MEDICINE'S CRYSTAL BALL

SJ scientists can now predict if you'll get cancer or Alzheimer's. Would you want to know?
For Beth Peditto, learning she had stage three colon cancer at age 45 – two weeks after burying her father – was devastating enough. But the idea that she may have passed on a gene mutation to her three sons, making it almost inevitable they would share the same fate – seemed almost unbearable.

Nonetheless, Peditto, a hairdresser from Westville with a family history of cancer, wanted answers. Most of all, she wanted to find out if she was a carrier of a hereditary condition called Lynch syndrome, which typically strikes its victims young and puts them at high risk for other diseases.

“Knowledge is power,” says Peditto, who is recuperating at home after an intensive year of chemotherapy, radiation and surgery to remove a foot of her colon. “I realize not everyone feels that way, but I had to know for my sons.”

If your doctor had a crystal ball that showed your medical future – if you’ll get cancer or develop Alzheimer’s – would you look?
While much is still unknown about many life-threatening illnesses, a wealth of information has been unraveled about our genes, and heredity’s role in the development of diseases. Since 2003, some 20,000 to 25,000 genes in human DNA have been identified, ushering in the era of genetic testing.

As Michael Christman, president and CEO of Coriell Institute of Medical Research in Camden, sees it, this is an exciting time in the history of clinical care.

“It’s the convergence of some technology and some knowledge about the human genome,” explains Christman, an expert in genetics – the study of heredity – and genomics, the study of genes and their functioning.

“I think the complete sequencing of the genome in 2003 serves as a platform for understanding complex diseases like cancer. And now we also have some technologies for measuring human genetic variation. We have matured to a point where genetic tests are cost effective and fairly comprehensive. It’s the first time, historically, we are able to use testing for clinical care.”

Coriell, for example, offers free genetic testing to 10,000 volunteers willing to spit saliva in a test tube for analysis. Participants in the research project gain information on their genetic risk for a number of diseases as well as ways to share the information with their physicians. The goal of the program is to research whether personalized genetic information can be used to improve people’s health. The saliva samples are used to look for genetic variants associated with common diseases and responses to medication. Medical history, family history and lifestyle are also taken into consideration.

Also in SJ, another genetic program identifies high-risk individuals or families in hopes of catching cancer in its early stage – or preventing its onset altogether. Scientists in the Cancer Genetics Program at Virtua trace family roots thoroughly, analyze environmental factors and then, together with oncologists, develop a personalized action plan.

Currently, more than 1,000 genetic tests, costing between $200 and $3000, are available from testing laboratories. There are no regulations for evaluating the accuracy and reliability of the tests, even as for-profit companies promise to deliver everything from home-testing of DNA for cancer, to screening a child’s sports ability or even determining who will go bald. While much good has come out of genetic testing – with more benefits to come as testing becomes the standard of care in the future – there is much to be concerned about as well, experts say.

Christman notes that human error is a real worry, particularly when specimen samples are collected by individuals at home and shipped to faraway companies. To be accurate, genetic testing must be done under the highest of laboratory standards, he says, noting that such conditions are expensive to replicate and likely not practiced by every company hawking tests on the web.

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Just as concerning is the fact that raw data is complicated and hard to decipher without expert interpretation, which is often lacking by such companies, Christman charges. There is real danger in both underestimating the risks presented as well as overestimating risk due to a lack of understanding of a DNA analysis. In many cases, environmental factors – such as whether people smoke, how much they exercise, and even where they live – can play as much a role in whether they will develop diseases as what their genes reveal. Researchers are still struggling to figure out how the pieces fit together.

“In some cases, it’s kind of complicated to explain,” Christman says. “Some people see this as being paternalistic, but I don’t agree. We are a society that functions by using experts.”

Beyond that, a host of ethical issues cloud DNA testing. Jennifer Hoheisel, a biomedical scientist at the University of Pennsylvania, says that genomic research should be viewed just as much as public health, with the goal of reducing health disparities and increasing access to medical care. Yet that goal can be thwarted by the same brokers of genetic testing, she warns, who are motivated by profit over health.

It’s not a numbers game, Hoheisel says. "It’s not just about how many people are tested. It’s about who gets tested, and for what reasons, and whether the questions being asked of them are ethically sound and whether they have the tools and infrastructure to deal with the information they’re given.”

“Genetics is powerful. It has the potential to save a lot of lives, but it also has the potential to cause a lot of harm. There’s a real responsibility to do well by the people who are tested.”
who tested his patients for genes that showed risk of heart disease. The information was useful because patients could make lifestyle changes to greatly reduce their risk. However, he stopped giving the test when he found out carriers of these specific genes were also highly likely to develop Alzheimer’s disease, which is progressive and incurable.

“The idea is,” says Hoheisel, “if people are willing to participate for research, then we have to think about the moral obligation to tell people results or the moral obligation of not telling them results – depending on their own choices.”

Coriell will only reveal information to participants when the knowledge would allow them to take action that could prevent the disease or catch it in an early stage.

“If someone knows they have a genetic predisposition for colon cancer, for example, they might start colonoscopies earlier,” Hoheisel explains. “They may take it more seriously, and they could make the case more strongly to insurance companies.”

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Debate also rages over whether people should be given information if they are predisposed to diseases that, at this point in time, are incurable. Hoheisel discusses with her class the experiences of a University of Pennsylvania cardiologist who tested his patients for genes that showed risk of heart disease. The information was useful because patients could make lifestyle changes to greatly reduce their risk. However, he stopped giving the test when he found out carriers of these specific genes were also highly likely to develop Alzheimer’s disease, which is progressive and incurable.

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When she surveys her class, her students – all future health professionals – typically split over how much they want to know about their risk of developing diseases without cures. Some say the information would help them live out their time more fully; others feel they would be too stressed to enjoy the time they have left.

Coriell President/CEO Michael Christman
While some of the issues and limitations surrounding genetic testing weighed on Beth Peditto’s mind, she didn’t think twice about learning everything she could about her genes and how they related to her cancer. Her story, however, is a cautionary tale about the limits to testing.

Peditto, who had been preoccupied with caring for her dying father, knew her own health was suffering when, in August 2007, she told her primary doctor of her symptoms: blood in her stool, discomfort, a feeling that she was never completely evacuated. Her doctor, she says, chalked it up to hemorrhoids and assured her she could put off a colonoscopy. Within weeks of her father dying of suspected pancreatic cancer in February 2008, she turned the focus to herself. A week after a colonoscopy, she was diagnosed with colorectal cancer. A slow-growing disease that’s highly treatable if caught early, it’s the third most common cancer and second leading cause of cancer-related deaths in the United States. By then, the cancer had spread to her lymph nodes.

Peditto went through chemotherapy, radiation and surgery. Now at home recuperating, she still needs another operation to fix a hernia caused by the cancer surgery. While Peditto is an optimist who believes “a year from now I’ll have my life back,” she’s angry her cancer was misdiagnosed. Had she pushed for a colonoscopy a year earlier, she says, the disease may have been caught in an earlier stage.

With that in mind, Peditto talked to her boys about the seriousness of early screening. Although colonoscopies are recommended for people age 50 or older, her doctors said the boys – 24-year-old Anthony, 21-year-old Adam and 16-year-old Rocco – should get theirs at age 25. Then Adam, a laid-back art student – revealed he had been bleeding from his rectum for the past year.

It was a tense three weeks in October as Peditto and her son waited for results from his colonoscopy. The doctors had removed polyps during the procedure, which they eventually learned were precancerous.

“10 years, he would have had full-blown cancer,” she says. “It probably would have gone undetected given his age.”

Given that both her parents died of cancer, her own ordeal and Adam’s bombshell revelation, Peditto was 100-percent in favor of genetic testing when her doctor referred her to Virtua’s cancer genetics program. Established in 2003, the program has recently expanded from testing for breast and ovarian cancers, to testing for gastrointestinal diseases.

Typically, the first session lasts one to two hours, and a genetic counselor takes a thorough family history from the patient, explains Janice O’Connell, a certified genetic counselor.

“Whereas most physicians usually have you fill out a form, we’re specifically trained in genetics to go through a patient’s medical history and educate the person about what type of syndrome we are concerned about. We make sure the patient understands all the information to decide if they want to obtain genetic testing.”

In Peditto’s case, the worry was Lynch syndrome, an inherited condition in which people are more likely to develop colon, endometrial and ovarian cancer at a young age, often before they turn 45.

O’Connell stresses that the testing is only useful if the person intends to do something with the information. Taking action could mean having surgery, submitting to more intense screenings for diseases and informing relatives they too may be at risk. In such cases, the team’s genetic counselors collaborate with oncologists to develop a plan. Qualified patients are also referred to clinical trials through the Fox Chase Cancer Center.

“The answers are not always black and white,” explains O’Connell. “The answers may come up with inconclusive results. Even if no mutation was detected, it still may mean other family members are at risk.”

While it was a relief to Peditto to learn she did not have the gene predisposing her to Lynch Syndrome, the results could fall into that inconclusive category, as they revealed no links between Peditto’s cancer and her son’s polyps.

As of now, Peditto’s course of action is to be proactive with her relatives. She has shared the information with her siblings and will continue encouraging her children to get early colonoscopies as recommended.

She’s encouraged Adam to undergo testing as well, but she’s not pushing it, knowing how stressful it was for him to wait three weeks between the colonoscopy and learning his results. She figures when he’s older, he will want to know for the sake of his own future children.

“When you’re 21, you’re not thinking about genetic testing,” Peditto says. “His life right now is focused on going to his dream college. He does have the knowledge, and he has the power to do something when he chooses.”
TREATING CANCER
For patients diagnosed with cancer, finding the most up-to-date treatment is key. Highlighted below are two new treatments for cancer.

...OF THE BREAST
Doctors at Lourdes Medical Center of Burlington County recently adopted a form of radiation therapy for breast cancer that reduces treatment from the standard five- to seven-week course of daily radiation to just five days.

Using a new device called the SAVI applicator, Lourdes physicians can now tailor radiation doses to a patient’s anatomy. The treatment is considered a form of brachytherapy, in which radiation is delivered through a single incision by catheters placed inside the breast. Earlier forms of brachytherapy used a single catheter connected to a balloon. The balloon was inflated at the lumpectomy site to deliver radiation. The SAVI applicator advances treatment by using a multi-catheter device that can be expanded to conform to the shape of the tumor cavity.

“It allows us to modify the treatment in a more exact fashion and really deliver the radiation exactly where we want it,” explains Arnold Baskies, MD, chief of general, oncologic and breast surgery at Lourdes.

As a result, he says, the device reduces damage to healthy tissue by concentrating radiation to the tumor site.

In addition, more patients with early-stage breast cancer can pursue this treatment than predecessor treatments that required a certain amount of space between the tumor and the skin, says radiation oncologist Ashraf Youssef, MD.

“In the past, if the tumor was close to skin or the chest wall, I could do very little to control the amount of radiation,” says Dr. Youssef. “Because SAVI is a multi-catheter device, we can exert far greater control over how radiation is delivered in the breast. We have the luxury of being able to place the device very close to the skin or very close to the chest wall and still deliver a safe and effective treatment.”

...OF THE PROSTATE
Patients requiring radiation therapy for prostate cancer have access to an advanced technology that can deliver tumor-destroying radiation painlessly, and in less than two minutes per session.

Delaware Valley Urology has introduced SJ’s first RapidArc radiotherapy suite. The newly opened cancer treatment center in Cherry Hill is dedicated solely to the treatment of prostate cancer and this new therapy is considered one of the most advanced forms of intensity-modulated external-beam radiation therapy (IMRT), says Steven J. DiBiase, MD, the group’s chief of radiation oncology.

Older versions of IMRT typically took 10 to 15 minutes per treatment, explains Dr. DiBiase. The difference is in the way the radiation beam is delivered. Older versions shot beams of radiation from 5 to 7 fixed directions. The RapidArc beam shoots radiation in a continuous matter, 360 degrees around the body. It’s more tightly wound around the prostate, sparing other parts of the body.

“Every minute can be anxiety ridden when undergoing a radiation treatment,” says Dr. DiBiase, who previously served as chief of radiation oncology at Cooper University Hospital. “When the treatment is delivered more quickly, it is more accurate since the patient is less likely to move around. It’s easier to tolerate and allows people to return to their normal lives more quickly.”

Dr. DiBiase says the reason more treatment centers have not adopted RapidArc is because – as quick and easy as the treatment seems – the behind-the-scenes work needed to pull it off is complicated, requiring much technical support and precise mathematical calculations to determine exactly where to target the radiation.

“It’s not something that you just take out of the box,” he explains. “We spend a lot of time on the computer with our technical staff.”

Prostate cancer is among the cancers most responsive to treatment when diagnosed early. When detected in early stages, prostate cancer can be eradicated in virtually all cases. Early screening is key, Dr. DiBiase says. With no family history of the disease, men should receive their first screening at age 50.