**FACT SHEET**

**Disease Backgrounder: Phenylketonuria (PKU)**

**About Phenylketonuria (PKU)**

- Phenylketonuria (PKU) – is a debilitating, rare genetic metabolic disorder, affecting approximately 1,600 Australians.\(^1\,2\)
- It is an inherited disorder, caused by a deficiency of the PAH (phenylalanine hydroxylase) enzyme.\(^1\)
- A child is born with PKU when both parents are carriers of the defective gene.\(^3\) It is estimated that 1 in every 15,000 newborn babies in Australia will have PKU.\(^1\)
- All newborn babies in Australia are screened for PKU through the heel prick test 2-3 days after birth.
- Parents of a child with PKU have a one in four chance of having another child with the debilitating condition.\(^1\)
- PKU occurs when liver cells are unable to break down an amino acid called phenylalanine (Phe), which is found in most foods.\(^2\)

**PKU & Protein**

- As people with PKU cannot process Phe, they must adhere to a strict, lifelong low-protein diet.\(^1\)
- The amount of Phe needed and can be tolerated by each person with PKU varies, with adjustments to diet required according to blood Phe levels.\(^1\)
- The amount of protein a person with PKU can consume daily varies with development. It is calculated through blood Phe levels, age and growth.\(^1\)
- Maintaining the low-protein diet becomes particularly important during childhood to prevent damage to the growing brain.\(^1\)
- Recommended daily dietary intakes of protein typically include:
  - Adult male without PKU: 64 grams.\(^4\)
  - Adult female without PKU: 46 grams.\(^4\)
  - Person with PKU: 6-8 grams.\(^1\)
  - Pregnant or breastfeeding woman with PKU: 1-3 grams.\(^1\)

**Effects of PKU**

- In people with PKU, an accumulation of toxic Phe levels in the blood may lead to severe neurological damage if left untreated.\(^3\)
- Elevated levels of Phe can be toxic to the brain, leading to poor executive and social cognitive functioning, impairing concentration, mood and energy levels.\(^5\) As a result, people with PKU may experience difficulties with school work, job performance and social relationships.\(^6\)
- Without treatment, babies with PKU show signs of developmental delay and may develop severe brain damage.\(^7,8\)
- Reduced information processing speed in adults with PKU, along with attention deficits, have been found to affect job performance and social relationships, potentially leading to feelings of depression, anxiety and social isolation.\(^8\)
- People with PKU are 50 per cent more likely to develop psychiatric disorders.\(^9\)
- Women planning a pregnancy, or who are pregnant, need to have lower levels of Phe to protect their developing baby.\(^7\) The Australasian Guidelines for the Management of Maternal PKU recommend Phe levels be maintained at 70-250 μmol/L for 3 months prior to conception and throughout the pregnancy.\(^13\)

**Management and available treatment options**

- In Australia, the only currently available treatment option for PKU is through adhering to a strict, lifelong, low-protein diet.\(^1\)
- People with PKU must also support their low-protein diet with the use of Phe-free amino acid supplements, in order to prevent malnutrition.\(^1\)
- Some of the foods people with PKU are limited to on a daily basis include egg replacers, low-protein cheese, low-protein pasta, low-protein bread and low-protein cereal.\(^1\)
- Managing PKU through a low-protein diet and supplementation carries a heavy economic burden on patients and their families.\(^11\)
- Newborns and babies must be fed special Phe-free infant formula before they are allowed to be breastfed.\(^1\)
- Treating PKU with diet alone has been shown to have suboptimal outcomes.\(^6\)
- While people with PKU in other parts of the world have access to the only prescription medicine available to treat the condition, Australians with PKU are still not able to get subsidised access to this treatment through the Pharmaceutical Benefits Scheme (PBS).\(^13\)

**References**