

*Department of Genetics and
Genomic Sciences*

2022 Accomplishments

LEADING THE WAY
IN DISCOVERY &
INNOVATION IN
GENETICS RESEARCH
& MEDICINE

As a multidisciplinary powerhouse,

the Department of Genetics and Genomic Sciences integrates an array of genetics expertise across fields including neuroscience, oncology, immunology, human disease, computational biology, and data science, paving the way for **personalized medicine and groundbreaking targeted therapeutics.**

Our unique infrastructure

seamlessly combines basic science, data resources, cutting-edge technology, clinical trials, and patient care, creating a dynamic ecosystem that **accelerates the journey from investigation to transformative discovery.**

By developing new technologies

and methods for genetics research, improving predictive and preventive measures for disease, and identifying new therapeutic targets and medicines, we aim **to revolutionize patient outcomes and enhance quality of life.**

Our legacy,

defined by **groundbreaking treatments for rare diseases** such as Fabry and Niemann-Pick disease, and consistent ranking among the top five nationally-funded institutions for genetics research, is a testament to our **relentless commitment to excellence and innovation.**

→ #1

#1 in NIH funding for genetics research in the New York Tri-State area

→ #2

#2 in NIH funding for genetics research nationally

→ 600

Published over 600 scientific manuscripts

→ \$42,250,000

Received over \$42,250,000 in grant funding

Research and Discovery

- **Revealed the connection of a well-known risk factor for neurodevelopmental disorders to several other conditions** including obesity, kidney failure, hypertension, and obstructive sleep apnea, providing a foundation for further **insights into at-risk individuals and therapeutic strategies** targeting specific genetic variants. →
- **Identified five genes that affect Alzheimer's risk;** now investigating the top two candidates to use as potential **drug targets.**
- **Discovered that disease-related phenotypes** in disorders such as Niemann-Pick C1, a fatal genetic disease, **can be reversed by activating a protein called Trap1,** which could represent **an important new therapeutic for multiple similar conditions.** →
- Joined **NIH's Accelerating Medicines Partnership** to study the role of the human microbiome in **rheumatoid arthritis, systemic lupus, and other autoimmune diseases.** →

Development of Methods and Tools

- **Created a tailor-made gene sequencing method** that relies on a new machine learning algorithm. By helping researchers exclude bias from bacterial contamination in genetic studies of human disease it will **improve research accuracy and advance precision health initiatives.** →
- **Developed a new data integration method to describe the regulatory system between microRNA and messenger RNA.** This system is thought to be affected by environmental exposures and toxins that may lead to disease. By successfully detecting how important microRNA s are disrupted due to chemical exposure, this could help researchers studying the **impact of environmental exposures on disease to identify therapeutic targets.** →
- **Created a new machine learning method** for genetics research that **predicts the impact of genetic variants on protein function.** It can distinguish between pathogenic gain-of-function and loss-of-function variants and outperforms other tools currently used. →
- Developed a new **CRISPR imaging technology that paves the way for new approaches to targeting anti-cancer drugs** by allowing researchers to link specific genes to complex tumor characteristics at a scale and resolution not previously possible. →



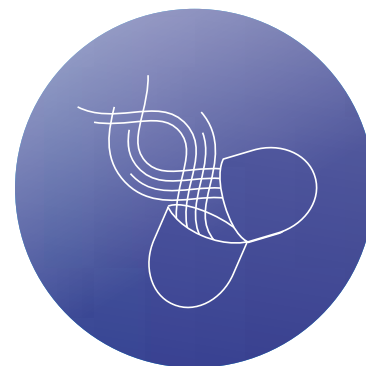
Bench



Drug Discovery and Clinical Trials

- **Developed a new approach to help patients with rare types of cancer.** By combining genomic profiling and drug screening **to identify drugs that specifically target a patient's tumor, it can greatly expand their treatment options.** The potential of this approach to improve clinical decision making for patients with rare or complex cancers has already been proven. [→](#)
- **Led 45 clinical trials addressing over 15 rare diseases.** Our clinical trials include industry sponsored, grant/philanthropy-funded, and investigator-initiated trials in Phases I, I/II, II, III, and IV.

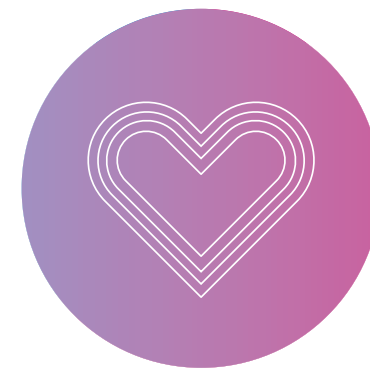
- The FDA approved the **first-ever treatment, which was developed by GGS researchers, for pediatric and adult patients with a rare progressive genetic disorder** known as acid sphingomyelinase deficiency (ASMD). Also served as a key site for the therapy's Phase III clinical trials. [→](#)
- Created opportunities for patients to participate in four sponsor-initiated **clinical trials for children, adolescents, and adults with Fragile X Syndrome;** supported NIH and Robert Wood Johnson Foundation funded basic science projects in Fragile X-associated tremor/ataxia syndrome.
- Designated as a **National Cancer Institute Proteogenomics Data Analysis Center to generate potential biomarkers and drug targets for cancer** and to develop tools for advancing cancer research. [→](#)



To

Innovation and Excellence in Medical Genetics

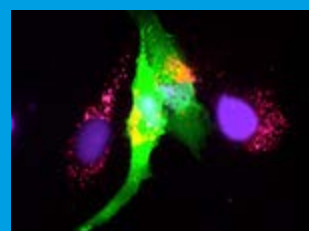
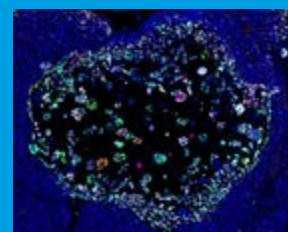
- Provided **specialized care for 11,158 patients** with known or suspected **genetic disorders, rare diseases, birth defects, reproductive complications, and cancer risks.**
- **298 patients with 50 distinct genetic predispositions to cancer** were cared for by our team of **cancer genetic counselors.** The Cancer Genetic Counseling Program assisted patients **at risk for over 15 different type of cancers and collaborated and assisted patients with care from more than 15 different specialties.**
- Reduced barriers to patients and increased convenience of on-site genetic counseling services by launching a **joint clinic** between the **Cancer Genetic Counseling Program** and the **Pancreatic Cancer Prevention and Surveillance Program.** As a result, almost three times as many patients were seen for this indication.
- **Welcomed 45 new patients to our Fragile X Syndrome and Fragile X-Associated Tremor Ataxia Clinics** for consultation or ongoing management of their conditions. Our Clinics are **regional/state referral centers for children** and belong to both the National Fragile X Clinical and Research Consortium and the International Fragile X Syndrome Clinical Consortium.



Bedside



Researchers use multiplex imaging to identify mutant cancer cells and study a mutation's intra- and extracellular effects: Brian Brown, PhD

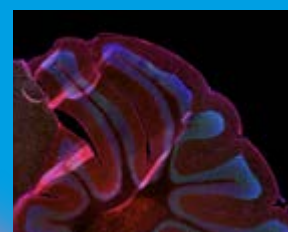


Researchers find activation of a protein called TRAP1 reverses neurodegenerative disease processes in Niemann-Pick C1: Yiannis Iannou, PhD



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212-659-8500



Researchers image microglial proteins in neurons implicated in Alzheimer's disease: Alison Goate, DPhil

<https://icahn.mssm.edu/research/genomics>