Help Me Understand Genetics

Precision Medicine


Lister Hill National Center for Biomedical Communications
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What is precision medicine?

According to the Precision Medicine Initiative, precision medicine is "an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person." This approach will allow doctors and researchers to predict more accurately which treatment and prevention strategies for a particular disease will work in which groups of people. It is in contrast to a one-size-fits-all approach, in which disease treatment and prevention strategies are developed for the average person, with less consideration for the differences between individuals.

Although the term "precision medicine" is relatively new, the concept has been a part of healthcare for many years. For example, a person who needs a blood transfusion is not given blood from a randomly selected donor; instead, the donor’s blood type is matched to the recipient to reduce the risk of complications. Although examples can be found in several areas of medicine, the role of precision medicine in day-to-day healthcare is relatively limited. Researchers hope that this approach will expand to many areas of health and healthcare in coming years.

Learn more about precision medicine:


Information about Genetics Home Reference and precision medicine is available from the interactive Journal of Medical Research (i-JMR) article, "Information Needs in the Precision Medicine Era: How Genetics Home Reference Can Help (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4917728/)."


Information about precision medicine (http://learn.genetics.utah.edu/content/precision/) is also available from the Genetic Science Learning Center at the University of Utah.
What is the difference between precision medicine and personalized medicine? What about pharmacogenomics?

There is a lot of overlap between the terms "precision medicine" and "personalized medicine." According to the National Research Council, "personalized medicine" is an older term with a meaning similar to "precision medicine." However, there was concern that the word "personalized" could be misinterpreted to imply that treatments and preventions are being developed uniquely for each individual; in precision medicine, the focus is on identifying which approaches will be effective for which patients based on genetic, environmental, and lifestyle factors. The Council therefore preferred the term "precision medicine" to "personalized medicine." However, some people still use the two terms interchangeably.

Pharmacogenomics (https://ghr.nlm.nih.gov/primer/genomicresearch/pharmacogenomics) is a part of precision medicine. Pharmacogenomics is the study of how genes affect a person's response to particular drugs. This relatively new field combines pharmacology (the science of drugs) and genomics (the study of genes and their functions) to develop effective, safe medications and doses that are tailored to variations in a person's genes.

Read more about precision medicine, personalized medicine, and pharmacogenomics:

A 2011 report from the National Research Council (http://www.plengegen.com/wp-content/uploads/4_Toward-Precision-Medicine.pdf) provides a detailed overview of precision medicine, including the reasoning behind the Council’s preference for the term "precision medicine" over "personalized medicine."

The journal Nature Medicine published an article on this topic called "Momentum grows to make 'personalized' medicine more 'precise'" (http://www.nature.com/nm/journal/v19/n3/full/nm0313-249.html).

What is the Precision Medicine Initiative?

The Precision Medicine Initiative is a long-term research endeavor, involving the National Institutes of Health (NIH) and multiple other research centers, which aims to understand how a person's genetics, environment, and lifestyle can help determine the best approach to prevent or treat disease.

The Precision Medicine Initiative has both short-term and long-term goals. The short-term goals involve expanding precision medicine in the area of cancer research. Researchers at the National Cancer Institute (NCI) hope to use an increased knowledge of the genetics and biology of cancer to find new, more effective treatments for various forms of this disease. The long-term goals of the Precision Medicine Initiative focus on bringing precision medicine to all areas of health and healthcare on a large scale. To this end, the NIH plans to launch a study, known as the All of Us Research Program, which involves a group (cohort) of at least 1 million volunteers from around the United States. Participants will provide genetic data, biological samples, and other information about their health. To encourage open data sharing, participants will be able to access their health information, as well as research that uses their data, during the study. Researchers will use these data to study a large range of diseases, with the goals of better predicting disease risk, understanding how diseases occur, and finding improved diagnosis and treatment strategies.

Learn more about the Precision Medicine Initiative:

The NIH website about the All of Us Research Program (https://allofus.nih.gov/) provides information about the goals of and participants in the Precision Medicine Initiative. This infographic (https://syndication.nih.gov/multimedia/pmi/infographics/pmi-infographic.pdf) gives a visual overview of the project. Additionally, the NIH Precision Medicine Initiative channel (https://www.youtube.com/channel/UCQId1TfpwPaYiDIGlxEhlkA) on YouTube includes talks by scientists and others about various aspects of the initiative.


Dr. Francis Collins, director of the NIH, and Dr. Harold Varmus, former director of the NCI, wrote more about their vision for the Precision Medicine Initiative (http://
What are some potential benefits of precision medicine and the Precision Medicine Initiative?

Precision medicine holds promise for improving many aspects of health and healthcare. Some of these benefits will be apparent soon, as the All of Us Research Program is set up and new tools and approaches for managing data are developed. Other benefits will result from long-term research in precision medicine and may not be realized for years.

Potential benefits of the Precision Medicine Initiative:

- New approaches for protecting research participants, particularly patients' privacy and the confidentiality of their data.
- Design of new tools for building, analyzing, and sharing large sets of medical data.
- Improvement of FDA oversight of tests, drugs, and other technologies to support innovation while ensuring that these products are safe and effective.
- New partnerships of scientists in a wide range of specialties, as well as people from the patient advocacy community, universities, pharmaceutical companies, and others.
- Opportunity for a million people to contribute to the advancement of scientific research.

Potential long-term benefits of research in precision medicine:

- Wider ability of doctors to use patients' genetic and other molecular information as part of routine medical care.
- Improved ability to predict which treatments will work best for specific patients.
- Better understanding of the underlying mechanisms by which various diseases occur.
- Improved approaches to preventing, diagnosing, and treating a wide range of diseases.
- Better integration of electronic health records (EHRs) in patient care, which will allow doctors and researchers to access medical data more easily.
Read more about the promise of precision medicine and the Precision Medicine Initiative:

Dr. Francis Collins, director of the NIH, and Dr. Harold Varmus, former director of the NCI, wrote about their vision for the Precision Medicine Initiative (http://www.nejm.org/doi/full/10.1056/NEJMp1500523) in the New England Journal of Medicine. Dr. Collins also gave a talk about his vision (https://www.youtube.com/watch?v=ObBYk0MOuDM) for the project, which is available on the NIH Precision Medicine Initiative channel (https://www.youtube.com/channel/UCQId1TfpwPaYiDIGlxEhlkA) on YouTube.

Examples are available of how precision medicine is helping Americans (https://obamawhitehouse.archives.gov/blog/2015/01/29/precision-medicine-already-working-cure-americans-these-are-their-stories).
What are some of the challenges facing precision medicine and the Precision Medicine Initiative?

Precision medicine is a young and growing field. Many of the technologies that will be needed to meet the goals of the Precision Medicine Initiative are in the early stages of development or have not yet been developed. For example, researchers will need to find ways to standardize the collection of clinic and hospital data from more than 1 million volunteers around the country. They will also need to design databases to store large amounts of patient data efficiently.

The Precision Medicine Initiative also raises ethical, social, and legal issues. It will be critical to find ways to protect participants’ privacy and the confidentiality of their health information. Participants will need to understand the risks and benefits of participating in research, which means researchers will have to develop a rigorous process of informed consent (https://ghr.nlm.nih.gov/primer/testing/informedconsent).

Cost is also an issue with precision medicine. The Precision Medicine Initiative itself will cost many millions of dollars, and the ongoing initiative will require Congress to approve funding over multiple years. Technologies such as sequencing large amounts of DNA (https://ghr.nlm.nih.gov/primer/genomicresearch/sequencing) are expensive to carry out (although the cost of sequencing is decreasing quickly). Additionally, drugs that are developed to target a person’s genetic or molecular characteristics are likely to be expensive. Reimbursement from third-party payers (such as private insurance companies) for these targeted drugs is also likely to become an issue.

If precision medicine approaches are to become part of routine healthcare, doctors and other healthcare providers will need to know more about molecular genetics and biochemistry. They will increasingly find themselves needing to interpret the results of genetic tests (https://ghr.nlm.nih.gov/primer/testing/interpretingresults), understand how that information is relevant to treatment or prevention approaches, and convey this knowledge to patients.

Learn more about challenges related to precision medicine:

The NIH Precision Medicine Initiative channel on YouTube (https://www.youtube.com/channel/UCQId1TfpwPaYiDIGlxEhIKA) offers videos of talks by leading experts on various aspects of the project, including issues related to data collection and sharing, storing data in electronic health records, and participant protection.

The Genetic Literacy Project provides the editorial “That ‘Precision Medicine’ initiative? A Reality Check” (https://geneticliteracyproject.org/2015/02/03/tha-
precision-medicine-initiative-a-reality-check/) outlining some of the possible challenges and limitations of the Precision Medicine Initiative.