Recipe for Survival

METABOLIC DISORDER CAMP HELPS YOUNG WOMEN LIVE FULL LIVES

Were Jean Handler suddenly capable of consuming whatever she liked, she would be a dedicated chocoholic. She would be a connoisseur of curly fries and a faithful patron of pizza parlors. She would eat ice cream and pancakes and ham and cheese omelets made of real scrambled eggs. Her cravings, at least in the first few glorious weeks of dietary freedom, would not be denied.

As it is, however, Handler adheres to a nutritional regimen that would make even the most abstemious vegan break down and cry: no meat, no fish, no poultry, no dairy, and no bread. That's because Handler, who turned twenty-three this summer, has a life-threatening metabolic disorder called maple syrup urine disease (MSUD). An exceedingly rare genetic condition—it strikes just one out of about 185,000 infants every year—MSUD is caused by the deficiency of an enzyme in the body that metabolizes protein. Left untreated, that deficiency leads to a build-up of certain amino acids in the blood and urine, causing irreversible brain damage and death.

So to stay healthy, people with MSUD, as well as those with a slightly more common metabolic disorder called phenylketonuria (PKU), must limit their intake of high-protein foods, the main source of the amino acids that their bodies cannot break down. They also have to supplement their diet with a special, and rather odoriferous, formula rich in the nutrients that they don't get elsewhere. "Oh, it smells horrible," says Handler. "But I'm very disciplined."

As she should be. Handler is, after all, a veteran of Emory's annual, research-based Metabolic Camp, which offers young women with PKU and MSUD a holistic approach to managing their disorders. Through activities like cooking classes and cooperative sports, field trips and discussion groups, campers can come together, have fun, and support one another as they make their own way through life.

"We focus on teaching these young women how to be independent, how to maximize and improve their quality of life," says Rani H. Singh, professor of genetics and director of Emory's Metabolic Nutrition Program, who founded the camp in 1995. "When I first started, it was a very emotional experience. These girls were very lonely. They had never met other girls with PKU or MSUD, and there was a lot of sadness."

Now, with Internet chat groups and the emergence of vegan clinics across the country, she says, things have improved. "At least now they're aware of other young women with the same disorder."

Males are affected, too, Singh adds; but one of the objectives of the camp is to educate young women about the consequences of going off-diet—for their own health and for that of their children.

The newborn screening program includes among its arsenal of tests for genetic diseases the Guthrie test, named for Robert Guthrie, the scientist credited with pioneering the screening for PKU in the late 1960s. Not until 2000, however, did the National Institutes of Health recommend, for all people with PKU, a low-protein "diet for life."

It used to be, says Singh, "that kids were taken off-diet at five or six because by that age, it was believed, the brain is fully developed." Years later, scientists learned they were wrong: adults who had
been diagnosed with PKU as newborns had suffered a drop in IQ as well as neurological degeneration.

No less troubling was the finding that many of the children these women had given birth to were severely delayed or mentally handicapped themselves. “We call that maternal PKU,” Singh explains. “The babies don’t have PKU, but the high level of phenylalanine [an amino acid essential to human nutrition] in the mother’s blood is very teratogenic to the fetus—even more so than fetal alcohol, and the symptoms are worse.”

Singh published a paper with the Centers for Disease Control and Prevention reporting that among seventeen women who had gone off-diet, sixteen had mentally disabled babies. Still, she adds, there was reason for hope: a national maternal PKU study published around the same time found that if women with PKU went back on-diet prior to getting pregnant, they were capable of having a normal, healthy baby. The problem, Singh knew, was simply a lack of awareness.

“I felt it was critical to catch these women early on and to teach them about prevention,” she says. “So we started the camp.”

Anne Allison Parker, a camp alumna with PKU, recent college graduate, and now an elementary school teacher in north Georgia, recalls attending the inaugural session in 1995, the youngest of the crew. She looked forward to that week every summer—the games, the new recipes, the friends who understood.

“I think my favorite was the time we made imitation cheesecake—the old kind with the marshmallows,” she laughs. “I licked the bowl clean.”

These days, she says, the camp experience has taken on new relevance as she and her husband plan a family. According to Singh, since the camp began fourteen years ago, seven women have gone on to have children; only one had a mentally disabled child.

“So we’re definitely having an impact,” Singh says. “Even if they’re not concerned about their own health, we’re able to educate them about the consequences for their baby, and they go back on-diet.”

Unfortunately, says Singh, there’s one challenge that the camp can’t help with: the cost of the special formula that gives people with amino acid metabolic disorders the vital nutrients that they can’t get from the food they eat. Because the formula is considered a food, not a drug, many insurance companies refuse to cover it, likening it to a dietary supplement such as Boost. “The average cost of the formula in our clinic is about $7,500 a year,” says Singh. “That is a huge financial burden for some young couples.”

Some states do have legislation requiring insurance companies to prepay for medical foods and specialized formulas. Georgia is not one them, but that didn’t deter Parker from making her case. She called her insurance company every day for a month and assembled a thirty-page packet of information that she had researched, highlighting key points. “I made them realize that it would cost them more in the end if they didn’t pay for the formula,” she says.

Back in Boca Raton, Jean Handler has her hands full as well—with a boyfriend, homework, exercise, and classes at her community college. Handler plans to become a psychologist for troubled teens once she finishes her degree. But she needs a new liver first, so she’s waiting for a transplant. If successful, it would free her from diets and formulas forever, from the frustrations of eating out and those awkward moments on dinner dates.

That doesn’t mean, of course, that she’d quit going to Metabolic Camp. She’s been looking forward to her fourteenth session for months. “I often think that my disorder has made me who I am,” Parker says. “You know, very strong-willed, very self-disciplined, able to rise to a challenge. I guess that’s what happens when you have to work hard just to eat.” —Patrick Adams 08MPH