Newborn Metabolic Screening

Newborn babies in the state of Oregon are routinely tested for various metabolic disorders. The disorders covered by the screening program are rare, collectively affecting about 1 in 2,000 infants, so the chance that any single infant will be affected is remote. The cost of not diagnosing one of these conditions, both in human suffering and financial terms, is immense, because early diagnosis and treatment can result in normal growth and development. Babies with these conditions appear normal at birth. It is only with time that the biochemical abnormality affects the baby's health and development. By the time clinical symptoms appear, the damage may be permanent.

The goal of the Northwest Regional Newborn Screening Program (NWRNSP), which includes Oregon, is to identify all affected infants before this damage can occur.

A summary of disorders screened by this program include:

Condition	*Incidence*	*Symptoms if not treated*	*Treatment*
Hypothyroidism	1 in 3,000 births	Mental retardation, other brain damage, growth delay	Thyroid hormone
Congenital Adrenal Hyperplasia	1 in 12,000 births 1 in 300 Yupik Eskimos	Addisonian Crisis in all infants; salt wasting in 2/3; hyperkalernia	Glucocorticoid and/or mineralcorticoid
Hemoglobino pathies, including sickle cell anemia	1 in 15,000 1 in 400 African Americans	Death by sepsis (infection) or splenic sequestration in sickle cell disease, anemia	Penicillin & comprehensive care
Hyperphenylalaninemia including phenylketonuria (PKU)	1 in 11,000	Profund mental retardation; seizures	Low phenylaline diet
Biotinidase deficiency	1 in 60,000	Mental retardation, seizures, skin rash, alopecia (hair loss), hearing loss, death	Biotin
Galactosemia	1 in 60,000	Severe brain damage, liver disease, cataracts, death	Galactose-restricted diet
Maple Syrup Urine Disease	1 in 150,000	Neonatal coma, convulsions, mental retardation, acidosis and death	Diet low in branched chain amino acids
Selected Amino Acid disorders	1 in 11,000 – 1 in 250,000	Varying symptoms	Dietary therapy / Medicatior
Selected Organic Acid disorders	Varies	Varying symptoms	Dietary therapy / Medicatior
Selected Fatty Acid disorders	Varies	Varying symptoms	Dietary therapy / Medicatior

*** All of these tests are screening tests**. Abnormal results will be phoned to provider and will need full evaluation/discussion with a consultant before a diagnosis is confirmed or treatment is started.

The purpose of newborn screening is to identify infants at risk and in need of more definitive testing. As with any laboratory test, both false negative and false positive results are possible. Screening test results alone are insufficient information on which to base diagnosis or treatment.

Parent Refusal to Have Baby Tested

The parent may refuse testing for personal and/or religious beliefs. A signed Informed Dissent document must be kept in the infant's medical record if the parents refuse the screen.

Is the blood test safe for my baby?

Yes. Your baby's heel will be pricked. A few drops of blood are collected on a special filter paper. There is a small risk of infection when the skin is broken. This risk is low. Your provider will take precautions to keep your baby safe. Over a million newborns have been screened in Oregon since 1961, and, according to state records, not one newborn has had a problem from the heel prick.

What about using cord blood for the first test?

Cord blood could be used to detect certain abnormalities, like biotinidase deficiency or galactosemia. At least 90% of cases with congenital hypothyroidism can also be detected in cord blood. On the other hand, in conditions such as PKU, abnormalities will not be detected because they only appear in the infant's blood until after the birth. Cord blood is therefore NOT acceptable for PKU, Maple Syrup Urine Disease, or galactosemia.

Is this a one-time test? When is it done?

Ideally, to test for all the conditions, two tests must be done. The first test, done before 72 hours of age, is valid for most of the conditions listed. There is a small chance that aminoacidopathies like PKU may be missed. In Oregon, six percent of all infants with PKU and 10% of those with hypothyroidism are found only on the second test. The second test is done between 5 and 14 days of age, generally at the two week mark.

Parents do have the option of choosing only one test for their child. The most ideal time for a one-time test would be at day three. Parents do need to realize that by refusing the second test, there is a chance that their baby could be affected by a disease and they would not be aware of it. A signed Informed Dissent document must be kept on file in the infant's medical record if this is the option the parents choose.

I/We have read the information provided by our midwife on newborn metabolic screening in Oregon. We have decided to:

(please check one of the following)

[ ] Have the newborn screen performed twice in the neonatal period, at three days and two weeks after the birth

[ ] Have the newborn screen perfomed once in the neonatal period, at three days after the birth

[ ] Decline the newborn screening for my newborn baby

_____ _____

Mother's Signature Partner's Signature

_____ _____

Date Date

_____ _____

Midwife's Signature Date