ASHG
American Society of Human Genetics

Success Stories
in Human Genetics and Genomics Research

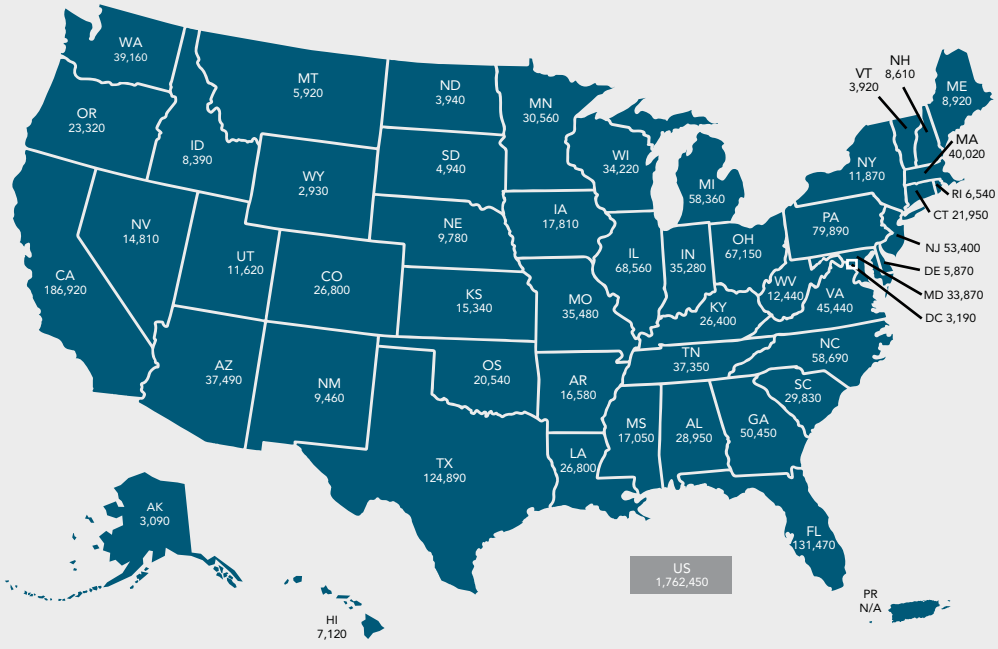
CANCER GENETICS AND GENOMICS

Cancer, a disease caused by mutations in the human genome, is the second leading cause of death in the United States. Basic genetics and genomics research funded by the National Institutes of Health (NIH) has led to a greater understanding of the causes of numerous types of cancer, which in turn is leading to advances in clinical care. The development of early prevention and detection tools, as well as increasingly effective targeted therapies, has improved the prognosis for patients.

Cancer Incidence by State

Estimated numbers of new cancer cases for 2019, excluding basal cell and squamous cell skin cancers and *in situ* carcinomas except urinary bladder.

Estimates are not available for Puerto Rico.



Source: American Cancer Society. Cancer Facts & Figures 2019.

Genetic Underpinnings of Cancer

Cancer is characterized by changes to genes that control the way our cells function, especially how they grow and divide.[1] Although the causes of cancer are not completely understood, many factors are known to increase its occurrence, including inherited genetic mutations and aspects of environment and lifestyle.[2]

Large-scale research projects, such as The Cancer Genome Atlas[3] funded by the NIH, have used DNA sequencing to investigate many types of cancer. These efforts have revealed genetic similarities driving the development and growth of different tumors. For instance, researchers have discovered that mutations in one gene can cause a number of cancers, including breast, bladder, pancreatic, and ovarian.[4]

Diagnostic Advances

Inherited genetic mutations contribute to about 5- to 10-percent of all cancers and researchers have linked specific mutations to over 50 hereditary cancer syndromes.[5] Genetic tests detecting these mutations are particularly useful for assessing cancer risk in individuals with a family history of cancer. For instance, clinicians perform testing for mutations in the genes *BRCA1* and *BRCA2* to assess women's increased lifetime risk for breast and ovarian cancer.[6] Early detection of cancer-promoting genetic mutations allows patients to take steps to lower their risk of developing cancer. New tests are also being developed to find and treat cancer at an earlier stage. This includes recent advancements in a DNA blood test that is able to detect more than 50 cancer types across different stages and the locations of these cancers in the body with both high specificity and sensitivity.[7]

Personalized Treatments

Cancer genomics research has led to advances in precision medicine, using genetic information to tailor treatment strategies to a patient's tumors.[8] These targeted therapies comprise drugs that block the growth and spread of cancer by interfering with specific molecules in the body.[9] Many targeted cancer therapies have been approved by the Food and Drug Administration to treat specific types of cancer, including CAR-T therapies, which uses patients' genetically engineered immune cells to target cancerous cells. More potential therapies are being studied in clinical trials with patients and in preclinical testing in animal models.

Examples of precision medicine already in clinical use to help patients with cancer include:

- Trastuzumab (Herceptin) is a drug that targets the overexpression of the protein HER-2 in certain breast and stomach cancers.[10]
- Kymriah and Yescarta are CAR-T therapies for the treatment of acute lymphoblastic leukemia and B cell lymphoma, respectively.[11]
- Lung cancer patients with a mutation involving the *ROS1* gene often respond well to treatment with the drug crizotinib.[12]
- Patients with metastatic pancreatic cancer and a *BRCA1* or *BRCA2* mutation survive longer when treated with the drug olaparib.[13]
- The drug vemurafenib (Zelboraf) targets a mutant form of the *BRAF* gene in many patients with inoperable or metastatic melanoma.[14]

The Future of Cancer Research

Researchers have not yet discovered all the genetic changes that cause cancer to develop, grow, and spread, but they are making progress. NIH-funded scientists are collecting and sharing large genomic datasets, boosting the power of the data and opening new opportunities for discovery.[15] Such research collaborations are critical for further investigating the genomic basis of cancer development, metastasis, and drug resistance.[16]

How Can Congress Support Research?

To find better ways to detect, treat, and prevent cancer, researchers need robust and predictable NIH funding. Translating genomic discoveries to clinical advances requires further testing in models of cancer such as cell lines, organoid tissues, or animal models. Congress can continue to support this research and help advance precision treatments for cancer through sustained NIH funding.

Additional Resources

Cancer Genomics
genome.gov/dna-day/15-ways/cancer-genomics

National Cancer Insitiitute
cancer.gov/

References:
ashg.org/advocacy/fact-sheets/

6120 Executive Blvd, Suite 500, Rockville, Maryland 20852 · policy@ashg.org · (301) 634-7300 · www.ashg.org

@GeneticsSociety