Overview: WA newborn screening

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Newborn screening (NBS) is a public health service available to all infants in Australia. Performed shortly after birth, it involves testing asymptomatic babies to identify and treat problems before they occur. A heel prick blood sample is screened for several different diseases – an abnormal result requires follow-up testing to confirm a diagnosis. Most infants with abnormal screening results will have normal follow-up testing – initial screening results can be abnormal because the blood was taken too early, the baby is premature, and many other reasons. If an infant truly has a disease, treatment is started immediately.

Newborn screening in Western Australia

The WA NBS Program operates from the Department of Clinical Biochemistry at Princess Margaret Hospital for Children and screens all babies born in WA. The disorders screened are shown in Table 1. The Program collaborates with physicians in the Metabolic Clinic (PKU, galactosaemia, amino acid, organic acid, and fatty acid oxidation disorders), Department of Endocrinology (congenital hypothyroidism), and Department of Respiratory Medicine and Genetic Services of WA (cystic fibrosis) at Princess Margaret Hospital to initiate any recall, diagnosis and treatment of infants identified through positive screening results.

Responsibilities of hospitals and midwives

It is the responsibility of the hospital and/or midwife to ensure that a sample is collected and sent to the screening laboratory in a timely manner. The success of newborn screening is dependent on the quality of the samples collected. It is critical that dried-blood samples are taken at the correct time and in the correct manner. The sample should be taken between 48-72 hours of age or on discharge, whichever is sooner. If the infant is discharged before 48 hours, another sample will need to be collected later because false negative results can occur if the sample is taken too early.

Parents must be provided with the information pamphlet *Your Newborn Baby's Screening Test*(1) prior to the blood sample being taken from their baby. The midwife or nurse discusses the collection and testing with the parents, obtains their verbal consent, and completes all relevant documentation. The parents should also be informed they will only be contacted if the test is positive or if another sample is required. Relevant information should be recorded (parental consent details, sample collection details) and information recorded in the mother's medical record/ personal health record.

Advances - tandem mass spectrometry

In December 2004, the WA NBS Program introduced expanded newborn screening for a range of amino acid, organic acid, and fatty acid oxidation disorders (Table 1). These disorders are individually rare but common as a group. This followed the evaluation and introduction of a new screening technology called tandem mass spectrometry (MSMS).

MSMS testing is different. It tests for many disorders at one time whereas before MSMS, there was a one-test, one-disease model in newborn screening. While getting more information from one test would seem advantageous, it creates new challenges for newborn screening programs.

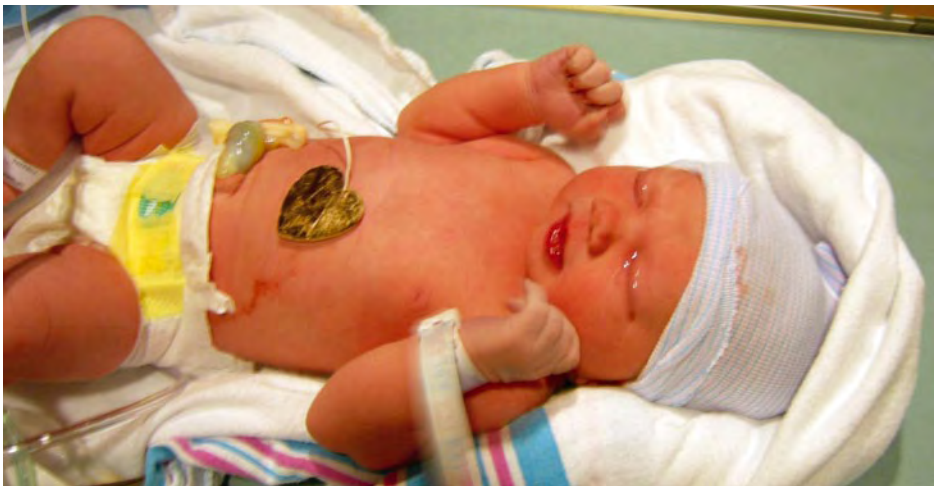

Table 1 : Disorders screened for in WA

Disorder	Screening Started	WA Incidence
Phenylketonuria	1969	1:16,000
Galactosaemia	1980	1:80,000
Congenital hypothyroidism	1981	1:3,500
Cystic fibrosis	2000	1:2,500
Other amino acid disorders	2004	1:15000
Fatty acid oxidation disorders	2004	1:10000
Organic acid disorders	2004	1:15000

Limitations of screening

Screening tests can indicate that an infant may be at risk for a disorder included in the testing panel but additional tests are necessary to determine if the infant actually has a disorder.

Screening tests are capable of both false positive and false negative results. Their accuracy depends on various factors, including the way the specimen is collected, infant's age at testing, birth weight, gestational age, feeding type, transfusion status, medications and the presence of co-existing illness or medical conditions.

Any infant with a normal NBS test result but symptoms indicative of a potential disorder should be followed up immediately.

Children with these conditions can usually live normal, healthy lives if they are detected and started on treatment early In Western Australia, the newborn screening program tested more than 32,000 (99%) babies in 2008. While participation in this effective public health screening program is entirely voluntary, the Department of Health (WA) strongly recommends that all babies are screened for these treatable conditions. ■

For more information refer to the the WA NBS website: http://www.pmh.health.wa.gov.au/services/newborn/index.htm

Ref: (1) www.pmh.health.wa.gov.au/services/newborn/parents/pamphlet_par.htm