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Answers in the genes : Testing, counseling help patients make medical decisions

DAVE MASON, NEWS-PRESS STAFF WRITER

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Liz Beckner knew it was time to stop rolling the dice in a life-and-death game called genetics.

The Santa Barbara resident, 38, made that decision after her baby, Dashiell, died in 2010 when he was 8 months old from Zellweger syndrome, which caused seizures in their boy.

Mrs. Beckner and her husband, Ben, 39, didn't know they were carriers of the gene that causes Zellweger syndrome until Dashiell was diagnosed when he was 6 weeks old. She talked about the baby and the parents' efforts now to have twins as she sat cross-legged on a bed in a private room at Santa Barbara Cottage Hospital, where she is on bed rest until the boys are born. (Doctors would like her to give birth ideally no sooner than her 34th week of pregnancy, which comes in September.)

The Beckners wanted to try again to have a baby, but didn't want the heartbreak of watching another child die. So they turned to genetic testing to make sure the next child didn't have Zellweger syndrome.

They had embryos fertilized in vitro. The Santa Barbara Fertility Center implanted two without the disease in Mrs. Beckner. (A third remains frozen.)

"We didn't have to roll the dice," she said.

Still, the prognosis is good for one twin, Nolan, but poor for the other, Judah, because of complications not related to genetics.

More and more, people are using genetic counseling and testing from blood or saliva samples to determine the risks of developing a disease or passing one on to future generations as well as the best treatment.

Physicians and counselors say testing is commonly done in the fields of prenatal care, pediatrics and oncology.

"It's becoming a big thing in just the last decade," said Megan McKenna, who started in July 2010 as a genetic counselor at the Cancer Center of Santa Barbara.

Mrs. McKenna, 26, said the mapping of the human genome in 2003 as well as the move toward more personalized medicine has revolutionized genetics. "They're looking at small variations in our DNA that can show how we react to our environment and can increase the risks for conditions such as heart disease and ... colon cancer."

"Personalized medicine is looking at these small variations," Mrs. McKenna added. "What are you at risk for? What are you not at risk for? Are there particular medications that you might react better to? The answers are based on your genetics."

In the past, family history has been something a doctor briefly discusses during a physical. Genetic counseling delves deeper.

"I can spend anywhere from 30 minutes to an hour going over family history," said Mrs. McKenna, who earned a master's degree in genetic counseling in 2009 at Cal State Northridge. "I ask about all the cancer diagnoses in the family. I ask about the ages of diagnoses and characteristics of the cancer as well."

To demonstrate where the questioning leads, Mrs. McKenna used a genetic family tree for one patient, in which the squares represent males; the circles, females. Diagnoses are marked in black.

"This particular patient had a melanoma diagnosis at 7. Her mother had a breast cancer diagnosis at 34. A maternal aunt had a brain cancer at 27," Mrs. McKenna said. "Another maternal aunt had breast cancer at 29. A grandmother had ovarian cancer at 40."

She uses several statistical models to determine the patient's risks for developing cancer, saying the most common genes for hereditary cancers are BRCA1 and BRCA2 — breast and ovarian cancer.

Mrs. McKenna said reviewing the family tree is less about statistical probabilities and more about recognizing patterns. "We look at what kinds of cancers are in the family. We're trying to determine which cancer syndrome we should be testing for."

Information from counseling can influence decisions on treatments, as it did for a patient sitting across from Mrs. McKenna on this day.

"I found a lump in my right breast in May," said Nadine Dwan, 31, a Santa Barbara health insurance broker. "My OB/GYN referred me for an ultrasound." A biopsy tested positive for breast cancer.

She said genetic counseling helped determine it was best to have both breasts removed. After finishing chemotherapy, Mrs. Dwan plans to undergo a bilateral mastectomy because of the risk she'll develop cancer in the other breast.

Genetic counseling can also be used to help parents make decisions about whether to have children or prepare for diseases their offspring may inherit.

"Most of the time, it reassures parents that their child will be healthy," said Dr. Alex Soffici, director of maternal-fetal medicine in the Cottage Health System,

He explained testing can show whether children will be born with congenital diseases, cystic fibrosis, or developmental disabilities such as Down syndrome.

"Under California law, all pregnant women are required to have been offered genetic screening by their obstetrician."

Dr. Soffici, 51, explained testing can be about whether the unborn child has a missing, extra or damaged chromosome.

In addition to family history, doctors are looking at anatomical evidence in the ultrasound. A bulge in a

neck, said the doctor, might indicate Down syndrome, and that can be confirmed by DNA testing. Down syndrome is among the diseases for which there's an extra chromosome.

Like the Beckners, many parents don't know they're carrying genes for certain diseases, and testing alerts them, noted Dr. Rene Allen, president and physician of the Santa Barbara Fertility Center. Carrying a gene doesn't necessarily mean the child will get it, and for many diseases, both parents must have the gene for the unborn child to inherit it, he said. "If we know both parents are carrying the disease, we can test the embryo."

He said the mapping of the human genome has greatly enhanced the ability to screen for genetic diseases.

But Dr. Allen, 35, added, "No test will be 100 percent accurate." He said there's always a small chance of error.

Testing is one way to take chance out of genetics, but can pose ethical questions given that it can mean picking a healthy embryo over one with likely birth defects.

Doctors and genetic counselors, though, say the ultimate goal is to protect the health of the patients and their children.

Genetic exams are also being used in pediatrics to seek the underlying causes of developmental disabilities, said Dr. Gary Feldman, a Ventura pediatrician and medical geneticist who works at Tri-Counties Regional Center in Oxnard.

"Genomatic medicine is the next big thing," Dr. Feldman, 68, said. "I had it happen to me."

He needed Plavix, a blood thinner, but couldn't use it because of a genetic defect that doesn't allow him to sufficiently metabolize the drug. Genetic testing showed his problem, and after trying another drug he couldn't use because of its side effects of headaches, he now takes aspirin to thin his blood.

Dr. Feldman said he sees this in the future — the right medications for individuals will be based on their genetic makeup.

That can mean one less toss of the dice as people try to avoid gambling with their health.

email: dmason@newspress.com

FYI

?For more about the Hereditary Cancer Risk Counseling Program, call the Cancer Center of Santa Barbara at 898-2204 or go to www.ccsb.org/genetics.

Santa Barbara Fertility Center is at 536 E. Arrellaga St., Suite 201. For more information, call 965-3400 or go to www.santabarbarafertility.com.