Media Release

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Living with a Rare Disease:
Phenylketonuria (PKU) in the Australian Aboriginal community

Saturday 28 February is World Rare Disease Day which raises awareness about rare diseases and the impacts they have on those affected personally and their families. Rare Diseases are defined as conditions, syndromes or disorders that affect 1 in 10,000 people or less. 80% of rare diseases affect children and most begin in childhood and continue throughout life. In the Australian Aboriginal community, 6-8% of the population are living with rare diseases.

Phenylketonuria (PKU) is a rare inherited disease with significantly harmful impacts if not managed from an early age and with the right foods. PKU prevents the body from metabolising a certain amino acid, a protein building block, known as Phenylalanine. In the Australian Aboriginal community, it is known that there are over 500 people living with PKU.

In PKU, when the body is exposed to too much Phenylalanine and it does not produce the enzyme to break it down, the Phenylalanine builds up in the blood and tissues of the individual. When this happens the phenylalanine build up causes the brain to not develop properly and leads to intellectual disability from infancy which, if allowed to continue, can progress to cause significant impairment and disability.

Professor John Christodoulou, Director of The Western Sydney Genetics Program says “As long as babies, children, youth and adults with PKU are provided the right treatment, a low phenylalanine diet, and their blood phenylalanine levels are maintained within the target range, there is every expectation that the person should be able to lead a normal life. People with PKU are connected with a clinical screening service which runs routine blood tests to help people with PKU manage their special dietary needs.”

“Women with PKU who are considering having babies do need to be especially observant of their PKU monitoring and diet. In the event PKU levels in women are excessive at the time they become pregnant, there is a high risk of harm to the developing infant.”

Newborn Screening services started in NSW in 1964 for Phenylketonuria, and by 1973 all babies born in NSW hospitals were screened using the ‘heel prick’ test.

“People with PKU are entitled to the same treatment, access and quality of care as any other patient. PKU needs a lifetime of monitoring and a lifetime of management to enable people to have the health outcomes they desire.” says Prof Christodoulou.

Wendy Bryan-Clothier, the Aboriginal Health Management Advisor for the Sydney Children’s Hospitals Network says, “I encourage young Aboriginal people and/or the carers of young Aboriginal people living with a rare disease, to continue to seek the support they need from their medical team.”

For ongoing medical support for Aboriginal infants, children and young people with PKU –or for more information about the Phenylketonuria (PKU) clinic, call the Metabolic Clinic: (02) 9845 3452 or visit the website. For more information about Rare Diseases, head to rarediseasedayaustralia.com.au/australia/

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