

INTRODUCTION

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Newborn Screening is an essential, preventive public health program for early identification of disorders that can lead to severe health problems. Screening of newborns for specified disorders began in the late 1950s and has since become widely accepted throughout the world as an important and effective public health activity. Newborn Screening Programs identify, in the newborn period, certain disorders which, if untreated, result in mental retardation, disabilities or even death. Early identification allows for early definitive diagnosis and treatment and potentially better outcomes.

As of July 1, 2008 the Kansas Newborn Screening Program in the Kansas Department of Health and Environment screens for twenty-nine disorders, including hearing. Statewide phenylketonuria (PKU) screening began in 1965. The screening program expanded in 1977 to include congenital hypothyroidism (CH), followed in 1984 by galactosemia (GALT). Hemoglobinopathies, such as sickle cell disease, became the fourth condition to be screened, starting in 1993. Hearing screening for newborns began in 1999. The expanded metabolic newborn screening follows the American College of Medical Genetics recommended core panel. A listing of the complete twenty nine tests can be found in Table 1. The disorders screened for by the Kansas Newborn Screening Program (NBS) are chosen because:

- the disorder occurs with significant frequency
- an inexpensive and reliable method of testing exists
- an effective treatment/intervention exists
- if untreated, the baby may die or develop severe mental retardation or disability
- the affected baby may appear normal at birth

The disorders screened for are each individually rare, so the chance that any single infant will be affected is relatively small. But the costs of not diagnosing these conditions, both in terms of human suffering and in financial terms, are substantial. Early diagnosis and treatment typically results in normal growth and development and/or can reduce morbidity and mortality. Most infants with one of these disorders appear normal at birth. It is only with time that the disorder affects the baby's health, growth, and development, but by then the damage may be permanent.

The goals of the Kansas Newborn Screening Program are to ensure that:

- each baby born in Kansas receives a newborn screening test for these disorders
- all infants with screening results outside of normal limits for the disorder receive prompt and appropriate confirmatory testing
- when appropriate, individuals diagnosed with a disorder receive early intervention to ensure better outcomes and are maintained on suitable medical and dietary therapy

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. It is important to remember that a screening test is not a definitive diagnosis and all abnormal results need to be confirmed and treated by a specialist for that disorder.

This manual describes the operation of the newborn screening program, the disorders currently detected by the program, standards for follow-up of out of range screening test results and recommended current, appropriate medical management of diagnosed cases.

Type of Disorder	Name of Disorder	Acronym for Disorder
Amino Acid Disorders	Phenylketonuria	PKU
	Maple Syrup Urine Disease	MSUD
	Homocystinuria	HCY
	Tyrosinemia, Type I	TYR I
	Arginosuccinic Acidemia	ASA
	Citrullinemia	CIT
Fatty Acid Disorders	Trifunctional Protein Deficiency	TFP
	Carnitine Uptake Defect	CUD
	Medium Chain acyl-CoA dehydrogenase Deficiency	MCADD
	Long Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency	LCHADD
	Very Long Chain Acyl-CoA Dehydrogenase Deficiency	VLCHADD
Organic Acid Disorder	Isovaleric Acidemia	IVA
	Glutaric Aciduria, Type 1	GA-I
	3-hydroxy-3-methylglutaryl CoA Lyase Deficiency	HMG
	Multiple Carboxylase Deficiency	MCD
	Methylmalonic Acidemia (Mutase)	MUT
	Methylmalonic Acidemia/Vitamin B12 Disorders	Cbl A & B
	3-methylcrotonyl-CoA carboxylase deficiency	3MCC
	Propionic Acidemia	PROP
	Beta Ketothiolase Deficiency	BKT
Hemoglobinopathies	Sickle Cell Anemia	SCA
	Beta Thalassemia	Hb S/Th
	Hemoglobin C disease	Hb S/C
Other Disorders	Hypothyroidism	HYPOTH
	Biotinidase	BIOT
	Congenital Adrenal Hyperplasia	CAH
	Galactosemia	GALT
	Cystic Fibrosis	CF
	Hearing	HEAR

Table 1

An efficient and effective newborn screening program requires coordinated efforts from a variety of health care providers:

- **PRACTITIONERS:** Prenatal, perinatal and newborn care providers are responsible for the appropriate collecting and handling of screening specimens, for providing families with accurate and current information about the tests and the disorders, and for assuring cooperation in obtaining prompt follow-up in the event of an out of range result.
- **SCREENING LABORATORY:** The Kansas Newborn Screening laboratory is responsible for performing appropriate tests on submitted specimens, for record keeping, for assuring quality control of laboratory procedures, and for notifying providers and the Kansas Newborn Screening Follow-Up Program of abnormal range results.
- **NEWBORN SCREENING FOLLOW UP:** The staff of the Newborn Screening Follow Up Program are responsible for notifying the physician of record of the out of range result, tracking, and follow-up of all children identified as having an out of range or unresolved result, administering an effective and efficient program, educating practitioners and the public, and for assuring adequate communication among all providers.
- **DIAGNOSIS AND TREATMENT:** The primary care physician is responsible for assuring prompt completion of confirmatory tests of infants with out of range screening results, and for making a referral to a medical specialist. Medical specialists are available through the Kansas Newborn Screening Program to complete confirmatory tests, to manage confirmed cases, to assist practitioners with management of confirmed cases, and to serve as a consulting resource for practitioners and the Newborn Screening Program. Once diagnosed, assistance may be available through Children and Youth with Special Healthcare Needs (CYSHCN) at the Kansas Department of Health and Environment.