Newborn Screening for Congenital Metabolic Disorders (Commonly referred to as Newborn Screening)

---Helps detect some newborn congenital metabolic disorders at an early phase and provide timely and proper treatment to prevent any physical compromise and/or mental retardation as a result of these disorders.

Dear Moms and Dads,

A healthy child is the source of merriment for a family. Any ailment or illness in the process of a child's growth often impacts the family and the society at various degrees. Hence, we are kindly reminding you that it is very important to let your children receive newborn screening and relevant physical checkups as soon as possible.

Newborn screening helps detect some congenital metabolic disorders without distinct symptoms at an early phase, provide proper diagnosis and treatment during the golden treatment period, and minimize the disease's damage on the body and/or mind.

The goal of newborn screening is to detect several congenital metabolic disorders early, preferably before the onset of symptoms, so timely treatment can be initiated. With treatment, these resultant physical and mental problems may be prevented.

To check if your baby suffers from these congenital metabolic disorders, a nurse or a professional at the hospital/clinic of birth will prick your baby's heel to collect several drops of blood after your baby is 48 hours old. The blood sample will be sent to a newborn screening center appointed by the Bureau of Health Promotion (BHP). Government offers subsidies for the newborn screening of the following disorders:

I. Congenital hypothyroidism:

This disorder occurs in about one in every 3,000 newborns. Babies with this disorder are not observed of any abnormality at birth. Symptoms surface within two to three months after birth. A major cause for the disorder is lack of thyroid hormone in the baby's body, which undermines the growth and development of the cranial nerves and physical parts. If the condition is not treated within the first six months after birth, most children will suffer from mental retardation, slow growth and development, and a short and small stature. But if found and treated within one to two months after birth, the babies can have normal intellectual and physical growth and development.

II. Phenylketonuria (PKU):

About one in every 35,000 births is born with Phenylketonuria (PKU) in Taiwan. The symptoms usually appear three to four months after birth, such as: slow growth and development, moldy and stinky urine and physical smell, and development of serious mental retardation. PKU occurs

mainly because babies cannot process the protein in their diet effectively. If the disorder is found early, and the babies are placed on a special diet and/or other treatments starting within 2-3months after birth, and are followed up periodically, most cases can be treated to ensure normal intellectual development of the babies.

III. Homocystinuria (HCU):
About one in every 100,000 to 200,000 babies is born with this disorder in Taiwan. Babies with this disorder cannot process the protein in their diet effectively. If left untreated, complications, including skeletal malformation, mental retardation, and thrombus formation will progress. If found at an early stage and treated with a special diet and supplementary vitamins, mental retardation can be prevented.

IV. Galactosemia (GAL):
About one in every one million babies is born with typical galactosemia in Taiwan. It occurs when babies cannot process galactose properly. Babies with this disorder tend to vomit and become lethargic after milk intake and can have eye, liver and/or brain damages. If found early and the babies are removed from breast milk or ordinary commercial formulas and put on lactose free formulas sever, damages can be prevented.

V. Glucose-6-phosphate dehydrogenase deficiency (G6PD deficiency, commonly known as Favism):
This is a common inherited disorder in Taiwan, found in about three in every 100 babies. It occurs when babies cannot process the glucose in red blood cells properly. Babies with this disorder suffers from neonatal jaundice and acute hemolytic anemia in response to ingestion of fava (broad) beans, or contact with naphthalene balls (moth balls), use of gentian violet, and intake of sulfa-drugs and antipyretic analgesics drugs. Without timely treatment, it may result in jaundice, mental retardation, and deaths. Identifying the babies' health conditions at an early stage and avoiding contact with the above-mentioned pathogenic factors can decrease the injuries to the babies.

Currently many medical facilities are introducing and using tandem mass spectrometers in their newborn screening, which can simultaneously screen multiple congenital metabolic disorders. To provide more complete and accurate newborn screening services, our government has since July 2006 added six items that need to work with the tandem mass spectrometer onto the existing screening for the foregoing five congenital metabolic disorders. The newly-added and subsidized newborn screening tests are as follows:

VI. Congenital adrenal hyperplasia (CAH):

About one in every 15,000 babies is born with this disorder. It is commonly caused by 21-hydroxylase enzyme deficiency. The clinical symptoms vary with the quantity and quality of the 21-hydroxylase enzyme deficiency including: (1) Salt-losing type: Most babies with this disorder experience life-threatening situations as a result of undiagnosed drastic loss in salt. (2) Simple virilizing type: Female babies with this disorder will have inappropriate sexuality, have no menses in the process of growth, be excessively malelike, be barren, and have abnormal growth and development. The male babies with this order will also experience developmental problems. If not found at an early stage, it will be really difficult to rectify both physiologically and mentally. (3) Late-onset type: Symptoms appear after infancy. Except for the late-onset type, babies can be diagnosed with congenital adrenal hyperplasia at an early stage through screening and be treated with appropriate supplements to avoid life at risk and ensure normal growth and development.

VII. Maple syrup urine disease (MSUD):

The incidence rate of this disorder is still unknown in Taiwan, but it occurs in one in every 120,000 European Caucasian babies and every 250,000 American babies. It is named such for the characteristic sweet odor of a patient's body fluid and urine that smells like maple syrup. Babies with this disorder typically have symptoms or signs of vomiting, drowsiness, loss of appetite, shortness of breath, and convulsions, that appear gradually after starting to feed for a few days. In extreme cases, babies will become unconscious and fall into a coma or even die. It is a rare disorder of abnormal metabolism of branched-chain amino acids (valine, leucine, and isoleucine). Early detection and treatment are essential for the babies to have relatively normal growth and intellectual development.

VIII. Medium chain acyl-CoA dehydrogenase deficiency (MCAD):

The incidence rate of this disorder is still unknown in Taiwan, but it occurs in about one in every 15,000 babies in Europe and America. It is one of the most common fatty acid oxidation deficiencies, whose clinical symptoms appear in the first two years after birth. The lack of the medium chain acyl-CoA dehydrogenase stops the fat metabolism from working properly and results in high levels of partially broken down fats, which are toxic to the brain and nervous system. Symptoms include vomiting, fatty liver, non-ketotic hypoglycemia, confusion, coma, and convulsions. Although some patients have no symptoms, twenty-five percent of the cases die at the first episode , and hence it is often mistaken as sudden infant death syndrome (SIDS). Early screening helps prevent attacks of this disorder. For acute cases, hypoglycemia should be treated immediately and quickly. Long-term treatment involves avoiding long periods of time without eating and having snacks that are high in carbohydrates before bedtime and treating

accidental situations such as infection or gastroenteritis aggressively. If we can prevent attacks of this disorder, the final outcome should still be very good.

IX. Glutaric acidemic type I (GAI):

The incidence rate of this disorder in Taiwan is still unknown. However, it occurs in about one in every 20,000 US babies. It is a rare disorder of abnormal amino acids metabolic deficiency. Due to lack of glutaryl CoA dehydrogenase enzyme, babies' body cannot process amino acid lysine and tryptophan properly, and excessive toxic products (such as glutaric acid) accumulate in the blood and tissue, resulting in progressive neural symptom(s) and acute metabolic abnormality. Babies may not show any abnormality or just have macrocephaly without symptoms in the first few months after birth. However, at the later stage of infancy, symptoms start to surface, including dyskinesias, progressive choreoathetosis, dystonia, muscle rigidity, paralysis, and opisthotonis (the limbs turn outwardly, and the body is like a bow). There might also be acute seizures such as epilepsy, lethargy, or coma. Early detection and treatment are essential to ensure relatively normal growth and intellectual development of the babies.

X. Isovaleric acidemia (IVA):

The incidence rate of this disorder in Taiwan is still unclear. However, about one in every 50,000 babies is born with isovaleric academia internationally. It is a rare disorder of organic acids metabolic deficiency. Lack of isovaleryl-CoA dehydrogenase enzyme makes babies unable to process leucine properly and results in excesses of the toxic product, isovalerate, which further invades the nervous system and the hematopoietic system. Bases on the seriousness of symptoms and the time point of the first episode, this disorder can be typical and non-typical. Babies of the typical disorder may look normal but symptoms like fatigue, nausea, vomiting, lethargy, lack of appetite and convulsions will gradually appear. Isovalerates accumulate in the body in large quantities and result in a foul "sweaty feet" odor on a baby's body and urine. If not diagnosed and treated properly, patients will grow into a coma. On the other hand, the non-typical disorder has a late attack time and less obvious symptoms. It is usually not diagnosed until a year after birth and hence is sometimes mistaken as one of other similar disorders. Early detection through newborn screening, dietary control and periodic follow-ups can bring rather good treatment results.

XI. Methylmalonic acidemia (MMA):

The incidence rate of this disorder in Taiwan is still unclear. It is a rare disorder of organic acids metabolic deficiency. Abnormal function of methylmalonyl CoA mutase enzyme or abnormal metabolism of cobalamine vitamin B12 leads to excessive accumulation of organic acids such as methylmalonic acid and propionic acid inside baby's body and causes damage to the nervous

system. In extreme cases, it may cause ketoacidosis, hypoglycemia, hypermmonemia, and hyperglycinemia. Newborns and infants with this disorder suffer from a high death rate. Early detection and screening can prevent acute attacks and enable timely treatment to avoid acidosis. For those responding to vitamin B12, vitamin B12 must be given in the treatment. For the others not responding to vitamin B12, special infant formulas and high calorie diets must be given so that the concentration of methylmalonic acid in the blood and urine can be maintained within an ideal range.

※ Before knowing the screening result, please do not let your baby contact naphthalene balls (commonly called moth balls), or take any drugs; if there is any jaundice condition (skin with yellow color) and/or other health problem, please be sure to consult with your pediatrician immediately. A (possibly) positive screening result does not mean that the baby is confirmed to have a particular disorder. The hospital that collected the blood sample or conducted verifying checkup will assist your baby in further confirmation and examination in the shortest time possible. The result of the initial screening or the follow-up screening test can be known in about weeks after blood sample is collected. For any questions, please contact your blood collecting hospital, or check the website of the newborn screening center of Taipei Institute of Pathology (http://www.tipn.org.tw/) ; for disorder-related questions, please dial the direct line for health education consultation at TEL: (02) 8596-2065.

Bureau of Health Promotion, Department of Health, R.O.C. (Taiwan) cares about you!