CANADIANS AFFECTED BY RARE, BRAIN-THREATENING DISORDER MARK FIRST PKU AWARENESS MONTH IN MAY

Improved access to specialized care and treatment for PKU is needed throughout patients' lives

TORONTO, ON (May 9, 2012) – During Canada's first PKU Awareness Month, the Canadian PKU and Allied Disorders Inc. (CanPKU) is striving to increase awareness and understanding of the tremendous challenges faced by patients living with the rare, inherited metabolic disorder known as phenylketonuria (PKU). At birth, all Canadian babies are tested for PKU through government-provided newborn screening programs [i] so that treatment can begin immediately, however lifelong specialized care and treatment is needed to protect patients' brains from harm throughout their lives.

"Over forty years ago, Canadian governments were amongst the first jurisdictions to introduce universal newborn screening programs for PKU, enabling treatment for these vulnerable patients to begin immediately to protect against brain damage," said CanPKU president and CEO John Adams, whose 25-year-old son lives with the disease. "It is our hope that with increased awareness and education, our governments will expand their commitment to screening for PKU with lifelong access to all the tools in the toolbox for patients living with this brain-threatening disease."

Risk of neurocognitive impairment

PKU occurs when the body is unable to process phenylalanine, or "Phe," an essential amino acid found in the protein in most foods. [ii] The standard treatment for PKU, involving a lifelong adherence to a severely restrictive low-Phe diet reliant on synthetic formulas and medical foods, can also lead to negative health impacts including nutritional deficiencies, weight issues and bone disease. [iii] Despite patients' best efforts in dietary planning, adherence to the low-Phe diet and blood Phe level monitoring, many are still unable to adequately control their Phe levels, leaving them at risk for neurocognitive impairment with psychosocial impacts. [iv]

"From infancy to adulthood, individuals with PKU and their families experience the negative impact of the disease in profound ways," said Dr. Komudi Siriwardena, metabolic geneticist and medical director of the PKU program at The Hospital for Sick Children (SickKids) in Toronto. "Many adult patients require improved access to specialized clinics, while older children and teenagers can have difficulties adhering to the challenging dietary regimen and require access to additional treatment options to help them achieve the best possible outcome."

Lifelong access to specialized care and treatment

PKU, which is diagnosed at birth through bloodspot collection, is found in approximately one in 12,000 to 15,000 infants born in Canada. [v] Bloodspot collection is a simple procedure that involves pricking the heel of a newborn baby to collect a few drops of blood, which are sent to a specialized laboratory to test for certain rare genetic disorders. [vi] Once PKU is diagnosed, it is critical to the child's brain development and function that treatment is initiated immediately and maintained throughout life.

"I feel incredibly fortunate to have been born two years after newborn screening for PKU was introduced in Ontario," said Kelli Gibney, a PKU patient living in Sutton, Ontario. "The needs of PKU patients like me extend beyond the introduction of dietary treatment at birth, and improved access to specialized care and treatment options would help us live well with the disease."

On May 10 in Ontario, and May 15 in British Columbia, PKU patients and their families will meet with their elected members of provincial parliament to ask that PKU Awareness Month be recognized in their respective legislatures, and to share their personal stories about the challenges they face living with this brain-threatening disease.

About Canadian PKU and Allied Disorders Inc.

Canadian PKU and Allied Disorders Inc. is a non-profit association of volunteers, dedicated to providing accurate news, information and support to families and professionals dealing with PKU and similar, rare, inherited metabolic disorders. Our mission is to improve the lives of people with PKU and allied disorders and the lives of their families. By allied disorders we mean other rare, inherited metabolic disorders also detected by newborn screening. For more information, visit **www.canpku.org,** and our new comprehensive resource for patients and families, **PKU and the Brain.**

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For more information and to schedule an interview, please contact:

Beth Daniher
Cohn & Wolfe
647-259-3279
beth.daniher@cohnwolfe.ca

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