluating the Clinical and Economic Value of Sequencing-Based Diagnostic Tests for Patients With Rare and Undiagnosed Diseases

BACKGROUND

For patients with rare and undiagnosed diseases, genomic sequencing — determining a patient's entire unique DNA makeup — may be an extremely valuable tool for discovering the genetic alterations that contribute to disease development, as well as for influencing treatment decision-making. Patients with rare and undiagnosed diseases are most often children who may have already undergone single-gene testing but still have not received a definitive diagnosis. They are sometimes referred to as "diagnostic odyssey" cases, as a child and family can bounce around various medical centers for years while numerous diagnostic procedures are performed and health care costs are accrued. All the while, the disease goes undiagnosed.

As they go through their diagnostic odysseys, these patients' diseases progress and they lose time during which they could potentially receive effective therapies or be enrolled in a clinical trial.

OBJECTIVE

PMC organized a project planning committee to develop a request for proposals for a study that would demonstrate the clinical and economic value of sequencing for rare and undiagnosed diseases. A research proposal developed by Peter Neumann, Sc.D., at the Institute for Clinical Research and Health Policy Studies at Tufts Medical Center was selected. The study will address the primary hypothesis as stated in the proposal: "Genomic sequencing testing can speed diagnosis and initiation of appropriate care and can be cost-effective when performed early on in clinical course."

Interested partners will be included on a project steering committee that will help guide the project, review progress at various milestones, and ensure the use of appropriate and up-to-date data. The study will lead to a peer-reviewed publication whose purpose will be to provide evidence for sequencing platform developers, payers, and providers that sequencing-based diagnostic testing is both clinically useful and economically efficient when applied to the right patient population.

RESEARCH OUTLINE

The project, to be completed in 6–9 months, will involve the development of a clinical and economic value model validated with existing real-world data from institutions that are regularly sequencing patients (and in some cases, the child and both parents) with diseases of unknown etiology.

The project may also involve a value of information analysis and the identification of factors that significantly impact the cost-effectiveness of sequencing-based diagnostic testing.

Research will utilize existing data sets and/or ongoing studies to the extent possible — supplemented by the collection of data from health system and medical records.

As the convener of the study, PMC will coordinate a meeting with the research team, steering committee members and other partners to determine what data sets should be explored for use in the study and how we will gain access to these data sets.

The project's aim is to assess the value of sequencing rare and undiagnosed disease patients in general. The study will not compare technology platforms.

A payer and employer advisory committee to include health insurers, large employers, and employer benefits management groups will also be formed to provide guidance, especially related to cost/benefit assumptions utilized in the value model, so the project results will incorporate the perspectives of these key stakeholders, increasing the likelihood that the results will be useful in informing the coverage and reimbursement process. Results of the study will be published in a peer-reviewed journal and disseminated through a robust communications plan to increase awareness and become a resource for decision-makers in government, industry and health care.

CONCLUSION

Many organizations within the personalized medicine community have called for evidence demonstrating the value of genomic sequencing in rare and undiagnosed disease. This project is PMC's answer to that call.

STUDY 5

Improvements in Clinical Care Associated With Personalized Medicine

BACKGROUND

Integrating personalized medicine into health care delivery systems requires a shift from traditional "one-size-fits-all" practices to a more efficient approach based on understanding and addressing individual patient characteristics and circumstances, therefore requiring an investment in workforce and financial resources associated with new technologies, policies, practices and education. As providers consider integrating personalized medicine approaches into their health care delivery systems, they increasingly want evidence showing that the implementation of personalized medicine improves clinical care.

To gain a better understanding of the current landscape of personalized medicine integration, PMC coordinated a study to capture a holistic picture of the complete range of personalized medicine implementation strategies and technologies being undertaken at a representative sample of health care delivery institutions across the United States. Based on a quantitative framework that assesses progress toward personalized medicine integration at the institutional level with consideration of multiple clinical areas, institutions were assigned scores between one (minimal personalized medicine integration) and five (expansive and systematic integration). The findings show that U.S. health care organizations are widely distributed in terms of integration across clinical areas, but most are at level two or level three.

Level four and level five institutions stand out as pioneers in personalized medicine but are still not common. While many of these institutions are thought of as outstanding health systems, there is a need for evidence linking the personalized medicine approach to improved clinical care. A better understanding of the level of integration of personalized medicine associated with improved health care delivery and clinical outcomes may help justify the adoption of policy changes and investments associated with its implementation.

OBJECTIVE

This project will examine the effect of integrating personalized medicine approaches in driving improved clinical care and systemic efficiency. The extent to which a health care delivery institution has integrated personalized medicine will be based on integration scores as determined through PMC's *Integration of Personalized Medicine in U.S. Health Systems* landscape analysis. Improvements in clinical care and systemic efficiency will be considered as part of a matrix of elements contributing to the increased delivery of high-quality health care practices and services that can improve clinical outcomes and/or provide greater access to relatively higher value care, and will include both quantitative and qualitative measures.

The results will be included in a report whose purpose will be to provide key personalized medicine stakeholders — including providers, payers, clinical guideline developers, pharmaceutical and diagnostic manufacturers, patients and policymakers — with evidence of the value of integrating personalized medicine into clinical practice. This, in turn, can help inform efforts to address the most critical outstanding integration challenges.

PROJECT OUTLINE

The project, to be completed in 12 months, will involve the development of an evidence-based framework linking personalized medicine integration scores with improved clinical care, along with subsequent analysis that will help demonstrate the current value of personalized medicine integration.

A project steering committee composed of partner members of PMC's Health Care Working Group will guide the development of the value framework with the inclusion of appropriate evidence; track progress at various milestones; and ensure that results are meaningful to key audiences.

The analysis will include examination of a representative sample of U.S. health care delivery institutions including academic health centers; urban, suburban, and rural community hospital systems; and integrated payer/provider systems.

A payer advisory committee will also be formed to provide guidance especially related to assumptions about improved clinical care utilized in the value framework. The advisory committee will help ensure that the project results incorporate the perspectives of these key stakeholders, increasing the likelihood that the results will be useful in informing coverage and reimbursement processes.

Results of the study will be published in a peer-reviewed journal and become a resource for decision-makers in health care delivery, industry, and government.

CONCLUSION

The personalized medicine community needs an evidence-based analysis linking the implementation of personalized medicine to improved clinical care to help advance personalized medicine integration efforts and to make clear to health care decision-makers that they should develop policies and practices that drive personalized medicine. *Improvements in Clinical Care Associated With Personalized Medicine* is designed to address this need.

STUDIES 6–9

Examining Clinical Integration Strategies

The final four projects are designed to understand and address the challenges that health care providers face when integrating the principles of personalized medicine into their clinical workstreams. The studies are focused on prompting the more widespread clinical adoption of genomic testing technologies by highlighting underappreciated benefits of testing; examining the barriers that are discouraging the clinical integration of genetic testing and the appropriate utilization of testing results; and addressing equity and data management challenges that are inhibiting progress in health care and personalized medicine.

STUDY 6

Defining the Clinical Utility of Genomic Testing in Cancer Care

BACKGROUND

The use of advanced diagnostic tests to detect predictive and prognostic biomarkers is a cornerstone of personalized medicine, providing a way to guide treatment and prevention strategies based on individual patient characteristics. However, despite varying levels of evidence supporting the clinical utility of many biomarker-based strategies, advanced diagnostic testing is often not appropriately or effectively used in clinical practice. The consistent use of biomarker testing faces implementation challenges related to insufficient awareness and education, a lack of recognition by payers and providers of the clinical value of testing, and outdated practices, policies and processes. A firm understanding among all health care stakeholders of all the ways in which biomarker testing has clinical utility will be instrumental to overcoming implementation challenges and providing the most efficient and effective personalized health care.

Developing this expanded definition of clinical utility will require a better understanding of current provider and payer perspectives on biomarker testing along with a comprehensive examination of how testing strategies can benefit patient care. This, in turn, can inform further efforts to address challenges associated with personalized medicine implementation.

OBJECTIVES

PMC will examine current perspectives on the use of personalized medicine strategies in health care delivery and identify all of the factors that can help determine the clinical utility of multiplex genomic testing in clinical practice, such as for use in patient screening, prognosis, treatment decisions, clinical trial recruitment, and to inform potential off-label uses. Based on these findings, PMC will develop recommendations to broaden the definition of the clinical utility of multiplex genomic

testing in patient care. The recommendations will be made to key personalized medicine stakeholders, including health care providers, payers, clinical guideline developers, clinical laboratories, and patients, to help inform their policies and processes and support their educational efforts.

PROJECT OUTLINE

The project will involve examination of current perspectives on the use of multiplex genomic testing and the subsequent development of recommendations to key personalized medicine stakeholders for the inclusion of a comprehensive set of value factors within a broadened definition of clinical utility.

Phase 1: Stakeholder Roundtable Event

a. Objectives:
   i. Bring together health care providers and payers to identify the components of what an expanded definition of clinical utility could and should consist of
   ii. Align on recommendations for how expanding the definition of next-generation sequencing clinical utility can be implemented in the community to enhance patient care
      › To do this, PMC envisions the experts discussing their clinical practice (case studies), responding to pre-prepared topics/questions related to utility factors, and describing how their practical experience could inform the development of recommendations on an expanded definition of clinical utility

b. Format:
   i. Moderated roundtable discussion to examine case study examples and respond to pre-prepared topic points related to different elements of the clinical utility of advanced genetic tests
   ii. Roundtable participants will discuss how recognition of clinical utility can help address practice gaps and develop recommendations on how to broaden the perception of utility and value amongst providers

c. Who:
   i. 10–20 experts
      › 4 academics (representing multiple tumor types)
      › 6 community (mix of hospital system and stand-alone practice)
      › 2–3 payers (1–2 attached to practice also in the room — commercial, CMS)
      › 2–3 molecular pathologists/laboratory directors
      › Representatives from AACR (GENIE)/ASCO/NIH (All of Us and NCI Lung-MAP)

d. Pre-meeting materials may include:
   i. Prepared document with discussion topics, considerations, and potential suggestions for review by roundtable participants
   ii. PowerPoint presentation for roundtable discussion
   iii. Press release
   iv. Blog, website and social media content
   v. Email to membership and other identified partners

Phase 2: Report/Recommendations

a. Value data
b. Clinical recommendations
c. Operational recommendations
d. Influence on guideline development bodies (such as NCCN) and payers
e. Willingness of attendees to kick off studies at their institution
f. Request attendees to advocate for this topic during future speaking engagements

CONCLUSION

Many organizations within the personalized medicine community have emphasized the importance of driving a better understanding of the clinical utility of genomic profiling in cancer care amongst all stakeholders, including payers and providers. This project is PMC's answer to that call.

STUDY 7

Addressing Challenges in Using Health Data to Advance Personalized Medicine

BACKGROUND

Recognizing that genetic test results and other biomarker information, diagnostic images, and information about individual patient circumstances are increasingly being generated and utilized to improve patient care and advance research in personalized medicine, proponents for personalized medicine have begun to envision a data-driven era in which health care is tailored more closely to the biological and environmental factors influencing each patient's health and well-being. Because the success of these endeavors will depend on patients themselves to report, generate, and consent to the use of their health care data, strategies for data-driven personalized medicine must also capture the imagination of the American public on topics related to the future of health and health care, examining societal values and ethics and demystifying the technical and engineering aspects of data and digital health. Foundational work is therefore needed to frame the patient experience as one made better by data and technology while supporting the development of scalable and expandable data infrastructure and analytical methods that can help health care providers organize and systematically access knowledge about each individual patient.

OBJECTIVE

Recognizing the opportunities availed by the incoming Presidential administration and a new Congress in the United States, *Addressing Challenges in Using Health Data to Advance Personalized Medicine* will identify, develop, and advocate for regulatory and policy solutions that encourage the responsible use of health care data to advance patient-centric drug development, personalized medicine, and a learning health care system. Proposed solutions may be designed to:

- Encourage the re-use of electronic health record data;
- Protect patient privacy while encouraging innovation in health care; and
- Establish data standards.

PROJECT OUTLINE

Innovation Horizons, a health care and data analytics consultancy group, will conduct in-depth surveys and interviews to gain perspectives on the topics that will be used to shape concepts and recommendations geared toward addressing future policy issues in the one- to five-year time horizon. From the assembly of interviews and PMC reference materials, a white paper and presentation documents will be prepared, reviewed, and published.

STUDY 8

Addressing Practice Gaps in the Implementation of Personalized Medicine in Cancer Care

BACKGROUND

Predictive biomarker testing to help identify patients who could benefit from targeted therapies is a cornerstone of personalized medicine in cancer care, informing treatment decisions that can lead to better patient outcomes and improve the efficiency of the health care system. Predictive biomarker testing technologies are relatively new, however, and providers face several barriers as they seek to integrate the tests into clinical work streams. For example, although more than 30 percent of patients with non-small cell lung cancer (NSCLC) have tumors that are linked to genetic driver mutations, many NSCLC patients do not receive genomic testing. Furthermore, a recent Personalized Medicine Coalition report has estimated that only 65–75 percent of NSCLC patients with an actionable mutation as determined by genomic testing actually receive targeted therapies.

A recent analysis by Diaceutics estimated that more than 30 percent of cancer patients in the U.S. never receive targeted therapies that they may have benefitted from. The practice gap between patients that are eligible for targeted therapies and those that receive them is attributed, in part, to:

1. Limited access or availability of predictive biomarker tests;
2. Sample processing constraints;
3. Test performance;
4. Challenges related to reporting and interpretation of test results;
5. Limited access to targeted therapies; and
6. Lagging awareness of the rapidly evolving field of personalized medicine.

In order to help optimize the clinical use of genomic testing in cancer care and to accelerate the clinical adoption of personalized strategies, a better understanding of the associated clinical practice gaps is necessary.

OBJECTIVE

This project will utilize the Diaceutics Global Diagnostic Index (GDI) database to examine the practice gaps associated with biomarker testing-informed personalized medicine strategies in NSCLC care. The study will draw conclusions about specific practice challenges, including those related to test access and availability, sample processing, test performance, test interpretation, and utilization of results. The study will also estimate the impact that each of these factors has on the delivery of personalized cancer care. Implementation challenges will be considered as part of a matrix of elements contributing to the overall biomarker testing practice gap. The final report will offer testing optimization recommendations.

The results will be included in a report whose purpose will be to provide key personalized medicine stakeholders, including providers, payers, clinical guideline developers, pharmaceutical and diagnostic manufacturers, and patients with insight into how biomarker testing is being implemented throughout the health care system and the impact of several potential implementation barriers. These insights can inform efforts to optimize predictive biomarker testing in clinical practice and therefore help drive the delivery of personalized medicine to all cancer patients who can benefit.

PROJECT OUTLINE

The project, to be completed in 6–9 months, will involve the development of a matrix of practice elements contributing to sub-optimal use of personalized medicine strategies in cancer care and will estimate the impact of each practice element toward the overall clinical practice gap.

Research will utilize existing data sets from the GDI database, which includes practice-level data for approximately 260,000 NSCLC patients who were diagnosed between October 2018 through September 2019, showing their testing rates, test performance, and treating habits.

A project steering committee will help guide matrix design and assure that results are meaningful to key audiences.

The project's aim is to assess the practice gaps related to biomarker testing and targeted therapy in NSCLC in general. The study will not compare technology platforms.

Based on the analysis, PMC will make clinical implementation recommendations, which will be published as a white paper alongside the results.

STUDY 9

Addressing Disparities in Research Advancing Personalized Medicine

BACKGROUND

In order for personalized medicine to be universally applicable, research and development of new treatment and prevention strategies that are tailored to the biological characteristics and circumstances of individuals must include data from all types of people, thereby capturing the diversity of patients represented within the population. However, racial and ethnic minorities, the elderly, women, and patients from disadvantaged socioeconomic groups are often underrepresented in clinical trials. The problem persists even in disease areas characterized by elevated disease incidence and mortality counts among understudied patient populations.

As electronic health records (EHRs) and other digital technologies become ubiquitous in health care settings, scientists are also increasingly conducting practice-based research and machine learning studies to help inform personalized medicine implementation strategies and policy development. Little is known about how the use of these data sources to examine clinical utility, utilization and access challenges associated with various biomedical interventions compares to more traditional research strategies, and some researchers, patients and policymakers worry that an overreliance on data built with underlying health care inequities has the potential to further exclude patients from underrepresented populations.

Health research data can fail to reflect the demographic characteristics of populations being studied due to patient mistrust and transparency concerns, outreach and education barriers, cultural barriers, overly restrictive study design eligibility criteria, cost concerns/disproportionate representation among the uninsured, language/linguistic/literacy challenges, and practical obstacles such as proximity or access to health care facilities.

The advancement of personalized medicine underlines the importance of overcoming health disparities because it anticipates targeted treatments based on a patient's individual characteristics and health needs, including variables such as the patient's race, ethnicity, age, sex, and socioeconomic status. This relies, however, on an inclusive and equitable representation of patients with these diverse characteristics and health needs in personalized medicine research and development. A better understanding of how sociocultural, behavioral, and health system factors converge and contribute to unequal participation in clinical research and differential representation in research data is therefore needed to help ensure that personalized medicine scientific advances are impactful to all patients.

OBJECTIVE

This initiative will bring together key stakeholders representing public, private, and nonprofit organizations that are working with or are part of communities that are underrepresented in health care research to comprehensively address disparities in clinical trials participation and a lack of racial, ethnic, demographic and socioeconomic equity in health data used to guide research and development as well as the delivery of personalized medicine technologies. The findings will be included in a report whose purpose will be to provide key personalized medicine stakeholders, including clinical and health outcomes researchers, information management and digital health technologies organizations, providers, pharmaceutical and diagnostic manufacturers, payers, patients, and lawmakers with insight into barriers to equitable personalized medicine research. The report will provide recommendations on how to ensure that innovations underpinning personalized medicine are accessible to all patients.

PROJECT OUTLINE

This project will involve identifying a health equity task force consisting of key stakeholders, partners, and experts that have knowledge of health research data and are working with or are part of communities that are underrepresented in health care research. A series of interviews with the task force will be conducted to elucidate inequities within health data related to current clinical research, practice, and community access. The interviews will help define barriers to the development and use of representative health data for personalized medicine research and development.

PMC, working with this health equity task force, will then conduct a comprehensive policy review, to include: an examination of trial protocols, patient engagement approaches, existing strategies to address health care data disparities, and coverage, reimbursement, and utilization policies that can lead to inequalities in access to health care. The policy review will also examine the different impacts of research practices and policies on different underrepresented populations.

Based on the policy review, a set of draft recommendations to help increase the proportion of underrepresented groups in health research and appropriately account for diverse population demographics data will be developed.

Patients and community leaders will be surveyed to get their feedback on the draft recommendations and to gauge community perceptions of their potential impact and feasibility.

A report will be published with a set of final recommendations based on the community feedback and rankings.

The project will involve a qualitative analysis of inequities in various forms of data used for health research, including data from clinical trials, practice-based research studies, genetic and disease-specific databases, pooled electronic health records, health insurance claims databases, and population data used in machine learning algorithms.

MISSION

The Personalized Medicine Coalition (PMC), representing innovators, scientists, patients, providers, and payers, promotes the understanding and adoption of personalized medicine concepts, services, and products to benefit patients and health systems.

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