ASHG
American Society of Human Genetics

PRECISION MEDICINE: THE FUTURE OF HEALTHCARE

MEET JAKE

JAKE HAS ASTHMA.

JAKE'S ASTHMA DIDN'T RESPOND TO STANDARD MEDICATION

Like many others, he started using the usual medication—but it didn't help much. He kept having symptoms, and nothing seemed to work.

GENETIC TESTING REVEALED A VARIANT

So, his doctor decided to dig deeper. She ordered a genetic test to see if Jake's DNA held any clues. And it did! The test showed that Jake had a specific genetic variant—one that changes how his body responds to certain asthma medications.

NOW HE'S THRIVING ON A TREATMENT TAILORED TO HIS DNA

Armed with this information, his doctor switched him to a different treatment—one that works better for people with Jake's unique genetic makeup. This time it worked.

WHAT IS PRECISION MEDICINE?

USING PERSONAL INFORMATION LIKE GENETICS TO FIND A TREATMENT THAT WORKS BEST FOR EACH PERSON

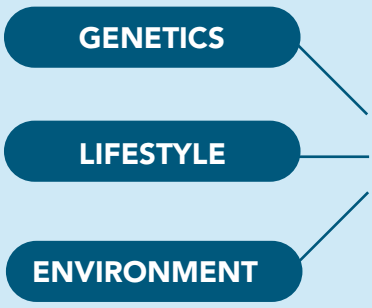
Precision medicine goes by many names:

- Personalized medicine
- Individualized medicine
- Tailored medicine

They all point to the same idea: using what we know about a person to give them the best possible care. Unlike traditional medicine that uses a "one-size-fits-all" approach, precision medicine takes into consideration a person's genetics, lifestyle, and environment to better inform healthcare decisions.

Care designed for YOU—not 'one-size-fits-all.'

Using what we know about a person to provide the best possible care:



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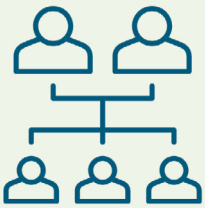
HOW IS PRECISION MEDICINE CURRENTLY BEING USED?

Doctors are already using precision medicine by looking at lifestyle, environment, and health data to tailor treatments. Tools like wearable devices, imaging, and electronic health records help personalize care without using genetics.

But now, we are entering a new era—thanks to the Human Genome Project. By understanding a person's DNA, doctors can target treatment for diseases more accurately than ever before.

PRECISION MEDICINE IS EVOLVING HEALTHCARE FROM "WHAT WORKS FOR MOST PEOPLE" TO "WHAT WORKS BEST FOR YOU"

PREDICTING DISEASE RISK AND GUIDING PREVENTION STRATEGIES

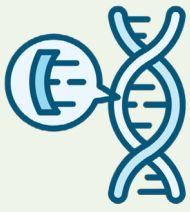


Genetics, lifestyle, and environment can influence a person's risk for health conditions such as cancer, diabetes, and heart disease. A healthcare provider can use a person's family history of these diseases to inform people who may be at higher risk for certain diseases or may benefit from genetic testing. This combined knowledge of family history, genetic testing, and lifestyle choices (such as smoking, alcohol use, and nutrition) can predict a person's likelihood of developing a chronic disease. A healthcare provider can then recommend steps the person can take to prevent the disease or find the disease early.

INFORMING MEDICATION DOSAGE AND RESPONSE

A person's DNA can affect how they respond to medications. Pharmacogenomics is a type of precision medicine that uses a person's genetics to identify the right drug and the right dose for an individual. Before prescribing certain medications, a doctor may recommend genetic testing to determine if the medication will be effective or if it will cause an adverse reaction in that individual. Healthcare providers often use pharmacogenomic testing to inform cancer treatment. Because every cancer is different, not all cancers will respond to the same treatment. A healthcare provider will test for the specific type of cancer, for example, HER2-positive breast cancer, to determine the best medication that will target that specific cancer. Healthcare providers also use pharmacogenomic testing to inform prescription and dosage for other medications, from antidepressants to heart medication to blood thinners.

TREATING AND MANAGING DISEASE



Gene therapy is a form of precision medicine that uses a person's genetics to treat rare diseases and other conditions with a genetic component. Gene therapy uses gene editing technologies such as CRISPR/Cas to cut the DNA in precise locations and then uses DNA's natural repair process to insert, change, or delete that section of DNA. The Food and Drug Administration (FDA) approved the use of gene therapy to treat sickle cell disease, and clinical trials are underway for the use of gene therapy to treat diseases such as cancer, inherited blindness, and cardiovascular disease.

WHAT ARE THE CHALLENGES OF PRECISION MEDICINE?

WHILE PRECISION MEDICINE HAS MANY BENEFITS, IT ALSO COMES WITH CHALLENGES.

COST AND ACCESS

- While the cost of genome sequencing has decreased in the last two decades, these technologies are still relatively expensive.
- Limited availability is also a significant barrier to access.
- Access is particularly limited for people with lower income, people living in rural areas, and people who are uninsured/underinsured, and this may lead to increased health disparities.

EQUITY IN BENEFITTING FROM RESEARCH

- Existing genomic databases consist mostly of participants of European ancestry.
- Without ensuring that people from a variety of backgrounds are represented in research, precision medicine treatments may not be as effective for all people.

Programs such as the *All of Us* Research Program are working to address these challenges. *All of Us* is building a large genomic and health database of people from different ethnic and socioeconomic backgrounds to inform research studies on a wide range of diseases and treatments and make sure precision medicine benefits everyone.

WHAT IS THE FUTURE OF PRECISION MEDICINE?

Precision medicine has the potential to transform the future of healthcare. Advances in genomic technologies have already led to major breakthroughs in preventing and treating disease. As the cost of technology continues to decrease, genetic testing may become routine in healthcare, and targeted treatments will likely become available for more conditions. Jake—and thousands of people around the world whose medications don't work or who have a currently untreatable disease—will see improved health and hope thanks to innovations in precision medicine.

LOOKING AHEAD

Human genetics and genomics research has achieved extraordinary progress, laying the foundation for innovations that improve health and well-being worldwide. Looking ahead, the next generation of discoveries – driven by emerging scientists and transformative technologies – holds even greater promise. Through continued investment, we can ensure these advances benefit all people, reflecting our broad community and global impact: One Humanity, Many Genomes™.

FROM ONE-SIZE-FITS-ALL → ONE-SIZE-FITS-YOU

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