

Tennessee Newborn Screening Program



Guide for
Practitioners

State of Tennessee Department of Health

Women's Health and Genetics

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health.state.tn.us/womenshealth/NBS

A program administered by Tennessee Department of Health with the assistance of Hospitals, Primary Care Providers, Pulmonologists, Endocrinologists, Genetic and Sickle Cell Centers from across the state.

I. Tennessee Law

Tennessee Code Annotated (TCA 68-5-401) Part 4 – Newborn Testing – Metabolic Defects

The general assembly hereby declares that as a matter of public policy of this state and in the interest of public health, every newborn infant shall be tested for phenylketonuria, hypothyroidism, galactosemia and other metabolic/genetic defects that would result in mental retardation or physical dysfunction as determined by the department, through rules and regulations duly promulgated.

II. Excerpts taken from Rules and Regulations

Chapter 1200-15-1-.01 Tests. The Department of Health will designate the prescribed effective screening tests and examinations which will be performed on the blood samples submitted in accordance with 1200-15-1-.02 for the detection of metabolic/genetic disorders in newborns. Tests are to be conducted for Biotinidase Deficiency, Congenital Adrenal Hyperplasia (CAH), Congenital Hypothyroidism, Galactosemia, Hemoglobinopathies, Homocystinuria, Maple Syrup Urine Disease (MSUD), Medium-Chain Acyl CoA Dehydrogenase (MCAD) Deficiency, Phenylketonuria (PKU), and other metabolic/genetic tests as designated by the Department of Health. Results of the Newborn Hearing Screening, if conducted, are to be submitted in conjunction with the blood sample procedure for the detection of disorder in accordance with 1200-15-1-.02.

- (1) Exemption for religious beliefs. Nothing in this part shall be construed to require the testing of or medical treatment for the minor child of any person who shall file with the department a signed, written statement that such tests or medical treatment conflict with such person's religious tenets and practices, affirmed under penalties of perjury pursuant to T.C.A. 68-5-403. The newborn screening refusal form provided by the State should be completed and retained in the medical record for the period of time defined by the hospital or provider policy.
- (2) Failure to have a child tested for the genetic/metabolic disorders is a Class C misdemeanor. Reporting of hearing screening is not to be construed as mandatory testing, therefore, failure to have a child tested for hearing loss will not be considered a misdemeanor pursuant to T.C.A. 68-5-404.

Chapter 1200-15-1-.02 Institutions Responsible For Test For Newborn Infants

- (1) The following persons or institutions shall be responsible for having tests made on newborn infants:
 - a) Every chief administrative officer of a hospital and the attending physician in each instance shall be responsible for submitting a specimen of blood to the State of Tennessee Laboratory, State Department of Health, in a manner as directed by the department. This sample shall be collected before newborn infants are discharged from the nursery, regardless of age.
 - b) Every chief administrative officer of a hospital and the attending physician shall direct every parent, guardian, or custodian to bring the infant, if the infant was initially screened before twenty-four (24) hours of age, back to the hospital or to a physician or the nearest local health department to be rescreened for Biotinidase Deficiency, Congenital Adrenal Hyperplasia (CAH), Congenital Hypothyroidism, Galactosemia, Hemoglobinopathies, Homocystinuria, Maple Syrup Urine Disease (MSUD), Medium-Chain Acyl CoA Dehydrogenase (MCAD) Deficiency, Phenylketonuria (PKU), and other metabolic/genetic tests as designated by the Department of Health within twenty-four to forty-eight (24-48) hours after birth. In the case of a premature infant, and infant on parenteral feeding or any newborn treated for an illness, who is not discharged from the nursery in a timely manner, the sample should be collected not later than the seventh (7) day of age.
 - c) Any healthcare provider(s) of delivery services in a non-hospital setting shall be responsible for submitting a specimen of blood to the State of Tennessee Laboratory, or directing every parent, guardian, or custodian to bring the infant, between twenty-four and forty-eight (24-48) hours of age, to a hospital, physician or local health department to be screened for

Biotinidase Deficiency, Congenital Adrenal Hyperplasia (CAH), Congenital Hypothyroidism, Galactosemia, Hemoglobinopathies, Homocystinuria, Maple Syrup Urine Disease (MSUD), Medium-Chain Acyl CoA Dehydrogenase (MCAD) Deficiency, Phenylketonuria (PKU), and other metabolic/genetic tests as designated by the Department of Health.

- d) Any parent, guardian, or custodian residing in Tennessee, of an infant born in Tennessee, outside a Tennessee health care facility and without assistance of a health care provider, shall between twenty-four to forty-eight (24-48) hours of the birth of said infant, present said infant to a physician or local health department for testing for the purpose of detecting Biotinidase Deficiency, Congenital Adrenal Hyperplasia (CAH), Congenital Hypothyroidism, Galactosemia, Hemoglobinopathies, Homocystinuria, Maple Syrup Urine Disease (MSUD), Medium-Chain Acyl CoA Dehydrogenase (MCAD) Deficiency, Phenylketonuria (PKU), and other metabolic/genetic tests as designated by the Department of Health.
- e) The original blood specimen shall be collected between twenty-four and forty-eight (24-48) hours of age. Repeat blood specimens shall be collected before two (2) weeks of age.
- f) Every chief administrative officer of a hospital that performs physiologic newborn hearing screening shall be responsible for reporting the results of the newborn hearing screening test performed prior to discharge from the health care facility. Results of the hearing screening are to be reported to the Department of Health on the form designated for newborn screening blood spot collection or a similar form designated by the Department.

III. Hospital Responsibility

A. Completion of the Newborn Screening Form

- ☞ Collection forms are available from the local Health Department.
- ☞ It is important to fill out all information on the Newborn Screening collection form completely and accurately. Some results are based on age, weight and/or feed status of the infant at the time of collection.
- ☞ Adoption Cases: If a specimen needs to be repeated, a letter will be sent using the information listed on the form. **Do Not** put birth mothers information on the form, list either adoptive parents, adoption agency or lawyer. Also write ADOPTION CASE on the collection form.
- ☞ Death of a Newborn: If a screen was collected and a newborn has died, notify the newborn screening follow-up program by fax. Include the child's name, birth date, mother's name and the date of death. Follow up will close the case so mother will not receive letters requesting a repeat specimen if needed. Fax # (615) 262-6458

B. Obtain a Satisfactory Specimen

- ☞ A satisfactory specimen is: Drops of whole blood applied evenly and allowed to soak through the filter paper and can be seen clearly with no white showing through on either side. Preferably these spots should be large enough to punch at least 6 - 1/8 inch discs with no white areas.
- ☞ Recommended techniques are available upon request. All Tennessee birthing hospitals/facilities and health departments were provided with a newborn screening video, "Let's Do it Right the First Time". The video demonstrates collection procedures and reviews the diseases screened for by the State of Tennessee.
- ☞ An unsatisfactory specimen of a newborn with one of the disorders can cause a possible delay in diagnosis and treatment. A specimen can be considered unsatisfactory for several reasons, including quantity insufficient, blood did not soak completely through filter paper, specimen was contaminated or arrived in a plastic bag. A complete list of descriptions for unsatisfactory specimens is available upon request.

- ☞ Reports are mailed to each birthing hospital on a quarterly basis. These reports alert you to the number of unsatisfactory specimens collected at your hospital. Monitor these reports and take the steps to decrease the number of unsatisfactory specimens submitted.

C. Transfused Newborns

- ☞ **Always collect a newborn screening before any transfusion** even if the infant is < 24 hours old, the **hemoglobin and biotinidase enzyme** results **will be accurate** and will not need to be repeated if the results are normal.
- ☞ **All tests except Hgb:** Collect a filter paper **4 days** after the last transfusion if baby did not have a normal newborn screen on lactose feed or if the 1st specimen was collected at < 24 hours of age.
- ☞ **Hgb (hemoglobin):** Will need to be collected **3 months** after the last transfusion if a filter paper was not collected prior to transfusion identifying a normal hemoglobin. This specimen will need to be collected in a microvette tube and sent to the Meharry Sickle Cell Center in Nashville for hemoglobin confirmation.
- ☞ If an infant has symptoms such as vomiting, diarrhea, dehydration and/or jaundice in which case the test should be repeated immediately and the Genetic Center should be contacted.

D. Parent Education and Pamphlets

Chapter 1200-15-1-.03 Metabolic/Genetic Newborn Screening, Pamphlet Provided To Parents.

The chief administrative officer of each hospital shall order the distribution of a pamphlet on Biotinidase Deficiency, Congenital Adrenal Hyperplasia (CAH), Congenital Hypothyroidism, Galactosemia, Hemoglobinopathies, Homocystinuria, Maple Syrup Urine Disease (MSUD), Medium-Chain Acyl CoA Dehydrogenase (MCAD) Deficiency, Phenylketonuria (PKU), and other metabolic/genetic tests as designated by the Department of Health, to every parent, guardian or custodian of an infant screened for these conditions. The pamphlet, distributed by the Department of Health, educates and prepares the family for newborn testing on their infant. If an infant's screen was collected earlier than twenty-four (24) hours after birth and the patient is discharged home, the health care facility must review the information on the back of the pamphlet with the family, which requires them to present the infant to the hospital, physician or health department within 24-48 hours for a repeat screen. The pamphlet will have a perforated page that may be signed by the parent and placed in the medical record as documentation that the pamphlet was provided.

Pamphlets are available upon request.

E. Quality Assurance

- ☞ Set up a system ensuring every infant born in your facility has a screen collected.
- ☞ Set up a system ensuring results are received on all NBS specimens your facility submits.
- ☞ Teach new personnel the proper methods of specimen collection and review with existing personnel on a regular basis.
- ☞ Review quarterly unsatisfactory specimen reports and take steps to lower the rate by identifying areas of weakness in your internal procedures.

IV. Newborn Screening State Laboratory and Follow-up Section Responsibilities

The Laboratory performs tests on all specimens, reports the results to both the provider and to the hospital of collection listed on the NBS form. Presumptive positives for diseases are immediately reported to the follow-up program. The follow up staff contacts providers and tertiary centers about the abnormal results, and follows up to ensure the patient has confirmatory testing, diagnosis and treatment when necessary. Follow up also informs the parent and provider by letter of the need for a repeat specimen due to either abnormal values, unsatisfactory specimen, transfusion, specimen collected at <24 hrs of age or possible hemoglobin trait.

V. Weekend and Holiday Calls

When the result of a specimen is abnormal, the lab repeats the test in duplicate. Anytime a critical result is identified on Friday and needs to be repeated, the lab personnel will come in on Saturday to complete the test. Results will be called to follow up, who will contact the provider listed on the newborn screening form and the on call personnel with the appropriate tertiary center. On any holidays greater than 3 days, lab personnel will come in to perform testing, specimens will not go longer than 3 days without being tested.

VI. Primary Care Provider Responsibilities for Follow-up

When the laboratory receives specimens, they are separated according to the age of the infant and the quality of the specimen then assigned a Tennessee Department of Health Number (TDH#) before tests are performed.

- A. Specimens Within Normal Limits (WNL) - Reports of normal specimens are mailed within 5-7 working days from receipt of specimen to provider and hospital of collection listed on the newborn screening form. No follow-up is needed, although providers are responsible for making sure their patient has had a newborn screen, reviewed and interpreted results with respect to blood transfusion and diet status. The provider is also responsible for informing parent/guardian of the results.
- B. Unsatisfactory Specimens - Medical technologists closely examine each specimen for quality and quantity before performing tests. There are several reasons a specimen might be marked unsatisfactory, some of the more common are: quantity not sufficient, the blood spots did not uniformly soak through the filter paper, the specimen was too old by the time it arrived in the state lab or the baby was <24 hours of age when collected. When a specimen is identified as unsatisfactory, the lab notifies the provider and hospital of collection by mail the next working day. Follow up staff also notifies the provider and parents by mail, and requests a repeat specimen to be obtained. It is the responsibility of the parents and provider once notified to obtain a repeat specimen. Specimens are mailed to the state laboratory; a second unsatisfactory specimen at this point can cause a costly delay in diagnosis and treatment. A description of unsatisfactory specimens is available upon request.
- C. Process for Presumptive Positive for Disease - The laboratory reports a presumptive positive result to follow up as soon as it has been determined, generally within 24-48 hours after the specimen is received. Follow up staff notifies the provider listed on the newborn screening form by telephone and fax to initiate confirmatory testing, follow-up, and treatment of the patient. Follow up also notifies the appropriate Endocrinologist, Genetic, Cystic Fibrosis or Sickle Cell Center. Results will be mailed to provider and hospital of collection when other tests are completed, within 5-7 days from receipt of specimen. Remember, this is a screening program and further testing will need to be performed prior to diagnosis and treatment.
- D. Unable to locate - When follow up and/or the physician are unable to contact or locate an infant for repeat testing due to unsatisfactory or abnormal results, the local health department should be contacted to assist.
- E. **Keep in mind; this is a SCREEN, not a diagnostic test.** The newborn screening (NBS) test can be affected by baby's age, medical or treatment status at the time of specimen collection; the quality and quantity of the specimen or other variables and may not detect all affected babies. The possibility of false negative or false positive results must always be considered when screening newborns for metabolic disorders. Regardless of NBS results, diagnostic evaluation should be performed on an infant presenting with clinical symptoms.

VII. Newborn Screening Video - "Let's Do it Right the First Time"

"Let's Do it Right the First Time" is a self instructional CD-ROM for practitioners. This course was designed for as a multidimensional tool which includes information about the disorders, techniques needed to properly collect blood specimens that are acceptable for the laboratory screening process, hearing screening information and the newborn screening follow up programs duties. For ordering information visit: health.state.tn.us/womenshealth/NBS

VIII. Newborn Screening Program Voice Response System (VRS)

The VRS is intended to provide quick access 24 hours a day 7 days a week to screening results via telephone and/or fax. The system provides step by step instructions to obtain Newborn Screening Information.

Local (Nashville): 262-3041

Toll Free: (866) 355-6132

When dialed, that number will connect you with a computerized voice response system which will access the newborn screening computer system. Using the keypad on a touch tone telephone, you will be able to request test results from the computer's database. In order to gain access to the system you must have:

- (1) Valid Tennessee Physician's License Number
- (2) Personal Identification Number (PIN)
- (3) Touch tone phone
- (4) Mother's Social Security Number
- (5) Fax machine, if you wish to receive faxed results

In order to gain access to the voice response system or receive more information about it contact the Newborn Screening Program at (615) 262-6153 or (615) 262-6304.

IX. List of Endocrinologist, Genetic and Sickle Cell Centers

A. Pediatric Endocrinologists

☛ T.C. Thompson Children's Hospital
Pediatric Endocrinology
Chattanooga (423) 778-6405

☛ Jackson Pediatric Center
Endocrinology
Jackson (731) 664-9928

☛ East Tennessee State University
Pediatric Endocrinology
Johnson City (423) 439-7320

☛ East Tennessee Children's Hospital
Department of Pediatric Endocrinology
Knoxville (865) 971-7400

☛ U.T Medical Group
Department of Pediatrics
Division of Endocrinology
Memphis (901) 287-5096

☛ Monroe Carell Jr.
Children's Hospital at Vanderbilt
Department of Pediatric Endocrinology
Nashville (615) 322-7427

☛ The Endocrine Clinic
Memphis (901) 763-3636

B. **Genetic/Metabolic Centers**

☞ University of Tennessee
Department of Medical Genetics
Knoxville (865) 305-9030
(800) 325-3894

☞ University of Tennessee
Division of Medical Genetics
Memphis (901) 448-6595

☞ Vanderbilt University Medical Center
Division of Medical Genetics
Nashville (615) 322-7601

C. **Satellite Genetic Centers**

☞ East Tennessee State University
Medical Genetics Center
Johnson City (423) 439-8541

☞ T. C. Thompson Children's Hospital
Division of Medical Genetics
Chattanooga (423) 778-6112

D. **Hematology Centers**

☞ T.C. Thompson Children's Hospital
Pediatric Hematology
Chattanooga (423) 778-7289

☞ University of Tennessee
Department of Medical Genetics
Knoxville (865) 305-9030
(800) 325-3894

☞ Meharry Sickle Cell Center
Meharry Medical College
Nashville (615) 327-6763

☞ St. Jude Children's Research Hospital
St. Jude Hematology
Memphis (901) 495-5670

E. **Cystic Fibrosis Centers**

☞ T.C. Thompson Children's Hospital
Pediatric Pulmonology
Chattanooga (423) 778-2001

☞ Vanderbilt University Medical Center
Division of Pediatric Pulmonary, Allergy,
and Immunology
Nashville (615) 343-7617

☞ University of Tennessee
at Le Bonheur Children's Medical Ctr.
Pulmonary Medicine
Memphis (901) 287-5222

☞ East Tennessee Children's Hospital
Pediatric Pulmonology
Knoxville (865) 637-8481
(865) 541-8698

X. Metabolic/Genetic Disorders

Disorder Incidence Genetics	Defect	Clinical Symptoms (untreated)	Screening Method	Goals of Screening	Pitfalls of Screening
Phenylketonuria (PKU) 1/14,000 AR Screening Began:1968 An amino acid disorder	Enzyme defect (phenylalanine hydroxylase);increased phenylalanine/phenylketones	Mental retardation Seizures	Elevated phenylalanine	Identify all infants with elevated phenylalanine levels. Assess for therapy before day 14 of life.	Inadequate oral/IV protein or amino acid intake
Congenital Hypothyroidism (CH) 1/3,000 Sporadic AR for hormone Dysgenesis Screening Began:1980	Insufficient production of thyroxine due to absent, dysfunctional or ectopic thyroid gland (Primary CH) or to defective TSH secretion by the pituitary (Secondary CH).	Most newborns show none. Jaundice, constipation, coarse facies/tongue, delayed skeletal maturity, posterior fontanelle, bradycardia, hypothermia	Elevated TSH When pituitary is normal indicates absent or hypofunctioning thyroid gland.	To identify all infants with primary CH and initiate therapy by day 14 of life.	Early samples inconclusive due to TSH surge at birth TSH screen only identifies Primary CH Some (VLBW) infants with CH display delayed TSH rise
Hemoglobinopathy 1/350 African Americans AR Screening Began:1988	Abnormal Hb (homozygous SS, doubly heterozygous SC, or heterozygous AS, AC)	Sickle cell disease associated sepsis, pain crises, pneumonia, anemia, gallstones, splenic enlargement etc.	Isoelectirc focusing, with HPLC or cellulose acetateelectrophoresis for confirmation	Identify infants with sickle cell disease for case management; identify infants with trait conditions for genetic counseling.	Abnormal hemoglobins must be confirmed. RBC transfusion affects results; screen before transfusion
Galactosemia 1/53,000 (classical) 1/6,000 (variants) AR Screening Began:1992	Enzyme defect (transferase) Elevation of galactose and galactose metabolites	<u>Classical:</u> Sudden death (E. coli sepsis); jaundice, hepatomegaly, acidemia, cataracts, mental retardation <u>Variants:</u> milder	Elevated total Galactose in blood RBC based assay (Enzyme screened in specified situation only; RBC based)	Identify all infants with classical galactosemia, prevent death, and begin diet immediately. Identify all treatable variant forms	Test not reliable on non-lactose/IV intake, or post RBC transfusion.
Congenital Adrenal Hyperplasia (CAH) 1/19,000 AR Screening Began:2000	Enzyme defect in cortisol and aldosterone synthesis. Leads to high ACTH, over secretion of adrenal androgens, and virilization of genitalia. Death from circulatory collapse and salt loss (salt wasting form).	Virilized female genitalia, males not apparent. Vomiting, circulatory collapse, hyponatremia, and hyperkalemia as early as 5 days of life.	Elevated 17 α -hydroxyprogesterone (17-OHP)	Identify all infants with the salt wasting form, 21 hydroxylase deficiency and treat within first week of life. Correct gender assignment in affected females.	Early sample inconclusive due to placental 17-OHP. Screening only identifies 21-Hydroxylase form (90% of cases).

Disorder Incidence Genetics	Defect	Clinical Symptoms (untreated)	Screening Method	Goals of Screening	Pitfalls of Screening
Biotinidase Deficiency 1/61,000 AR Screening Began:2003	Decrease in biotinidase activity which is needed to free biotin from protein which is required for carboxylases to function properly. When carboxylase is unable to perform their normal functions, altering fat, carbohydrate and protein metabolism, harmful byproducts collect in the body.	Hypotonia, seizures, coma, tachypnea, stridor, alopecia, conjunctivitis and dermatitis	Biotinidase Enzyme Activity	Identify all infants with deficient biotinidase activity	Severity of symptoms and age of onset can vary. False negative test results may occur with the use of sulfonamides, repeat filter paper 5 days after sulfonamides discontinued.
Maple Syrup Urine Disease (MSUD) 1/230,000 AR Screening Began:2004 An amino acid disorder	Enzyme defect or deficiency which is needed for metabolism of leucine, isoleucine and valine, amino acids. Life threatening complications may occur due to accumulation of derivatives of above amino acid.	Maple syrup odor of urine/sweat. Poor feeding, high pitched cry, vomiting, mental retardation hypertonia or hypotonia, convulsions and/or coma.	Elevated Leucine and/or Valine Levels	Identify all infants with elevated leucine levels. Assess need for diet and cofactor therapy and begin immediately.	Inadequate oral/IV protein/amino acid intake can delay rise in leucine levels. Infant receiving hyperalimentation may have mildly elevated leucine levels.
Medium Chain Acyl-CoA Dehydrogenase (MCAD) 1/12,000 AR Screening Began:2004 A fatty acid disorder	Deficiency of the enzyme MCAD, which is necessary for the breakdown of certain fatty acids leading to the accumulation of Acyl-CoA derivatives of fats in the liver and the brain.	Triggered during periods of fasting. Hypoglycemic, lethargy, vomiting and/or liver mal-function. Viral illnesses that limit food intake may cause symptoms to occur.	Elevated C8 levels	Identify all infants with elevated Octanoylcarnitine levels. Assess need for therapy with low fat diet and carnitine; begin treatment immediately.	Confirmation may require analysis of urine organic acid, Acyl carnitine profile and DNA studies.
Homocystinuria 1/340,000 AR Screening Began:2004 An amino acid disorder	Reduced activity of cystathionine beta synthase, which is required for the conversion of homocysteine to cystathionine and cysteine, needed for proper growth and development.	Mental retardation, seizures, thrombosis and dislocated lens	Elevated Methionine levels	Identify all infants with elevated methionine levels. Assess and initiate diet and cofactor treatment immediately.	Inadequate oral/IV protein/amino acid intake can cause methionine to rise slowly.
Amino Acid Disorders AR Screening Began:2004	Defect in amino acid metabolism caused by a specific defect in the biosynthesis of one of the enzymes.	Hypotonia, hypothermia, poor feeding, persistent vomiting, developmental delays, damaged to vital organs, seizures or coma. The effect of the disorder will depend on the age at which symptoms occur.	Elevated metabolites using MS/MS related to specific disorder	Identify all infants with elevated metabolite levels. Assess need for diet and cofactor therapy and begin immediately.	Inadequate oral/IV protein/amino acid intake can delay rise in amino acid levels. Infant receiving hyperalimentation may have mildly elevated levels.

Disorder Incidence Genetics	Defect	Clinical Symptoms (untreated)	Screening Method	Goals of Screening	Pitfalls of Screening
Organic Acid Disorders AR Screening Began:2004	Defect in protein metabolism where an essential enzyme is absent or malfunctioning causing accumulation of organic acids in blood and urine.	Vomiting, metabolic acidosis, ketosis, hyperammonemia, lactic acidosis, hypoglycemia, failure to thrive, hypotonia, global developmental delay, sepsis, hematological disorders and ultimately death. The effect of the disorder will depend on the age at which symptoms occur.	Elevated metabolites using MS/MS related to specific disorder	Identify all infants with elevated metabolite levels. Assess need for diet and cofactor therapy and begin immediately.	Confirmation may require analysis of urine organic acids, blood Acyl carnitine profiles and enzymatic studies
Fatty Acid Oxidation Disorders AR Screening Began:2004	Accumulation of fatty acids and a decrease in cell energy metabolism due to an enzyme defect in the fatty acid metabolic pathway (use of dietary and stored fat).	During the first crisis children have presented with metabolic acidosis, persistent vomiting, hypoglycemia, lethargy, apnea, encephalopathy, coma, cardiopulmonary arrest, or sudden unexplained death	Elevated metabolites using MS/MS related to specific disorder	Identify all infants with elevated metabolite levels. Assess need for diet and cofactor therapy and begin immediately	Confirmation may require analysis of urine organic acid, blood Acylcarnitine profile and DNA studies
Cystic Fibrosis AR Screening Began:2008 1/3,000 Caucasian 1/6,000 Hispanic 1/10,000 African-American 1/90,000 Asian-American	Defect in cystic fibrosis transmembrane conductance regulator (CFTR) gene. CFTR regulates the movement of salt (sodium chloride) and water into and out of cells, when it does not work properly high levels of salt are found in the sweat. This also results in thick, sticky secretions in the respiratory and digestive tracts, as well as in the reproductive system.	Failure to thrive, bulky and greasy stools (steatorrhea), failure of newborn to pass stool, frequent sinopulmonary infections, nasal polyps, clubbing, excessive salt in sweat, dehydration	Elevated immunoreactive trypsinogen (IRT), a pancreatic enzyme elevated in most CF affected newborns	Identify all infants with elevated IRT so CF can be diagnosed early and patient referred to an accredited CF center for assessment and management. Aggressive nutritional support and respiratory care can be started before the infant gets sick.	IRT may be elevated in many newborns and slowly decrease to normal with age increasing the number of false positives identified. Delays in specimen transport or if exposed to heat may cause false negative results. Diagnostic evaluation should be performed on all infants with meconium ileus (MI) regardless of NBS results because they may have low initial IRT values.



Tennessee Department of Health
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