Over the last decade, physicians and scientists have become increasingly aware that genetic factors dictate our physical and mental characteristics, and can be used to detect and manage a wide range of health problems.

Feinstein Institute researchers are diving deeper and examining the genetics of those that are sick and those that are well to find a genetic definition of “what is well.” Defining “what is well” can help medical professionals see, depending on that person’s particular genetics, what is required to bring a patient to health. Through research using the biobank, Northwell Health researchers and doctors hope to soon use a patient’s own DNA to develop a personalized treatment plan.

The Northwell Health biobank, overseen and maintained by Peter K. Gregersen, MD, and James M. Crawford, MD, PhD, is one of the largest and most diverse collections of blood and tissue in the New York metropolitan area. Samples for the biobank come from Northwell Health’s 21 hospitals, which serve the most culturally diverse population in the US. Having such a diverse pool to study is advantageous to research because it allows examination of how different factors such as age, race and medical histories play into different diseases and conditions. Through strategic affiliations, we also provide the samples from our biobank to researchers outside of Northwell Health, giving them access to samples they wouldn’t have access to otherwise.

“With the biobank, we are aiming not only to advance research and discovery but to establish new standards of care for patients,” said James M. Crawford, MD, PhD, executive director and senior vice president.
1.6 million Americans will be diagnosed with cancer this year.

We aim to use genetics to develop a targeted diagnosis and treatment plan.

doing this research, the Feinstein Institute and others to Northwell Health patients for first-to-human clinical trials and therapy.

The biobank is a critical component of a successful strategic partnership in cancer with Cold Spring Harbor Laboratory (CSHL), a leader in molecular biology and genetics research. One of the reasons why cancer is so difficult to treat is because the cells in a tumor do not necessarily share features with their tissue-of-origin – they can exhibit the genetic characteristics of a different type of cancer. For example, a patient could have colon cancer that exhibits the same genetic characteristics typically found in lung cancer. This is where the biobank can be an asset to treatment. A patient’s tissue is taken during a procedure and brought to the lab to grow a copy of the patient’s tumor and then test that tumor’s genetic and functional characteristics. Once these characteristics are determined, the team can best identify an effective treatment for that patient. Going back to the example, it may be best to treat a patient’s colon cancer tumor with a treatment for lung cancer based on those genetic and functional characteristics.

A new member to the Feinstein Institute, investigator Kenan Onel, MD, PhD, is helping to spearhead the Institute’s genomics initiatives and is using this research to find treatments for pediatric cancer. Dr. Onel sees children as a potential barometer for the genetic causes of both pediatric and adult cancers because their bodies have not been exposed to as many environment or lifestyle factors as their adult counterparts. One of his studies is GREAT Kids (Genomics for Risk Evaluation and Anti-cancer Therapy in Kids), which is examining the genetics of children with cancer and their family members to look for early indicators of the disease. The hopes of this study are to help prevent the patients’ other family members from getting cancer and translate genetic markers found into cancer identifiers for other patients. Dr. Onel’s research focus then goes beyond cancer, through his LIFE (Long Island Family Empowerment) Project, which will follow 20,000 children treated by Northwell Health and their parents for their entire lives to examine the genetic and environmental factors which contribute to different health conditions. By understanding what contributes to disease and wellness, Dr. Onel and his team hope to develop preventative diagnostics and more targeted treatment.

Feinstein Institute Professor Peter K. Gregersen, MD, an expert in genetics, has established a unique biobanking program called the Genotype and Phenotype (GaP) registry. The GaP is essentially a “living biobank” of volunteer subjects who agree to be recalled for specific research studies based on their genetic makeup. Over 7,000 volunteers have signed up for the GaP over the last decade, and over 80 research studies have been carried out using this resource, including by investigators from leading research institutions nationwide. An expansion of the GaP to 50,000 volunteers is planned in order to accommodate interest from researchers around the world. One such study is directed to developing new diagnostic and treatment approaches for endometriosis. Endometriosis, which affects approximately 176 million women globally, is a condition in which tissue from the uterus grows outside the uterus, causing significant pain and discomfort — it may even lead to infertility. It often takes more than 10 years to diagnose as it can be mistaken for other conditions, and to confirm that it is endometriosis, patients have to undergo surgery. Dr. Gregersen’s team is developing a novel test that may allow for early diagnosis of the disease so that patients may be treated early and not have to undergo surgical treatments.

Drs. Crawford and Gregersen and George Raptis, MD, senior vice president, Cancer Service Line, acting executive director, Northwell Health Cancer Institute, are working to expand the scope of the benefits of the biobank to help all patients who enter Northwell Health hospitals. Following patient consent, Northwell Health hospitals will be able to test patients’ blood with a DNA “chip” called the Global Screening Array (GSA), which can quickly characterize a person’s DNA and identify particular genetic markers of disease. The information from this test can help medical professionals more rapidly diagnose conditions and develop a personalized treatment plan. Patients will also have the option to have these samples included in the Northwell Health biobank to help move research forward with the goal of providing personalized treatment based on genetics.

“Northwell Health is one of the first health organizations to incorporate a GSA into its standard diagnostic protocols,” said Dr. Gregersen. “This cutting-edge technology will help to accelerate diagnosis and identify effective treatment while also providing data to help researchers develop the next generation of targeted therapies.”

Northwell Health