



Has Your Infant been Screened for Metabolic Disorders

SCREENING FOR INHERITED METABOLIC DISORDERS

- What is a metabolic disorder?

It is an inherited disorder which affects the way the body changes food and essential chemicals into energy and waste matter.

The conditions screened for in Ireland are:

Phenylketonuria (PKU),
Galactosaemia,
Maple Syrup Urine Disease,
Homocystinuria and
Hypothyroidism

All of these conditions are treatable.

- How do we screen for metabolic disorders?

The Guthrie Test (Heel Prick Test) is a test carried out on new born infants to detect these conditions. A blood sample is taken from the infant's heel not earlier than 72 hours and not later than 120 hours after the infant's birth. If the infant is being breast fed, it is necessary to ensure that the infant has taken sufficient food before the test is carried out.

- Where is the test carried out?

The test may be carried out:

- (a) In the hospital where the infant is born
- or
- (b) by a public health nurse

or
(c) by a general practitioner.

On discharge from hospital, either parent should check if the test has been carried out. If the infant has not had the test either parent should ensure that it is carried out by bringing the infant back to the hospital or to the public health nurse or to a general practitioner not earlier than 72 hours and not later than 120 hours after the infant's birth.

- Why is the test important?

Early detection of a metabolic disorder means that essential treatment can be commenced quickly. This may prevent brain damage, serious physical illness or death.

- The Treatment

The treatment is provided and monitored on a nationwide basis by the

Metabolic Unit,
Temple Street Hospital,
Dublin 1
Phone(01) 874 8763 Ext. 317

The treatment is medication or a special diet for life.

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