

Real-time Testing for PKU

First-ever self-monitoring device is under development for rare genetic disorder.

BY T.J. BECKER

Home-testing kits have made it easier for diabetics and hypertension patients to track their conditions, but such self-monitoring hasn't been possible for people with phenylketonuria (PKU). Georgia Tech researchers and an Atlanta start-up company hope to change that by introducing the first home-testing device for PKU patients.

PKU is a genetic metabolic disorder in which the body lacks a liver enzyme needed to process phenylalanine, an essential amino acid, into another amino acid (tyrosine) used by the body. Left unconverted, excessive amounts of phenylalanine in the bloodstream are toxic to brain tissue and the central nervous system; if untreated in newborns, PKU can cause brain damage and mental retardation.

There is no drug that can cure PKU. It can only be treated through diet. "When phenylalanine levels become too high, PKU patients can suffer movement disorders, such as tremors, seizures and hyperactivity," explains project director Jeff Sitterle, chief scientist at the Georgia Tech Research Institute (GTRI).

Blood-testing requirements vary, but frequent monitoring is very important for children. Frequent testing can be costly and inconvenient for patients who must travel long distances to reach a center.

Because PKU is considered an "orphan disease" — one that affects a small sector of the population — large pharmaceutical companies haven't pursued a home test for the disease.

"Incidence of PKU varies around the world," says Richard Shunnarah, president of Atlanta-based MetGen Inc., which is developing blood-monitoring devices for metabolic genetic disorders. In the United States, PKU occurs in about one in 10,000 births, he adds.

Shunnarah launched MetGen in 2002 and approached Georgia Tech for help in developing a PKU home test. The testing device will store data, providing a history for doctors to review during patients' routine checkups. "That gives doctors a true trend picture, rather than blood levels that might have resulted from patients making a sudden effort to stick to their diet prior to the checkup," Sitterle adds.

The project is a collaboration between GTRI engineers, who built the device's electronics, and Regents Professor of Chemistry and Biochemistry Sheldon May, who developed a special reagent strip for the device. Graduate student Veronica de Silva assisted May.

Like diabetics using blood-glucose home tests, PKU patients prick their fingers and then place a drop of blood on the reagent strip inserted in the testing device. Phenylalanine in the blood prompts a reaction on the test strip. Then the device's electronics calculate how fast the strip absorbs a specific color of light — indicating the level of phenylalanine in the blood.

"That may sound simple, but it actually requires thousands of measurements and a mathematical algo-

rithm to pinpoint the right section of data to evaluate," explains Tim Strike, associate head of the Technology Application Branch in GTRI's Electronic Systems Lab.

As they intended, researchers limited consumer costs by using off-the-shelf components. "Because PKU is an orphan disease, we wanted a device that would be affordable to produce in smaller numbers," says project collaborator Ron Bohlander, director of GTRI's Commercial Product Realization Office.

Another challenge was user-friendliness. "If you have too many steps to follow, it can be confusing for new users — and downright irritating when you use it on a regular basis," Strike adds. "We wanted to make this as easy as buying a soft drink from a machine."

This past summer, scientists at Emory University — where Georgia's only PKU treatment center is located — worked to validate the technology. Since then, Georgia Tech has been helping MetGen find manufacturers for the chemical strip and electronic device. MetGen hopes to win FDA approval by the end of 2004 and begin marketing the PKU home test in early 2005.

Earlier this year, MetGen received an \$85,000 grant from the Georgia Research Alliance's (GRA) Innovation Fund. "Without GRA's assistance, we would have had to seek outside investors, and that would have been difficult due to the size and uniqueness of this market," Shunnarah adds.

© Contact: Jeff Sitterle, GTRI, 404-894-1254 or 404-894-3369 or jeffrey.sitterle@gtri.gatech.edu; or Richard Shunnarah, MetGen Inc., 404-256-1464 or rshunnarah@metgenco.com. Read more at: gtresearchnews.gatech.edu/reshor/rh-f04/pku.html

Researchers Tim Strike, left, and Ron Bohlander are among the Georgia Tech researchers working with Atlanta start-up company MetGen Inc. in the development of the first home-testing device for PKU patients.



PHOTO BY NICOLE CAPPELLO