RESAMPLE NEWBORN SCREENING
PHENYLKETONURIA (PKU)

What is the Newborn Screening Programme (NBS)?
The Newborn Screening programme is a health prevention measure designed to identify babies who may have certain treatable disorders. When treated early enough, babies with these disorders have a much better outcome. When your baby was about 2 days old a blood sample was collected from the heel and sent to the laboratory where it was tested for over 30 disorders. Most of these disorders are treated by diet and/or medication.

Why are babies screened for Phenylketonuria (PKU)?
PKU can slow the development of the brain leading to mental retardation unless treatment is started early. Babies with PKU are not obviously different from unaffected babies so a screening test is needed for diagnosis before any damage is done. Proper treatment leads to completely normal development.

Why does my baby need a second test for Phenylketonuria?
Your baby’s first result was slightly abnormal and the laboratory needs a repeat sample just in case your baby has a mild form of PKU. In the majority of babies the second test result is quite normal and nothing further needs to be done.
If, however, your baby’s second test result remains slightly elevated, the family will be asked to bring the baby to the Children’s Hospital at Westmead for additional tests. At this visit you will also be given a full explanation of PKU and how it is managed.

What is Phenylketonuria (PKU)?
PKU is a rare disorder where one part of protein (phenylalanine) is not broken down or is broken down more slowly than usual. If phenylalanine levels remain high for a long time, brain development slows down and can cause mental retardation.

Slightly high levels of phenylalanine do not cause problems but the baby needs to be monitored to be sure the levels do not rise above a safe level.

PKU occurs in about one in every 10,000 babies born in Australia.

How is PKU treated?
PKU is treated by giving a diet very low in protein. A special protein supplement that does not contain any phenylalanine is prescribed by the specialist doctor to give a balanced diet. When the baby is small, the diet is relatively easy, and allows you to breast or bottle feed plus give the special protein supplement.

Mild PKU may or may not require a special diet but regular blood levels are needed to monitor the level of phenylalanine in the blood.

All children with PKU are monitored regularly using blood from a heel or finger prick, soaked into cards like the newborn screening cards, and sent through the post.