Dear friends,

This past year has presented us with many opportunities to be adaptable and nimble. The COVID-19 pandemic has significantly impacted every aspect of our lives. Research and MyCode were not exempt from that.

But despite the challenges presented by a long-term pandemic, we continue to conduct research here at Geisinger. And MyCode continues to enroll participants and use the data we've gathered to support research projects.

We're also still informing some participants about important genomic information that will allow them to take better control of their own health and potentially help their family members.

As always, thank you for being a part of MyCode.

- The MyCode Team

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MyCode screenings lead to positive health outcomes

A team of Geisinger researchers has found that the MyCode DNA screening program can identify people with an increased risk of cancer and heart disease—which can lead to care that greatly reduces the risk of early heart disease, cancer and stroke.

The research, published in *Genetics in Medicine*, studied participants enrolled in MyCode, which discloses genetic results for about 30 inherited conditions. Two of these conditions lead to higher chances for certain cancers. A third leads to increased chances for early heart attacks and strokes.

**The team found that:**

- 87 percent of study participants with a gene change that increases risk for one of these three conditions didn't already know about their genetic risk.
- More than 70 percent of these then had a recommended procedure, such as a lipid panel or colonoscopy.
- More than 10 percent were then found to have high cholesterol, an early cancer diagnosis or a colon polyp.

"Without MyCode, these people might not have found these health problems until later, when health outcomes might have been worse," says Adam Buchanan, director of Geisinger's Genomic Medicine Institute. “This evidence suggests that genomic screening programs can effectively find people who could benefit from taking extra steps to reduce their risks for cancer or heart disease.”

MyCode has disclosed genetic results for one of these three genetic conditions to about 1,000 people. This makes MyCode the largest genomic screening program in the world.
Thanks to MyCode, her cancer didn’t get the chance to spread.

In October 2019, Dawn Snyder got a call from her primary care provider, giving her a heads-up that she’d soon be hearing from researchers at MyCode. They’d found a gene change that put her at risk for thyroid cancer.

Soon after that call, the researchers helped Ms. Snyder schedule appointments with clinical genetics specialist Gary Bellus, MD, PhD, and two genetic counselors, Gretchen Thone and Kerrianne Fry.

After the consultation, Ms. Snyder underwent tests and was referred to Timothy Lindemann, MD, a surgeon who specializes in thyroid and parathyroid surgery.

Having been a nurse for over 40 years, Ms. Snyder understood what her lab results in myGeisinger meant as soon as she saw them — her tumor markers for medullary thyroid cancer were elevated.

Dr. Lindemann performed an ultrasound and thyroid biopsy.

“He was very honest with me,” says Ms. Snyder. “He thought the cancer had probably been there for years, even though I had absolutely no symptoms.”

Luckily, a scan revealed that Ms. Snyder’s bones and organs were fine — the results were completely negative outside the thyroid.

Dr. Lindemann removed Ms. Snyder’s thyroid gland and central lymph nodes. Now, she just needs to take thyroid replacement medication daily.

Because gene changes can occur in relatives, Ms. Snyder’s counselors worked with her family members so they, too, could understand the risks.

Ms. Snyder says she’s extremely thankful that she chose to participate in MyCode.

“If I hadn’t gotten my results when I did, who knows what might’ve happened,” she says. “Thanks to MyCode, my family members can be prepared for a brighter future.”

MyCode sheds light on link between kidney disease and COVID-19 outcomes

Thanks to MyCode research, we now know that people with chronic kidney disease are among the most vulnerable to COVID-19-related hospitalizations.

The study was conducted to help answer the question, “Why do some people with COVID-19 have mild or no symptoms, while in others, the virus is so much more dangerous?”

Using MyCode and COVID-19 data, along with electronic health record information Geisinger has collected since 1996, researchers worked to pinpoint the role and influence of chronic disease on patient outcomes for COVID.

The study confirmed that factors like high blood pressure, diabetes, obesity, some chronic lung conditions, older age and being male put people at higher risks for COVID-19-related hospitalizations.

But the results related to kidney disease were more surprising. The MyCode information, coupled with the long-term Geisinger health record data, showed that people with end-stage kidney disease are 12 times more likely to be hospitalized due to COVID-19.

And among the small number of kidney transplant patients studied, almost everyone who had COVID-19 ended up in the hospital.

In addition to finding that chronic kidney disease seems to be one of the most critical factors in the severity of COVID-19, the research team believes that having the MyCode database, which is designed for research analysis, was crucial to conducting a nimble, thorough study of this type.
MyCode study seeks to reduce sudden cardiac death

MyCode researchers are studying a rare inherited disease called arrhythmogenic cardiomyopathy (ACM) that can change the heart’s electrical system and make the heartbeat abnormal. The study can help pay for clinical evaluations.

Doctors call abnormal heartbeats “arrhythmias,” and they can even make a person’s heart stop suddenly and without warning. In some families, the first sign of ACM is sudden cardiac death in a loved one who seemed healthy.

Genomic screening programs like MyCode are helping participants learn if they’re at risk for ACM before it develops. If a gene change is found, the MyCode team lets the participant and their doctor know, because there may be ways to keep ACM from developing or getting worse.

As of August 2020, 92 MyCode participants were found to have an ACM gene change. Many had little or no evidence of ACM. Others had some signs of ACM, but not enough to be diagnosed. And only two patients already had heart disease diagnoses that could be related to the gene change. Because of their ACM diagnosis, two participants had a device implanted that would help restart their heart if it were to suddenly stop.

MyCode patients with the ACM gene change can choose to regularly check on their hearts to make sure they stay healthy. Also, because these gene changes can be passed through families, these patients are encouraged to share their MyCode result with family members, so they know they could also be at risk.

If you have concerns about your heart health or a history of heart disease in your family, speak with your doctor. You can reach a cardiovascular genetic counselor at 814-235-5503 or ask your doctor for a referral to the Cardiovascular Genetics clinic. To learn more about the ACM research study, email heartresearch@geisinger.edu.
Discovering genetic links to brain disorders

For the past three years, researchers at Geisinger’s Autism & Developmental Medicine Institute (ADMI) have been reporting genetic results related to learning and mental health conditions including:

- Bipolar disorder
- Anxiety disorder
- Depression
- Learning disabilities
- Seizures
- Autism spectrum disorder

Collectively, these disorders impact about 14 to 18 percent of the nation’s children and adults.

A single genetic change can cause different brain disorders. Two people with the same genetic change may have different health histories, even in the same family. One person might have seizures, a mild learning disability and an anxiety disorder. Their sibling may have bipolar disorder. Knowing about a genetic cause for brain disorders can help people understand their past or present mental and physical health. This information can also help their doctors care for them better.
These genetic changes are called “deletions” because a section of DNA is missing. These sections are important for how our brains grow and develop. If one of these deletions is found in someone’s DNA sample, they can meet with a genetic counselor to learn more about the result.

ADMI genetic counselors Karen Wain, MS, CGC, and Emily Palen, MS, CGC, have expertise working with families with brain disorders. They explain the genetic results in detail and help people understand how their results may fit in with their personal history. They also talk about what the results might mean for other family members and can order genetic testing for them, too.

Over 200 participants have received genetic results related to brain disorders. Most have positive reactions and think being in MyCode was a good decision.

By doing surveys, interviews and tests from home, participants are helping researchers learn how these genetic changes affect people of all ages. Anyone who received a MyCode genetic result related to brain disorders can contact the ADMI team — genetic counselors are happy to answer any questions.

To set up a meeting or to learn more about other research projects related to brain disorders, call or email Karahlyn Holdren at 570-522-6295 or keholdren@geisinger.edu.

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Select participants’ responses to learning their results

“I’m glad I know now, because I would have kept wondering.”

“It’s nice to have an answer... it gives that peace of mind.”

“It’s better to know than to not know.”

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Participants’ perceived benefits of receiving genetic results

- Information was valuable
  - Helped make sense of family history
  - Helped make sense of personal history
- Medicalized diagnosis and/or experiences
- Relieved burden/guilt
Expand your MyCode involvement

_Ethics panel welcomes your input_

Would you like to be part of a panel that works to make sure MyCode adheres to the highest ethical standards?

The MyCode Ethics Advisory Council, formed six years ago, is recruiting patient-participant members to work with our expert researchers.

The expert members have a deep understanding of ethics, genomics and genetics. But our patient-participants bring firsthand experiences, which include:

- Being recruited to MyCode
- Undergoing the process of being informed about — and consenting to — participation
- Receiving genomic results that indicate they may have an elevated risk of disease

One of our top priorities is addressing concerns about the privacy and confidentiality of MyCode data, issues raised by focus groups held several years ago. We’re committed to keeping all data safe and secure, and we’d welcome your input to ensure we continue to meet this and other goals for maintaining the highest ethical standards.

If you’re interested in contributing to this important work, send a message to Dan Davis, PhD, at ddavis1@geisinger.edu.

Help us by responding to a survey

We’re distributing a pilot survey to a subset of MyCode’s 260,000-plus participants. It’s designed to gather information about you and other participants and includes questions about your age, gender, race and ethnicity. It also asks about your interest in participating in other MyCode-fueled studies.

If you receive the survey, please carefully consider these questions. We welcome your continued participation in MyCode and the other research that it makes possible.

Knowledge is power

When MyCode participants get clinical results for familial hypercholesterolemia, or FH, they often tell us they don’t fully understand the condition — or what it means for them and their families.

FH isn’t your average type of high cholesterol. It can lead to high risk for early heart attacks — yet, these issues are preventable if you know about FH and learn to manage other risk factors.

That’s why we developed a video to help. The video:

- Explains that FH is a genetic condition that affects how your body processes cholesterol
- Stresses the importance of treating your FH to avoid heart attack and stroke
- Discusses treatments that can keep you healthy

Watch the video at [geisinger.org/fh](https://geisinger.org/fh) — and be sure to share it with others who might have FH.
Research studies through MyCode

DNA and health data from MyCode research participants like you has contributed to over 250 research studies initiated since 2009. With the help of our participants, we’ve been able to study a broad range of health and disease.

50+ Brain Studies:
- Neuroscience
- Parkinson’s disease
- Autism & developmental medicine
- Psychology
- Addiction
- Behavioral health

40+ Heart & Blood Studies:
- Atrial fibrillation
- Peripheral vascular disease
- Cardiomyopathy
- Blood lipids
- Coronary artery disease
- Hemochromatosis

20+ Cancer Studies
- Bladder cancer
- Breast cancer
- Colon cancer
- Endometrial cancer
- Esophageal cancer
- Familial cancers
- Lung cancer
- Melanoma
- Prostate cancer

Pulmonary/Sleep Studies:
- Asthma
- Smoking
- Sleep disorders
- Pulmonary hypertension
- Chronic obstructive pulmonary disease
- Respiratory distress syndrome
- Cystic fibrosis

Research Studies:
- Population health
- Genomics
- Stem cell
- Pharmacogenetics
- Social media

Digestive Tract Diseases:
- Crohn’s disease
- Eosinophilic esophagitis

10+ Liver Studies:
- Fatty liver disease

10+ Kidney Studies:
- Chronic kidney disease
- End-stage renal disease
- Kidney stones
- Polycystic kidney disease

30+ Obesity & Nutrition Studies:
- Diabetes drug response
- Gestational diabetes
- Obesity
- Monogenic studies
- Gestational weight gain

10+ Bone & Joint Studies:
- Joint infection
- Osteoarthritis
- Pediatric orthopaedic conditions
Explaining the benefits of MyCode is the first step in encouraging people to participate. That's the role of precision health associates. These vital team members enroll patients and carefully check consent packets to make sure all paperwork is filed correctly, so personal information and our research data are secure and accurate. It's a crucial job that requires both attention to detail and a commitment to MyCode's mission of improving healthcare for each person who trusts us with their genetic data and the broader community.

“It's great to see that we are making a difference.”

– Chelsea Kunkel, Central Region precision health associates lead, who has been with the team since the start of her Geisinger career

“I've learned so much about genetics and how research works. I've heard so many different stories from patients of all walks of life. That's the focus of my job — patient interaction.”

– Alexei Rivera, precision health associate, whose fluency in Spanish and English helps him communicate with patients at family practice and outpatient clinics