

Adults with genetic disorder PKU need to get back to the clinic

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Genetic researchers at Children's Memorial Hospital, Chicago, are aggressively identifying adult patients who suffer from the genetic disorder, Phenylketonuria (PKU), and are presenting those findings at the 11th International Congress of Inborn Errors of Metabolism in San Diego, August 29 through September 2.

The findings have also been published in the current issue of [Molecular Genetics and Metabolism](#).

PKU is a lifelong [genetic disorder](#) in which a deficient enzyme prevents the body from metabolizing an essential amino acid, called Phenylalanine (Phe), which is found in most foods including meat, bread, eggs, dairy, nuts, and some fruits and vegetables. When left untreated, PKU patients who consume too much Phe are at risk for severe neurological complications, including IQ loss, memory loss, concentration problems, mood disorders, and in some cases, severe mental retardation.

Barbara Burton, MD, Director of the PKU and Metabolic Disease Program at Children's Memorial and Lauren Leviton, Education Coordinator in the Division of Genetics at Children's Memorial lead the adult PKU Outreach program. Through the program, the team has been able to identify and treat several adult PKU patients who have returned to treatment.

"It is so important for adults with PKU to know that it is never too late to

address the disorder and resume treatment," said Dr. Burton. "When PKU is no longer managed, that is when we see patients who suffer from mood and social disorders. For adults, this can also impact their concentration at work, stability in relationships and social interactions. As a healthcare provider, I feel that it is our responsibility to educate adult PKU patients and offer comprehensive medical care and social support systems."

Full results and an overview of how to develop such a program will be presented at the Congress.

Source: Children's Memorial Hospital

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