What Is Phenylketonuria?

Phenylketonuria (PKU) is a rare genetic condition that causes an amino acid called phenylalanine to build up in the body. Amino acids are the building blocks of protein. Phenylalanine is found in all proteins and some artificial sweeteners (Aspartame). Your body uses an enzyme called phenylalanine hydroxylase to convert phenylalanine into tyrosine, a nonessential amino acid. Your body needs tyrosine to create neurotransmitters, such as epinephrine, norepinephrine, and dopamine.

PKU is caused by a defect in the gene that helps create phenylalanine hydroxylase. When this enzyme is missing, the body is unable to break down phenylalanine. This causes a build-up of phenylalanine in the body. Early diagnosis and treatment can help relieve symptoms of PKU and prevent brain damage.

Ireland is one of the leading countries in Europe with instances of PKU, approximately one in every 4,500 babies born in Ireland have PKU in contrast to the United Kingdom where it is approximately one in every 12.000, and further afield in The United States where the condition is even rarer, only affecting about 1 in every 15,000 newborns each year.

In Ireland we follow “Diet for Life” where each PKU sufferer is continuously assessed from birth to determine how many grams of protein they can have on a daily basis. This assessment is determined from blood tests as well as weight and height measurements. These tests are ongoing for life in order to facilitate the healthy growth and development of PKU’s. To supplement the often very low amount of protein that PKU’s can have there are foods which are available on prescription through the Long Term Illness (LTI) card, (eg pasta pizza flour milk & bread), given that Ireland has one of the highest instances of PKU in Europe we have one of the smallest, and ever decreasing, variety of foods on LTI in comparison to our European neighbours. Given all of this and in order to follow a full Low Protein diet it is necessary to purchase a lot of foods from supermarkets which are low in protein. Unfortunately many of these foods (eg violife vegan cheese, fresh fruits & vegetables) are very expensive.

The PKU diet is extremely challenging for PKU patients and their families. The consequences of not adhering to the diet are serious: Untreated PKU can lead to brain damage, intellectual disabilities, behavioral symptoms, or seizures. Ultimately, facilitating PKU patients to adhere to their diet through the provision of a good range of medical and low protein foods, aswell as innovative medicines, will improve their outcomes, resulting in lower overall costs to the healthcare system, aswell as optimizing their ability to contribute to society.

Leaders Questions:

What is the LTI budget spend on PKU products year on year (please provide data from the last 5 years)?
Who is responsible for maintaining the list?
How often do they meet?
What criteria are used in the assessment of new products for inclusion?
How is the decision made to remove products from the LTI list?
Where can we access reports relating to the assessment of products (other than FOI)?
How long does it take for an application to be assessed?
How does the number of products available here compare to other EU countries?
Where can we review which products have been assessed, are waiting to be assessed, and when those in waiting are going to be assessed?
How is the patient/carer's voice provided in the approval process?
How can we, the end users give our input as to the value, or otherwise, of foods?
Is it acknowledged that an increased selection of foods does not necessarily equate to increased costs?

What progress, if any, have the HSE made to date on the National Rare Disease Plan recommendation that a working group to assess reimbursement of orphan medicines and technologies be set up?
Have they set up a working group or when do we hope to see the working group in existence?
The National Rare Disease Plan recommends the setting up of a national budget to fund such treatments so that there is not an impact on individual hospital budgets, example Temple Street.
Have the HSE made any progress to date?

"Domiciliary Care Allowance (DCA) is a monthly payment for a child aged under 16 with a severe disability, who requires ongoing care and attention, substantially over and above the care and attention usually required by a child of the same age. It is not means tested."

Why is it that Some PKU's are receiving Domiciliary Care Allowance whilst others are refused it?

What is the evidence based criteria used by the deciding officer?

Why are some families having to go through appeals in this process?

Surely as all PKU's suffer the same condition with the absolute same restraints and consequences why is there not a blanket Domiciliary Care Allowance payment for all PKU's from birth to 16yrs?

Given the increased costs to PKU's over 16yrs is it likely that having received DCA up to this age they will be in receipt of Disability Allowance when they apply for it past the age of 16?

(obviously with growth comes increased appetite = increased costs)

What are the evidence based criteria used by the deciding officer in these cases past 16yrs?

Further Reading/References:

http://www.hse.ie/eng/health/az/P/Phenylketonuria/Causes-of-phenylketonuria-.html

http://pku.ie/campaigns/