

ASHG Success Stories

in Human Genetics and Genomics Research



HOW HUMAN GENETICS DELIVERS **HEALTH ADVANCES**

Advances in federally funded genetics and genomics research are leading to new discoveries in preventing, diagnosing, and treating diseases—with much more to come. In this way, human genetics and genomics is providing hope for patients, families, and communities.

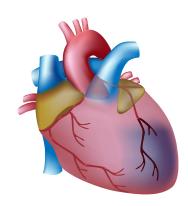
Chronic Diseases

Then: Heart Disease

Heart disease has been the leading cause of death in the U.S. and worldwide for decades.¹ Heart disease refers to a number of conditions that affect the heart's structure and functions. Risk factors for heart disease include behavioral, social and environmental factors, and one's genetics.²

Now

New drugs for lowering cholesterol, discovered through genetic studies, are contributing to a reduction in the number of heart disease deaths relative to the population. Also, researchers have recently developed a new method to calculate how changes across your entire set of DNA sequences, or genome, affect your risk of developing heart disease, which may enable screening to predict, identify, and prevent disease.34



Then: Cancer

Cancer, the second leading cause of death in the U.S. after heart disease, is a disease caused by changes in the genome, called mutations, leading to an uncontrolled growth, division, and spreading of cells.⁵

Now

Thanks to advances in research, new breakthroughs in cancer detection and treatment options are improving patient care. In 2017, the FDA approved the first gene therapy for children and young adults with acute lymphoblastic leukemia (ALL). In the years since, the FDA has approved four more gene therapies for other cancer types.⁶ In 2020, the first DNA blood test to identify multiple genetic changes in tumors, known as liquid biopsies, was approved.⁷ The tests help guide targeted treatment. Current studies are developing liquid biopsies for early screening and detection of multiple cancers.8

"So learning from the NIH that CAR T-cell therapy was an option for me at the University of Maryland Medical Center literally saved my life....Given the increasingly rapid advances in medicine, never lose hope!"

> -Sonia Su, Diagnosed with Non-Hodgkin Lymphoma in 2018

Imagine

Being able to prevent or cure heart disease and cancer and other debilitating chronic diseases such as diabetes, kidney disease, and Alzheimer's.

References:

ashg.org/advocacy/fact-sheets/