Parent's Information
Newborn Screening for Phenylketonuria (PKU) – a voluntary screening test for Caucasian babies

What is PKU?
Phenylketonuria is a rare genetic condition that results from an inherited defect in an enzyme responsible for metabolism of an aminoacid, *phenylalanine*. Patients with this defect cannot handle ordinary protein load in diet, resulting in accumulation of an excess of certain aminoacids that are toxic to the body. Untreated children will develop brain damage (mental retardation and epilepsy) and growth disturbance.
This is a treatable disease – with appropriate dietary modification started early in life, the child's brain can be protected and he/she can lead a normal life. The earlier the condition is diagnosed and treatment started, the better is the outcome.

Is PKU common?
It occurs in 1 out of 8000-15000 Caucasian babies (data from US and UK). PKU screening is part of the national screening for newborns in most European countries and states of America.

Why does my baby need a "newborn screening" test?
Affected child may not present early and the condition can be missed until he/she presents later with signs of brain damage. There is usually no positive family history to hint the diagnosis. However, abnormal blood phenylalanine level would be detectable 1-2 days after milk feeding has been started. By using a sensitive screening test to detect for this, we can diagnose the condition as early as possible so that treatment can be started.

What is the test like?
What we need to do is to have a heel prick test (figure 1) to obtain a few drops of blood from your child from day 2 to day 7 of life. The test would be sent to a private laboratory for analysis. Your child may feel a bit painful during the heel prick, but it is the safest way to obtain blood sample from newborns.

What would follow the screening if I opt to do it?
The blood sample (Guthrie card) would be sent to a local pathology laboratory (PathLab) for processing. Results will be available within 1 to 2 weeks. The result will be mailed to you within 2 weeks of the test. You will be contacted only if test result is abnormal.

What is meant by an abnormal screening result?
Just like all other laboratory tests, this screening test is bound to have "true" and "false" results. An abnormal result can either be "true positive" or "false positive". In such cases we will call you for a confirmative blood test for your baby, either to confirm or exclude the diagnosis. Fortunately, most of the "positives" are false.
False negative result, i.e. to miss a diagnosis for an affected child, is highly unlikely and should not, in general, be a concern.

What is the arrangement if I opt to have my baby screened?

As PKU screening is not part of the routine newborn biochemical screening for babies in Hong Kong, this test is not available in the public hospitals. To our knowledge, the '*PathLab Medical Laboratories LTD'*, a local private referral laboratory is currently providing such service for babies delivered in private hospitals. If you preferred to have blood taking (the test preferably to be performed within 36-144 hours of life to ensure that the baby was fed with milk for at least 24 hours) before hospital discharge, our doctors can provide assistance with the blood taking and the necessary arrangement. *You have to pay directly to the private laboratory by preparing a cheque with the amount of HK$250.00 (per test) payable to "PathLab Medical Laboratories LTD". This is a direct financial transaction between you and the private laboratory and our hospital/ staff will not be held responsible for any subsequent dispute arising from the test.* If your child is discharged earlier than that, or you want to have the test performed after discharge, we advise you to bring your child to your family doctor and arrange the test with the laboratory. We will provide you with the necessary information if needed.

Results will be mailed to your address, usually within 7-10 days after the test. A copy will be sent to our neonatal unit for follow up action if necessary. If you do not receive the result by 10 days, please call at 2861308 (PathLab).

Can I opt not to have it done?
Yes. It is entirely up to you to decide.

Where can I get more information on PKU?
Since it is a well-known condition occurring worldwide, there are a lot of websites on the internet that provide good information on the condition. Here are a few examples:

www.aafp.org/afp/991001ap/1462.html
www.nlm.nih.gov/pubs/cbm/pku.html

What is the treatment if my child is affected?
An affected subject should receive phenylalanine-restricted diet for life. Regular medical follow-up will ensure a good control of the disease and thus a good quality of life.

Contact and enquiry
If you have any enquiry on the screening test, please feel free to contact the nursing staff at 2255988 during office hours.

Figure 1: heel prick

Disclaimer notes
The test is arranged and done at a private laboratory (PathLab Medical Laboratories Ltd). There are many factors that may affect the delivery of valid samples to the laboratory e.g. preparation of samples, transport, losses due to human errors or unexpected conditions. This hospital and the staff of the Department of Paediatrics and Adolescent Medicine will not be held responsible for the failure of the test.

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