Study reveals new options for people with PKU

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(PhysOrg.com) -- For people with the genetic condition known as phenylketonuria (PKU), diet is a constant struggle. They can eat virtually no protein, and instead get their daily dose of this key macronutrient by drinking a bitter-tasting formula of amino acids. Yet drink it they must; deviating from this strict dietary regimen puts them at risk of developing permanent neurological damage.

In the near future, fortunately, a better option may become available.

In April, a team of University of Wisconsin-Madison researchers will publish the second of two key papers showing that a unique protein derived from whey — known as glycomacropeptide, or GMP — is safe for people with PKU to eat. GMP is the first known natural protein that is safe for this group, and these findings are poised to revolutionize the PKU diet. Already, Cambrooke Foods, a Massachusetts company that specializes in the manufacture of medical foods, is in the process of developing GMP-fortified snack foods for commercial sale.

"It's so important to individuals on the PKU diet to have new options, to have their diet liberalized. It's a quality-of-life issue," says Denise Ney, a professor of nutritional sciences who led the two studies. "Adolescents have an especially difficult time [staying on the diet], but it's so critical that they do."

People with PKU are born without the enzyme responsible for breaking down phenylalanine, one of the 20 major amino acids that form the

proteins we eat in everyday foods. While small amounts of phenylalanine are essential for PKU patients, excess amounts stay in their bodies indefinitely and interfere with brain function. Those who go off-diet often suffer from concentration problems and depression. Some even sustain permanent brain damage. The GMP protein isolated from whey, on the other hand, is the only known dietary protein that contains only trace amounts of phenylalanine; absolutely pure GMP, in fact, is completely phenylalanine-free.

The first GMP human feeding trial was published in February in the Journal of Inherited Metabolic Disorders. In it, Ney and her team describe the experience of an individual with PKU who volunteered to consume an all-GMP diet for 10 weeks. As the paper explains, not only did the subject enjoy the GMP-fortified snack bar, pudding and sports beverage that supplied most of his daily protein, but the amount of phenylalanine in his blood actually starting going down after he ate these items for a couple of weeks.

"And because the subject enjoyed the GMP foods, he was more inclined to eat them throughout the day, which helps keep the body's protein metabolism running efficiently all day long," says Ney. "When he went back to the amino acid formula, he went back to drinking it all in one sitting."

Ney's new study, publishing in the April issue of the American Journal of Clinical Nutrition and appearing online on Feb. 25, describes an 11-person trial of shorter duration involving PKU patients receiving care from UW-Madison's Waisman Center who agreed to spend six days at the University Hospital's Clinical and Translational Research Core. In the study, subjects adhered to the amino acid formula diet for four days, and then switched to the GMP diet for the following four. (Subjects spent the first two days of the study at home.) In the end, no adverse health problems were found, and 10 of 11 subjects claimed to prefer the

GMP diet, making the bottom line of this study the same as the first — that GMP is safe and acceptable.

In this shorter study, variations were seen among individual subjects, but the overall blood phenylalanine levels measured after meals were comparable in the two diets. Additionally, the GMP diet improved protein metabolism compared to the amino acid formula.

Members of the PKU community have been eagerly monitoring the progress of this research project for nearly a decade, since UW-Madison food engineer Mark Etzel published a key paper describing how to isolate GMP from whey. A few years ago, when things got to the point of starting human feeding trials, the researchers discovered they already had a long list of eager volunteers, including Matt Cortright of Wausau, Wisconsin, who developed seizures and other problems after abandoning the amino acid formula for a few years during early adulthood.

"The main reason I helped out with this study was so that newer generations won't have to go through the things I have. It makes me happy to see how far the studies have come since I was younger, and the choices future generations will have," writes Cortright, now 32, in a letter to Ney. "I just hope that I will also have the opportunity to use these [GMP food] products [in my own daily life] as well."

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