



Community Guide to Precision Health



INDEX

	Page
▪ Key Words	3
▪ What is precision health?	5
▪ How will genetics be used in precision health?	7
▪ Participation in Precision Health Research	9
▪ Historical Injustices and Ethical Guidelines	12
▪ For More Information	13
▪ About Us & Contact Information	14
▪ Sources	15

KEY WORDS

Precision Medicine is an approach to total health care (physical and mental) that takes into account factors specific to you—like where you live, what you do, and your family history—to create treatment and prevention strategies that are a better fit.

Microbiome is the collection of micro-organisms (bacteria, viruses, single-celled organisms) that make up the human body.

Race is associated with biology, whereas ethnicity is associated with culture.

Though all humans belong to the same species (*Homo sapiens*), there are genetic variations that result in different races with diverse physical appearances, such as in skin-color.

Ethnicity refers to the culture of people in a given geographic region, including their language, heritage, religion, and customs.

Homogenous: made up of all the same kind of parts

Ancestry and Family History

Ancestry can be more specific than race and ethnicity alone...considering one's personal and family history.

Equality

Equality is about sameness; it promotes fairness and justice by giving everyone the same thing. This only works if everyone starts in the same place. In this example, equality only works if everyone is the same height.

Equity

Equity is about fairness and ensures that people get access to the same opportunities while taking into consideration differences and/or history that can create barriers to participation.

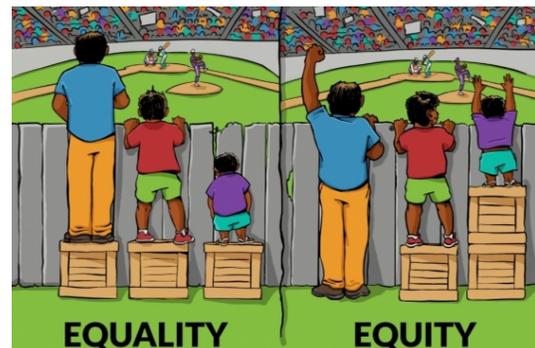


Figure 1. Illustration of “Equality” vs “Equity”.
Source: Interaction Institute for Social Change
(Artist: Angus Maguire)

Health inequity refers to unfair, avoidable differences arising from poor governance, corruption, or cultural exclusion while **health inequality** simply refers to the uneven distribution of health or health resources because of genetic or other factors or the lack of resources.

Health Disparity

Health and health care disparities refer to differences in health and health care between population groups. Disparities occur across many dimensions, including race/ethnicity, socioeconomic status, age, location, gender, disability status, and sexual orientation.

Genetics is a way for characteristics of living things, like humans, to get passed on from one generation to the next. Genes are biological code with information for passing on traits. Genes interact with many things, like your environment, to give rise to traits.

Epigenetics is the study of factors affecting how genes are activated and expressed

Genome: the complete set of genes or genetic material present in a cell or organism

Health Research is the process of finding out new things about improving people's health.

Research Protocol: the official procedure or system of rules used in a research study

Ethics are moral principles and rules of decency that govern a person's or group of people's behaviors or the conducting of an activity.

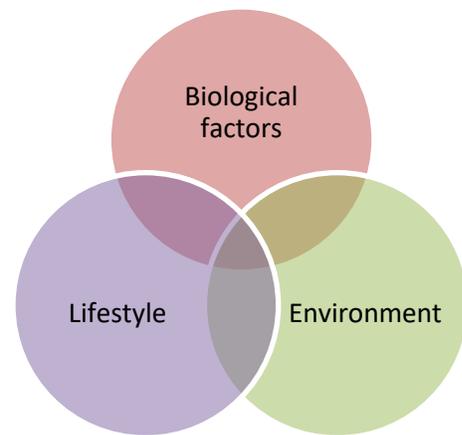
PRECISION HEALTH

Precision health is a way of approaching disease prevention and treatment that considers people's individual differences in genes, environment, and lifestyle. We are committed to precision health that focuses on health issues that the community cares about most.

Why is healthcare moving towards precision health?

“Most medical treatments have been designed with the average patient in mind. This approach works for some patients, but not for others. Where we live, our genetic makeup, our family medical history and our lifestyle choices all contribute to our health and well-being. Precision health takes into account these differences to create management, treatment, and prevention strategies. Together, this gives the medical team the tools to better understand how complex the human body is.”¹

Figure 2. Factors Affecting Health



Biological factors—demographic data like sex and age, as well as factors like genes, metabolism, microbiomes, etc.

Lifestyle—behaviors that can affect the health of people and populations

Environment—many things from our physical and social environment can affect our health, from exposure to unhealthy foods or poor air quality to neighborhood stress or limited access to health care.

Let's meet John Doe:

John is a 50-year-old Caucasian man who makes a living as a farmer in a rural town in Northern California. He spends most of his time outdoors and does not use sunscreen.

A. How do you think the biological, lifestyle, and environmental factors affect John's health?

- Biologically, John's light skin tone and older age increases his risk for skin cancer.
- Behaviorally, his outdoors farming lifestyle and lack of using protective sunscreen adds to this risk.
- Environmentally, living in a sunny region, as well as the possibility of having limited health resources from living in the countryside can affect his health. Chemicals used at his farm can cause epigenetic changes.

B. To best take care of John, health care providers must take all of this into account. While we may know the interaction of these factors for skin cancer risk, there remain many health conditions and factors that need to be studied to help all patients in a more precise and holistic way. This is the promise of precision health.

The importance of working with our diverse communities

Historically, most medical treatments have been designed for the average patient—too often not reflecting the diversity of patients in the real-world setting.^{2,3} Most physicians and scientists are informed by research findings from a largely homogenous population, usually white and male. Because of this, treatments may be characterized as being safe and effective for certain groups of patients but not for others. Racial and ethnic minorities have not fully benefiting from these clinical and biomedical advances.

Consider this example: The spirometer is a medical instrument used to measure lung function and is important to the diagnosis and management of many lung diseases. “Normal” lung function and lung capacity vary among groups. For example, African-American children were found to have lower lung volumes than White children⁴. Therefore, guidelines for spirometers should be created using studies that involve multi-ethnic data so that we can achieve more appropriate diagnosis and treatment.

The national effort of precision medicine focuses on involving people that have historically been underrepresented, who are from all areas of the US, and are in all life stages⁵. Along these lines, it has become obvious that precision health research must focus on truly engaging with community members.

The future of precision health relies on:

- Research that is guided by community values and input
- Diversity and inclusion of historically underrepresented groups
- Community representation in governance, design, conduct, dissemination, and evaluation
- Building a strong foundation of trust

Want to know how the people in your bay area community felt about precision health?

In 2016, SPHERE conducted 12 focus groups involving local community members and healthcare providers on their understanding and perception of precision medicine. Groups included: African America, American Indian, Chinese, Latino, Vietnamese, and various medical doctors from the area.

Theme 1: Understanding Precision Health

- All groups reported limited knowledge about precision health
- Most groups perceived that precision health is related to genes, and used to screen, prevent, and treat certain diseases (including genetic conditions) in specific racial/ethnic populations.

“It will help our future for our kids and our grandkids and everybody else” (Latino group)

“I’m suspicious of continued genocide. It’s just... it’s definitely paranoia, but it upsets me” (American Indian group)

Theme 2: Attitudes about Genetic Testing

- When asked about future genetic testing, all the groups mentioned considering undergoing testing under certain conditions: if disease is curable, enough information about the test, discussion with family first, consent before collecting samples, or physician expertise

Theme 3: Participation in Research

- When asked about participants’ willingness to participate in research, responses varied across and within groups by: generational status, type of data being collected, and type of research (i.e. experimental; how invasive it is; whether blood is involved).

Theme 4: Fears and Concerns

- Participants were worried about: Security and confidentiality of genetic information, cost and affordability of genetic tests and precision medicine, discrimination, and lack of trust

“We don’t want to be guinea pigs” (African American group)

“Language barrier is really huge because sometimes even though they have translators they cannot express in the fullest way that they want” (Vietnamese group)

“Genetic testing might create some sort of fear (of having certain disease/death) to some people” (Chinese group)

GENETICS AND PRECISION HEALTH

Note: It is important to note that precision health does not always involve genetics. Also, genetic testing and genetic research is not always the same thing.

What are genetics?

Genes provide a way for characteristics of living things, like humans, to get passed on from one generation to the next. While many traits are passed on, no one will receive exactly all the same genetic material from generation to generation. Genes interact with many things, like your environment, to give rise to traits. Therefore, genes may help us have a glimpse of what your health may look like, but it is only one part of the picture.

Genetic tests may be used to identify increased risks of health problems, to choose treatments, or to assess responses to treatments. There are many different types of genetic tests.

Financial: Genetic testing can cost anywhere from less than \$100 to more than \$2,000. Health insurance companies may cover part or all the cost of testing.

Discrimination:

Many people are worried about discrimination based on their genetic test results. In 2008, Congress enacted the Genetic Information Nondiscrimination Act (GINA) to protect people from discrimination by their health insurance provider or employer. GINA does not apply to long-term care, disability, or life insurance providers.

GENETIC TESTING
NHGRI FACT SHEETS
genome.gov

Genetic Tests Can Help to:

- Diagnose Your Disease**
- Pinpoint Genetic Factors That Caused Your Disease**
- Predict How Severe Your Disease Might Be**
- Choose the Best Medicine and Correct Dose**
- Discover Genetic Factors That Increase Your Disease Risk**
- Find Genetic Factors That Could Be Passed to Your Children**
- Screen Newborns for Certain Treatable Conditions**

NIH

Figure 3. Genetic Testing Fact Sheet. Source: National Human Genome

For more information about genetic discrimination and GINA, see:

<http://www.genome.gov/10002328/Genetic-Discrimination-Fact-Sheet>

For more information about genetic testing, please visit:

<https://www.genome.gov/19516567/faq-about-genetic-testing/>

Genetic Research is used to learn more about the diagnosis and treatment of genetic disorders, infectious diseases, and non-infectious diseases. Genetic research can include genetic testing, gene therapy, reproductive genomics, genetic databanks, and pharmacogenetics.

Why should I participate in genomics research?

Participating in genomics research is an opportunity to support scientific exploration of the genome and to help understand, prevent, detect, and better treat disease. Often, only through the recruitment of large numbers of participants from different populations can major scientific advances be achieved. In some cases, research participants may also gain some individual benefit, such as diagnosis of a disease or access to a treatment in development. A wide range of genetic research studies need participants, each with different levels of involvement, and it is important to understand the scientific purpose of any study and any potential personal benefit from participation.

What healthcare will I receive as part of the study?

As a study participant, you may receive some care as part of the research but only in accordance with the protocols of the research. To receive clinical care, you should consult your regular physician.

Financial: Participation in genetic research or clinical trials is free, and some studies may include compensation.

For more information, please visit:
<https://www.genome.gov/27561546/participating-in-genomics-research/>

PARTICIPATING IN PRECISION HEALTH RESEARCH

Health research is the process of finding out new things about improving people's health.

There are many different types of health research. All research projects collect data in some form, ranging from information you share in a questionnaire to parts of your medical record to biological samples like saliva or blood.

Behavioral Studies: These are studies that test how people act in different ways.

Clinical Trials: These are studies of a drug, surgery, medical device, or change in health behavior in volunteers or people who have a specific disease. See below for more information.

Genetic Studies: These are studies to find the role of genes in different diseases.

Observational Studies: These are studies in which a group of people are observed for a period.

Physiological Studies: These are studies to better understand how the human body functions.

Prevention Studies: These are studies that test ways to prevent specific conditions or diseases.

Public Health Research: This type of research can be one or a combination of the types of research mentioned above. Public health research tries to improve the health and well-being of people from a population-level perspective.

Why is health research important?

We all want to live long and healthy lives. People who do health research are identifying better preventive care to keep us healthy. And they are discovering new medicines and treatments for when we do get sick.

What is a clinical study?

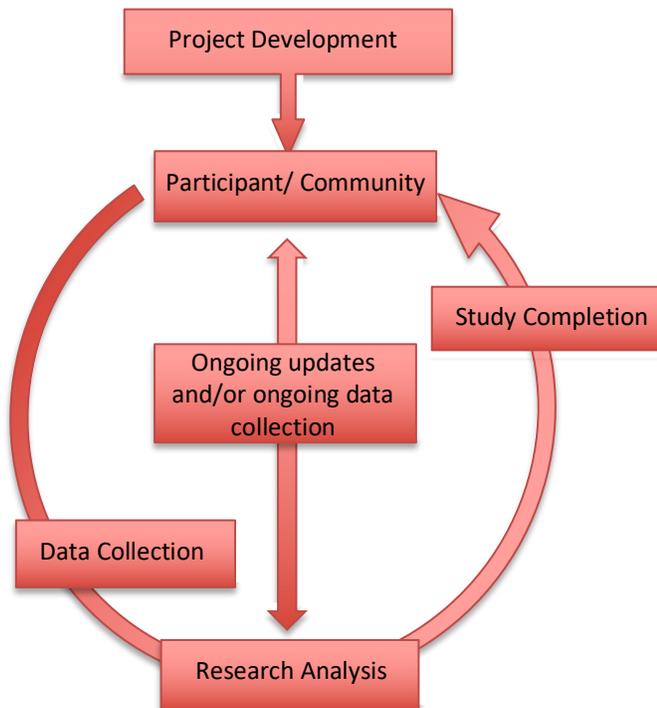
A clinical study involves research using human volunteers (also called participants) that is intended to add to medical knowledge. There are two main types of clinical studies: clinical trials (also called interventional studies) and observational studies.

In a **clinical trial**, participants receive specific interventions that may be any of many different things—medical products, such as drugs or devices; procedures; or changes to participants' behavior, such as one's diet. Clinical trials may compare a new medical approach to a standard one that is already available, to a placebo that contains no active ingredients, or to no intervention. Some clinical trials compare interventions that are already available to each other. When a new product or approach is being studied, it is not usually known whether it will be helpful, harmful, or no different than available alternatives (including no intervention). The investigators try to determine the safety and efficacy of the intervention by measuring certain outcomes in the participants. For example, investigators may give a drug or treatment to participants who have high blood pressure to see whether their blood pressure decreases.

In an **observational study**, investigators assess health outcomes in groups of participants according to a research plan or protocol. Participants may receive interventions (which can include medical products such as drugs or devices) or procedures as part of their routine medical care, but participants are not assigned to specific interventions by the investigator (as in a clinical trial). For example, investigators may observe a group of older adults to learn more about the effects of different lifestyles on cardiac health.

We believe patients and/or communities should be involved in research throughout the research cycle.

Figure 4. Process of Research



1. Project Development: A research question or idea is born, teams are formed, the process of the research (methods) is defined, grants are written to secure funding for the project, and if personal data is used then the project must be reviewed by a board for security.

2. Participants Involvement: People are recruited to the study; they are informed about the study and then asked to agree to participate in the study according to what the study requires by signing consent forms.

3. Data Collection: Can be self-report measures (e.g. disease symptoms), clinical data from electric health records, health exam data, biological samples, etc.

4. Research Analysis: Examination of samples or data using laboratory or software techniques

5. Study Completion: Final results returned to the community and published.

How are participants protected in research?

For legal as well as ethical reasons, there are procedures to ensure that:

- Researchers follow the four ethical principles of biomedical research (see below)
- The Institutional Review Board (IRB) reviews research that involves human subjects to ensure that the research is ethical.
- The identity of participants is protected (confidentiality) and the data is physically secure

What are the four ethical principles to biomedical research?

1. **Autonomy** - participants give informed consent to be a part of the research.
2. **Justice** - researchers must be fair and treat all participants equally.
3. **Beneficence** - research must be something that will be helpful to most people.
4. **Nonmaleficence** - research should not purposely cause harm.

What is Informed Consent?

With the exception of simple observation in public locations or the review of available documents, researchers must ensure that participants understand what they are expected to do, who is paying for the study, the risks and benefits, the level of confidentiality offered, their right to decline to participate, and their right to withdraw from the study at any time. This information is summarized in a form called consent. Participants agree to being involved by signing a consent form prepared by the research team.

What is an Institutional Review Board?

Most institutions and agencies now have an Institutional Review Board (IRB) to oversee research on human and animal subjects. One task of an IRB is to make a formal assessment of whether the potential benefits of the study outweigh the risk. The IRB is composed with professionals from different backgrounds and who have the knowledge and expertise to review the research plan. The make-up of the IRB committee at each institution reviewing the research project may differ.

What are transparency requirements?

“Full disclosure” requires researchers to be completely open with all aspects of the research project. This may pose a special problem to behavioral health researchers because knowledge of all aspects of the research may affect participants’ behaviors. A common solution is to withhold information during the session and debrief the participants afterward. Some researchers will tell people that they are taking part in a psychological experiment that will not involve pain or any physical danger and ask them for permission not to tell them about the experiment until afterward to obtain unbiased responses. Immediately following the session, each participant is given a printed sheet describing the purposes and methods of the study; the experimenter is present to answer any questions.

How is confidentiality ensured?

Confidentiality means that the respondent’s identity may be known to the investigator but protected from public exposure. The researcher keeps any identifying information out of published reports.

Some researchers use a special code at the time of the interview so that no names or other identifying information exist even in the researcher’s files. The exact methods used to protect your respondents will vary according to the situation, but it is important to realize that behavioral researchers do not have the right to communicate confidential information to those not involved in the study.

How is data physically protected?

Information will be stored on protected computers. Researchers should limit and keep track of who sees the information. You should be notified if there is a data breach.

HISTORICAL INJUSTICES AND ETHICAL PRINCIPLES

We are sensitive to the historical medical injustices that were endured by many minority groups in our country and want to respect the experiences and impact that these injustices have had. Such events include the Tuskegee Syphilis study in which readily available treatment for syphilis was withheld from African American men and the Havasupai Tribe lawsuit in which blood samples from the Havasupai tribe were misused in additional research analysis that the participants did not agree to.

We promise to abide by the highest ethical standards in our precision health endeavors, and to that end we promise to:

- *Actively engage with the community in discussion about historical medical injustices*
- *Honor the significance that bodily substances hold in many cultures*
- *Work with community partners to shape our ethical guidelines—which will guide how we conduct precision health research*

We are in the process of collaborating with community partners to develop Precision Health Ethical Principles which will be added to this guide upon completion.

FOR MORE INFORMATION

All of Us –National Institute of Health Precision Medicine Initiative

<https://allofus.nih.gov/>

Precision Medicine in Cancer Treatment

<https://www.cancer.gov/about-cancer/treatment/types/precision-medicine>

US Food and Drug Administration—Precision Medicine

<https://www.fda.gov/ScienceResearch/SpecialTopics/PrecisionMedicine/default.htm>

The White House on The Precision Medicine Initiative

<https://obamawhitehouse.archives.gov/node/333101>

Genetics Home Reference

<https://www.jax.org/education-and-learning/clinical-and-continuing-education/precision-medicine-for-your-practice>

National Human Genome Research Institute

<https://www.genome.gov/>

Precision Medicine for Your Practice—for Health Care Providers

<https://www.jax.org/education-and-learning/clinical-and-continuing-education/precision-medicine-for-your-practice>

UCSF Clinical Trials Finder website: clinicaltrials.ucsf.edu

Spanish (<https://clinicaltrials.ucsf.edu/about/spanish>)

Chinese (<https://clinicaltrials.ucsf.edu/about/chinese>)

Vietnamese (<https://clinicaltrials.ucsf.edu/about/vietnamese>)

Stanford Medicine Precision Health

<http://med.stanford.edu/precisionhealth.html>

UCLA Institute for Precision Health

<https://www.uclahealth.org/precision-health/about>

Precision Medicine at UCSF

<http://precisionmedicine.ucsf.edu/>

UC Davis Health Precision Medicine

<https://www.ucdmc.ucdavis.edu/precision-medicine/>

California Initiative to Advance Precision Medicine

<http://www.ciapm.org/>

“California unveils ‘precision medicine’ project”—NATURE Magazine

<http://www.nature.com/news/california-unveils-precision-medicine-project-1.17324>

ABOUT US

This guide was created by Thanh Truong, SMS IV, through a Valley Fellowship with the Office of Community Engagement, in collaboration with the SPHERE Consortium Core, our local community partners and community members.

PURPOSE OF SPHERE

The Stanford Precision Health for Ethnic and Racial Equity (SPHERE) center builds upon Stanford School of Medicine's leadership around precision health and is one of three NIH precision health centers committed to ending health disparities in the US. The SPHERE center is composed of 3 research projects and 5 working groups ("cores").

Description of Consortium Core – SPHERE's Community Advisory Board

- 14 Core members reflecting diverse communities
- Hold quarterly meetings as well as meetings with individual members
- Core members are compensated for their time

Role of Consortium Core Members

- Provide input and perspective on SPHERE core activities
- Guide the development of Pilot Awards Program and Focus Groups
- Design and implement ethical guidelines and community education tools

Consortium Core Members



More about Stanford Precision Health:

<http://med.stanford.edu/precisionhealth.html>

More About SPHERE:

<http://med.stanford.edu/phs/initiatives.html>

Sources

1. Health U. What is Precision Health. <https://www.uclahealth.org/precision-health/what-is-precision-health>.
2. Oh SS, Galanter J, Thakur N, et al. Diversity in Clinical and Biomedical Research: A Promise Yet to Be Fulfilled. *PLoS Med*. 2015;12(12):1-9. doi:10.1371/journal.pmed.1001918.
3. Hamel LM, Penner LA, Albrecht TL, Heath E, Gwede CK, Eggly S. Barriers to Clinical Trial Enrollment in Racial and Ethnic Minority Patients wit Cancer. *Cancer Control*. 2016;23(4):327-337. doi:10.1158/1940-6207.CAPR-14-0359.Nrf2-dependent.
4. Braun L. Race , ethnicity and lung function : A brief history. *Can J Respir Ther*. 2015;51(4):99-101.
5. Precision Medicine Initiative (PMI) Working Group. The precision medicine initiative cohort program – building a research foundation for 21st century medicine. *Precis Med Initiat Work Gr Rep to Advis Comm to Dir NIH*. 2015;Sept 17:1-108. <http://www.nih.gov/precisionmedicine/>.

<https://www.joinallofus.org/faq>

<https://www.kff.org/disparities-policy/issue-brief/disparities-in-health-and-health-care-five-key-questions-and-answers/>

<https://www.uclahealth.org/precision-health/>

<https://www.livescience.com/33903-difference-race-ethnicity.html>

<https://en.oxforddictionaries.com/definition/ancestry>

<http://www.sollis.co.uk/wp-content/uploads/2016/10/equity-vs-equality.jpg>

<http://www.globalhealthurope.org/index.php/resources/glossary/values/179-inequity-and-inequality-in-health.html>

<http://www.who.int/hia/about/glos/en/index1.html>

<https://www.uclahealth.org/precision-health/>

<https://www.ucdmc.ucdavis.edu/precision-medicine/>

<http://www.who.int/genomics/research/en/>

<https://www.genome.gov/27561546/participating-in-genomics-research/>

<https://guides.library.harvard.edu/c.php?g=389023&p=2639516>

<https://clinicaltrials.gov/ct2/about-studies/learn#HowAreParticipants>

<https://study.com/academy/lesson/beneficence-nonmaleficence-in-research-ethics.html>

<https://www.nlm.nih.gov/services/ctexpaccess.html>

<http://psc.dss.ucdavis.edu/sommerb/sommerdemo/ethics/protect.htm>