State of Illinois
Illinois Department of Public Health

Newborn Screening Practitioner's Manual

January 2011

Illinois Department of PUBLIC HEALTH

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Introduction

Newborn screening is a public health activity aimed at early identification of newborns affected with certain genetic and/or metabolic conditions. Early diagnosis and treatment of these conditions has been shown in many cases to reduce morbidity, premature death, mental retardation and other developmental disabilities. Newborn screening is recognized as one of the most successful recent public health accomplishments, and was the first population-based genetic screening program to become an integral component of public health practice.

While newborn screening disorders are individually rare, collectively the incidence of these disorders is around one in 500 births. In Illinois, each year around 350-400 babies are diagnosed with a condition identified by newborn screening. Newborn screening affects all health care practitioners who work with children and their families. During their practice of medicine, most pediatric and family medicine health professionals will receive a notice that a child in their care has a serious abnormal newborn screen, or has been diagnosed with a newborn screening disorder.

Health care practitioners are vital links within an effective newborn screening system, a system that includes hospitals, the state newborn screening laboratory and follow-up program, parents, health care providers, medical specialists and community service agencies. Health care providers serve as the front line in assuring that all newborns receive adequate screenings and when necessary, appropriate follow-up services within a medical home.

The Illinois Department of Public Health (Department) Newborn Screening Program developed this manual for health care professionals as a reference guide to newborn screening in Illinois. This resource provides protocols for specimen collection, laboratory testing, follow-up services, and the Department's reporting of both normal and abnormal screening results. Information about the disorders included in the current newborn screening test panel also is provided. This manual is intended to provide background information and general guidance on issues related to newborn screening, but does not replace the case specific medical advice available through consult with pediatric medical specialists, including those who may be contacted at the medical centers listed in the manual Appendix.

Overview

The Newborn Metabolic Screening Act (410 ILCS 240/) mandates newborn screening for all infants born in Illinois. This act authorizes the Illinois Department of Public Health to promulgate administrative rules for newborn screening (Title 77: Public Health, Chapter 1: Department of Public Health; Subchapter i: Maternal Child Health; Part 661 Newborn Metabolic Screening and Treatment Code). The Newborn Metabolic Screening Act and the newborn screening administrative rules may be viewed at the Department's Web site, www.idph.state.il.us.

All Illinois newborn infants are mandated to have a blood sample collected on the special filter paper specimen cards supplied by the Illinois Department of Public Health. The only valid exception is parental refusal based on religious beliefs and practices; in which case, a written refusal must be signed by the parents and documented in the infant's medical record.

Newborn screening blood spot specimens should be collected as soon as possible after the first 24 hours of life. If the baby is to be discharged from the birth center prior to 24 hours of age, the specimen should be collected before discharge. After drying, the specimen cards should be sent by courier to the Department's Newborn Screening Laboratory in Chicago for testing. When testing is completed, a report of all test results is issued by the Department's Newborn Screening Laboratory to the specimen submitter (usually the birth center). Birth centers are expected place the original screening report in the infant's medical record and to relay a copy of the results to the baby's primary care provider.

In addition to this laboratory report, abnormal, unsatisfactory and invalid test results are reported by the Newborn Screening Follow-up Program to the physician of record, the physician whose name appears on the specimen card. In some cases, hospitals may authorize reporting of results to a specified hospital contact person. The physician of record or the birthing hospital newborn screening contact person is expected to inform the mother, and should the baby have a new primary care provider, the new physician, of the abnormal test results and to facilitate any recommended follow-up activities. Necessary follow-up may include evaluation of the baby's medical condition and collection of a repeat newborn screening specimen or referral to a pediatric medical specialist for diagnostic testing. If the mother cannot be contacted, the assistance of the birth hospital and/or the local public health department may be needed to help locate the family. The Department's Newborn Screening Program should be informed of any difficulties in locating the family. Every effort should be made to assure that each baby who has an abnormal newborn screen receives the appropriate follow-up services in a timely manner.

In the cases of babies with any abnormal results requiring immediate referral to a pediatric medical specialist, a list of Department designated specialists, information about the suspected disorder and the actual test results will be provided to the physician of record or the hospital newborn screening contact. The American College of Medical Geneticists (ACMG), www.acmg.net, provides detailed action plans for follow-up of suspected newborn screening disorders. The University of Illinois at Chicago, Division of Specialized Care for Children (DSCC), www.uic.edu/hsc/dscc, provides additional information about these conditions and the importance of medical homes for children with special health care needs.

In addition to these resources, the Federal Maternal and Child Health Bureau of the Health Resources and Services Administration provided grant funding to create the Region 4 Genetics Collaborative, which includes Illinois, Indiana, Kentucky, Michigan, Minnesota, Ohio and Wisconsin. The goals of this multi-state collaborative include efforts to address inequities in genetics resources and to improve the quality of genetics services, including newborn screening throughout the region. The Region 4 Genetics Collaborative Web site, www.region4genetics.org, provides information about newborn screening, medical homes, specialty care resources and genetic counseling services available within this seven state region.

Illinois Newborn Screening Disorder List

Although additional disorders may be added as determined by the Illinois Department of Public Health under the advisement of the Genetic and Metabolic Disease Advisory Committee, the Illinois newborn screening panel currently includes the following disorders:

Endocrine Disorders
- Congenital adrenal hyperplasia (CAH)
- Congenital hypothyroidism

Hemoglobinopathies
- Sickle cell disease, trait conditions and other hemoglobinopathies
- Alpha thalassemia
- Beta thalassemia major

Amino Acid Disorders
- Homocystinuria (HCU)/Hypermethioninemia
- Maple syrup urine disease (MSUD)
- Phenylketonuria (PKU)/Hyperphenylalaninemia
- Tyrosinemia - tyrosine levels may not be sufficiently elevated for detection in first few days of life
- 5-Oxoprolinuria (5OXP) - may not be reliably detected in first few days of life

Urea Cycle Disorders
- Argininemia - extremely rare
- Argininosuccinic aciduria (argininosuccinate lyase deficiency - ASAL)
- Citrullinemia (argininosuccinate synthetase deficiency - ASAS)

Organic Acid Disorders
- 2-methylbutyryl-CoA dehydrogenase deficiency (2MBCD)
- 3-methylcrotonyl-CoA carboxylase deficiency (3MCC)
- 3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG)
- 3-methylglutaconic aciduria (3MGA)
- Beta-ketothiolase deficiency (BKT)
- Glutaric aciduria, type 1 (GA1)
- Isovaleric acidemia (IVA)
- Malonic aciduria (MA) - may not be reliably detected in the first few days of life
- Methylmalonic acidemia (MMA)
- Multiple carboxylase deficiency (MCD)
- Propionic acidemia (PA)

Fatty Acid Oxidation Disorders
- Carnitine/acylcarnitine translocase deficiency (CACT) - neonatal form is extremely rare
- Carnitine palmitoyl transferase deficiency, type 2 (CPT2) - neonatal form is extremely rare
- Carnitine palmitoyl transferase deficiency, type 1A (CPT1A) - may not be reliably detected in first days of life
- Carnitine uptake defect (CUD) - may not be reliably detected in first few days of life
- Glutaric aciduria, type 2 (GA2)/Multiple acyl-CoA dehydrogenase deficiency (MADD)
- Isobutyryl-CoA dehydrogenase deficiency (IBCD)
- Medium chain acyl-CoA dehydrogenase deficiency (MCADD)
- Medium/Short chain L-3-hydroxyacyl-CoA dehydrogenase deficiency (M/SCHADD)
- Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD)
- Short chain acyl-CoA dehydrogenase deficiency (SCADD)
- Trifunctional protein deficiency (TFP)
- Very long chain acyl-CoA dehydrogenase deficiency (VLCADD)

Other Disorders
- Biotinidase deficiency
- Galactosemia (Classical)
- Cystic fibrosis (CF)
- Lysosomal storage diseases
  - Pompe
  - Fabry
  - Gaucher
  - Krabbe
  - Niemann-Pick

Important Contact Information

Newborn Screening Follow-up Program
Illinois Department of Public Health
Genetics/Newborn Screening Program
535 W. Jefferson St., Second Floor
Springfield, IL 62761
Phone 217-785-8101
FAX 217-557-5396

Newborn Screening Laboratory
Illinois Department of Public Health
Division of Laboratories
2121 W. Taylor St.*
Chicago, IL 60612
Phone 312-793-4752
FAX 312-793-1054

Accounting Services
Illinois Department of Public Health
Account Services Billing Manager
Phone 217-782-5934

Web Sites
Illinois Department of Public Health Genetics/Newborn Screening Program -
www.idph.state.il.us/HealthWellness/genetics.htm

Illinois Department of Public Health Newborn Screening Laboratory -
www.idph.state.il.us/a-zlist.htm#N

***Note: Newborn Screening Shipping Labels**
Special courier service shipping labels are available to **birthing hospitals**. Please contact Newborn Screening Follow-up Program for more information about this service (217-785-8101).

All other submitters of newborn screening specimens are highly encouraged to utilize a courier service for prompt delivery of these dried blood samples, and shipping labels should be addressed to:

Newborn Screening Laboratory
Illinois Department of Public Health
2121 W. Taylor St.
Chicago, IL 60612

Practitioner's Newborn Screening Responsibilities

Specimen Collection

- Attending physician at birth or in the immediate newborn period has primary responsibility for collection of a specimen for newborn screening. The physician's responsibility may be delegated to the hospital administrator or the administrator's designee.

- If the birth is attended by a licensed nurse midwife, the midwife has primary responsibility for collection of a specimen for newborn screening.

- Parents should be informed that a blood specimen will be collected from their infant and printed information about newborn screening and how parents can access screening results should be provided.
    - *Newborn Screening Guide for Parents: Baby's First Steps in Life* document is available through the Department's Genetics/Newborn Screening Program. An electronic file of this document may be downloaded from the Department's Web site www.idph.state.il.us, and reprinted for distribution.
    - Documentation that a newborn screening specimen was collected and a copy of the screening results should be placed in the infant's medical record.
    - Parents should be informed that accurate contact information (emergency contact, current address and valid phone number) is vital, should their baby's newborn screening test be abnormal and additional testing or referral of the infant to a specialist become necessary.

- Physician or health care provider caring for the infant during the first month of life is responsible for newborn screening if -
    - Birth occurs outside of a hospital or medical facility.
    - Birth occurs without a physician or licensed midwife in attendance.

The American Academy of Pediatrics, August 2000 supplement to *Pediatrics*, "Serving the Family from Birth to the Medical Home," suggests that the role of the medical home health care professional include establishment of office protocol to retrieve the results of newborn screening for all newborns admitted to the practice when scheduling the first appointment. When screening results cannot be documented, a newborn screening specimen should be collected from the baby and submitted for testing.

- The Illinois Department of Public Health encourages primary care practitioners to provide medical homes, and to facilitate follow-up services for infants with abnormal newborn screening results.
    - The Federal Maternal and Child Health Bureau of the Health Resources and Services Administration (HRSA) funded Region 4 Genetics Collaborative Web site www.region4genetics.org provides information about newborn screening, medical homes, pediatric specialty care resources and genetic counseling services available within this seven state region.

- Primary care providers have an obligation to verify newborn screening results, and should not assume lack of notification indicates the baby's screen was normal.
    - Reports may be sent to the wrong health care provider.
    - Specimens may be lost in transit to the Department laboratory.
    - On rare occasions, hospitals may fail to collect a newborn screening specimen prior to hospital discharge or transfer.

- If there is no physician caring for the infant, the parents are responsible for obtaining newborn screening for their baby.

Newborn Screening Fee

- A fee will be charged for each specimen submitted to the Department Newborn Screening Laboratory for testing. This fee provides funding for testing, follow-up services for babies with abnormal results, and provision of certain medically necessary dietary treatment formulas.

- The Illinois Department of Public Health bills hospitals and health care agencies on a monthly basis for all newborn screening specimens submitted during that month.

Repeat Specimens, Diagnostic Testing and Referrals

- Physician of record or hospital designee is responsible for informing parents and/or infant's primary care/medical home provider of abnormal or unsatisfactory test results. If repeat screening is necessary, the physician of record is responsible for obtaining and submitting a repeat specimen and/or informing the infant's primary care provider of the need for additional testing.

- If referral of the infant to a pediatric medical specialist is necessary, the physician of record or hospital designee is responsible for assisting the baby's family and facilitating the referral. The Department highly recommends that the physician's office contact the medical specialist and provide the screening results to assure the referral is completed, and the screening results are accurately reported. Physicians and hospital staff should not refer to the newborn metabolic screen as the "PKU test." Use of this outdated, inappropriate terminology has often resulted in confusion about newborn screening results among primary care providers, medical specialists and parents.

Refusal of Newborn Screening

- Parents may refuse newborn screening only on the basis of religious beliefs and practices.

- If parents refuse newborn screening of their infant, parent education about the seriousness of newborn screening disorders should be provided, and the infant's primary care provider should be informed about the refusal. A written objection statement should be signed by the parents and placed in the infant's medical record, and a copy of the statement may be sent to Illinois Department of Public Health, Genetics/Newborn Screening Program.

Infants Born Outside State of Mother's Residence

- Illinois residents whose infants are born in other states may obtain newborn screening through the Department's Newborn Screening Laboratory. The physician should obtain the Department's filter paper specimen forms or order the screening through an Illinois birthing hospital. If an initial screening was performed in another state, a second newborn screening specimen may be submitted to the Department. Specimens should be collected and submitted as soon as possible after 24 hours of age. Newborn screening of infants older than 6 months of age is not recommended as the Department's normal values are based on normal analytes, distributions and controls for newborns. In these cases, a consultation with a pediatric sub-specialist is recommended.

- Infants born in Illinois whose mothers reside in another state must have a specimen sent to the Department's Newborn Screening Laboratory. Illinois provides screening for 29 of the disorders currently recommended by the American College of Medical Geneticists and the U.S. Centers for Disease Control and Prevention, plus several additional disorders, but parents who choose to are free to obtain a second screening within their state of residence.

Collection of Newborn Screening Specimens

Filter Paper Collection Form

- Only Department filter paper specimen collection forms are accepted by the Illinois Department of Public Health Newborn Screening Laboratory. The U.S. Food and Drug Administration now requires printed expiration dates on specimen cards, and health care providers are advised to check the card prior to specimen collection. See Appendix to view a copy of the Department collection form.

- Newborn screening filter paper specimen collection forms may be requested by contacting the Department's Division of Laboratories Springfield office, -217-524-6222 by phone, or may fax a request with appropriate contact information to 217-524-7924.

- Filter paper specimen cards should be stored in a cool, dry location out of direct sunlight. Cards should be stored in their original wrappings and stacked in a manner that avoids compressing the paper. When properly stored specimen cards have a shelf life of around two years. However, each card has a printed expiration date and once expired, the specimen cannot be considered valid. When expired cards are received, although testing is performed, the Department must issue a report requesting submission of a new sample for a valid screening.

- Birth history and identifying information requested on specimen collection forms should be complete, legible and accurate.

- Accurate personal health information is crucial to valid and reliable testing.

- Accurate personal contact information is vital to the physician for identifying the infant and contacting the parents, should abnormalities be detected in the blood sample.

Timing of Specimen Collection

- Newborn screening specimen collection from healthy newborns should occur as soon as possible after 24 hours of age. All newborns should have a sample collected within the first 24-48 hours of life. Specimens should <u>not</u> be collected prior to 24 hours of age with the exception of special circumstances.

- Special circumstances include:
    - Early discharge: If the infant is to be discharged at less than 24 hours of age, collect specimen prior to discharge. Inform parents that infant must be rescreened during the second day of life.
    - Transfers: If the infant requires transfer to another facility, if at all possible, a specimen should be collected prior to transfer, regardless of infant's age. If a specimen cannot be collected prior to transfer due to the medical instability of the neonate, the transferring facility is responsible for informing the admitting facility of the need for specimen collection prior to transfusion and/or within first 24-48 hours of life.
    - Transfusions: If the infant requires transfusion, specimen should always be collected prior to transfusion, regardless of infant's age.
    - Premature and sick infants: See Special Considerations section below.

- If a specimen is collected prior to 24 hours of age, **repeat specimen collection is necessary as soon as possible during the second day of life**.

Special Considerations

- Infants born outside of hospital/medical facilities: These infants should have a specimen collected at 24-48 hours of age.

- Premature and sick infants: If the infant's condition is medically unstable, the specimen should be collected at 24-48 hours of age.
    - All infants admitted to a neonatal intensive care unit (NICU) or special care nursery should have a routine second specimen collected at 14 days of age or prior to discharge from the unit, whichever comes first.
    - The "NICU" check box on the specimen card should be marked on specimens from all infants admitted to a NICU or special care nursery. Retest box should be marked for all repeat specimens.

- Infants receiving special feedings: Infants requiring soy formula, hyperalimentation or total parenteral nutrition (TPN), and those not yet receiving milk (galactose) feedings at the time of specimen collection require documentation of feeding type or status on the specimen card.
  - The feeding type box should be clearly marked for "breast," "soy," "other," "TPN" or "NPO" (nothing by mouth). This information is important to newborn screening laboratory staff and the baby's physician should an abnormality be detected.
  - Soy formula or lack of milk feeding **will affect** screening for galactosemia.
  - Hyperalimentation and TPN **may affect** tandem mass spectrometry screening for some amino acid, fatty acid oxidation and organic acid disorders.
    - If screening results suggest TPN effects, another specimen is requested when the infant has been off TPN for 48 hours, or on day 14 of life if the baby was admitted to NICU or special care nursery.

- Infants receiving antibiotics: When infants are receiving antibiotics at the time of specimen collection, the "antibiotic" box of the specimen collection card should be marked, as the presence of antibiotics and some other medication metabolites (valproic and benzoic acids) may be detected by tandem mass spectrometry. In these cases, a repeat sample will be requested.

- Transfusions: Collect initial specimens prior to transfusion, if at all possible. If this is not possible and the infant was transfused prior to specimen collection, indicate the **last transfusion date prior to the specimen collection** on the filter paper collection form.
  - Transfusions **will invalidate** screening for classical galactosemia.
  - Transfusions **will invalidate** screening for biotinidase deficiency.
  - Transfusions **will invalidate** screening for hemoglobinopathies.
  - Specimen collection immediately after transfusion **will affect** all newborn screening results. If a post-transfusion specimen is necessary, collection should be delayed for 48 hours post-transfusion.
  - If the infant's **initial specimen** was collected post-transfusion, a second specimen should be collected 48 hours post-transfusion, and a third specimen is required 90 days after the final transfusion.

Tips for specimen collection

- Electronic copies of the newborn screening specimen collection posters "Neonatal Screening: Blood Specimen Collection and Handling Procedure" and "Simple Spot Check" are available for download and printing through the Genetics/Newborn Screening Program, or for purchase through the Clinical Laboratory Standards Institute (CLSI). These posters provide an excellent training resource.

- The Genetics/Newborn Screening Program has provided to each Perinatal Network a copy of the Clinical Laboratory Standards Institute (CLSI) document, "Blood Collection on Filter Paper for Neonatal Screening Programs; Approved Standard"(LA4-A4) which is available on loan to birth centers.

- Heel stick is the preferred mode of collection for newborn screening whenever possible.

- If heel stick is not possible, use of a syringe to collect blood from an umbilical catheter is recommended by the CLSI.
  - If heel stick is not a viable option, see CLSI recommended procedures for specimen collection, Approved Standard LA4-A4.
  - Some screening results may vary slightly between heel stick specimens and venous or capillary specimens.

- Collection of specimens in capillary tubes is not recommended by the CLSI.
  - **Heparinized capillary tubes** should only be used when heel stick or collection by syringe is not possible.
  - Use extreme caution when applying a blood sample to the filter paper via capillary tubes. Caution is needed to prevent damage to filter paper and avoid "layering." Uniform application of the blood sample is required.
  - Layering can affect the validity and reliability of screening results. If heel stick is not an option, see the CLSI recommended procedures for specimen collection.

- **EDTA anticoagulant should never be used** for newborn screening specimen collection and will invalidate screening results and may cause false negative or false positive results.

Collection of Repeat specimens

- Repeat screening is requested by the Department when results of the original specimen were borderline abnormal, the specimen was declared unsatisfactory for testing, or declared invalid due to delayed submission or incomplete information on the specimen card.

- Routine repeat screening also is required for all infants admitted to the NICU or special care nursery. See Section "Special Circumstances – Premature and Sick Infants." Submitters should mark the retest box on the specimen card.

- Submitters should provide adequate information to allow matching of any retest specimens to the infant's original newborn screening record. All known names of the infant (beginning with the birth name), the mother's full name, date of birth and the infant's medical record number will greatly assist Newborn Screening Program staff in matching specimens.

Handling and Submission of Newborn Screening Specimens

Submitting Specimens

- It is highly recommended that completed newborn screening specimen collection forms be air dried for a minimum of three hours and submitted to the Department laboratory for testing **within 24 hours of collection** using a courier service.

- For details about the latest recommendations for specimen submission contact:

  Illinois Department of Public Health
  Genetics/Newborn Screening Program
  535 W. Jefferson St., Second Floor
  Springfield, IL 62761

- Currently, all Illinois **birthing hospitals** may utilize the Department supported courier service for pickup of newborn screening specimens and shipment to the Chicago Newborn Screening Laboratory. Contact the Newborn Screening program 217-785-8101 for more information about this service.

Timeliness

- Specimens should be submitted to Department Division of Laboratories on a **daily basis**, during regular business days Monday through Friday. Saturday UPS courier service pickup of specimens for **next business day delivery** also is available to birthing hospitals, but must be requested 24 hours in advance and arranged with the courier service. For more information contact the Newborn Screening Program 217-785-8101.

- Newborn screening disorders are serious and can be life threatening, therefore early detection and treatment is vital. Failure to submit specimens promptly may unnecessarily delay detection and treatment of affected infants. Batching of specimens from multiple collection days is **unacceptable**.

- Tracking courier service delivery of specimens to the Department's Newborn Screening Laboratory is the responsibility of birthing hospitals. Please contact the Newborn Screening Program at 217-785-8101 for more information about tracking deliveries and reporting courier service problems.

- Reports on the timeliness of specimen delivery for each hospital submitting specimens to the Department are available through the Newborn Screening Program.

Reporting of Screening Results

Normal Results

- Reported by written laboratory mailer sent to the submitting facility or submitting agent when testing is completed.

Abnormal Results

- Newborn screening is not diagnostic. Abnormal screening results are designated by Department laboratory staff as "presumptive positive" abnormal or "suspect borderline" abnormal.

- **Presumptive positive abnormal**: Indicates with high probability that the infant may have a disorder. Newborn Screening Program staff will recommend that infants with presumptive positive abnormal screenings be referred to medical specialists for consultation and/or diagnostic testing.
    - These results will be reported by phone call to submitting physician or submitting facility contact person, followed by a letter reporting the abnormal results and recommendations (sent by fax and mail).
    - A complete laboratory report (mailer) of all results is also sent to the submitting facility or submitting agent by Department Newborn Screening Laboratory for every specimen received.

- **Suspect borderline abnormal**: Indicates that the screening was slightly abnormal and that the infant needs a medical evaluation and a repeat newborn screen. If the infant has any symptoms of a disorder, referral to a medical specialist for diagnostic testing is indicated.
    - These results will be reported by letter indicating abnormal results and recommendations.
    - The letter reporting results and recommending additional follow-up screening or referral is sent by mail to the submitting physician or facility.
    - A complete laboratory report (mailer) of all results also is sent to the submitting facility or submitting agent for each specimen received.

Unsatisfactory Specimens

- Unsatisfactory specimen reports indicate the specimen was improperly collected, handled or submitted, as determined by the Department's Division of Laboratories. Specimens must be of good quality to assure reliable, valid newborn screening; unsatisfactory specimens require collection and submission of a new sample to assure that every baby receives a valid newborn screening.
    - These results are reported by letter indicating the unsatisfactory nature of the specimen and the need for immediate repeat specimen collection.
    - The letter is sent by mail to the submitting physician or facility.
    - Unsatisfactory specimen results also are included in the Department's laboratory report sent to the submitting facility or submitting agent.

- Reports on the number and type of unsatisfactory specimens are made available to hospitals by the Newborn Screening Program.

Referrals to Pediatric Medical Specialists and Other Agencies

Pediatric Medical Specialists

- Lists of Department designated pediatric medical specialists are provided by the Newborn Screening Program with letters reporting all presumptive positive abnormal results (those indicating need for referral to a medical specialist). See Appendix for a listing of medical centers providing pediatric sub-specialty services. These medical specialist lists are subject to periodic revision, and updated lists may be requested by calling 217-785-8101.

- Infants with family history of a disorder or those who appear symptomatic, require consult with a pediatric medical specialist. These infants should be referred for medical evaluation and possible diagnostic testing, regardless of newborn screening results or any Department recommendations suggested on the report.

Services for Infants with Sickle Cell Disease and Other Hemoglobin Disorders

- Family education, genetic counseling and diagnostic services are available to all families of infants with sickle cell disease, other hemoglobinopathies, and those who carry a hemoglobin disorder trait.
    - The Illinois Department of Public Health, through grants to university-based medical clinics, provides diagnostic and treatment services for infants and children identified with sickling hemoglobin disorders or traits. See Appendix for a listing of medical centers providing pediatric hematology services, or call 217-785-8101 for a listing of Department designated pediatric hematologists.
    - Additional services for infants, children and adults with thalassemia disorders are available through Children's Memorial Hospital Comprehensive Thalassemia Center, Division of Hematology-Oncology-Transplantation in Chicago; call 773-880-4125 for more information about these services.
    - In addition, by Memorandum of Understanding, the Department has joined with the Sickle Cell Disease Association of Illinois (SCDAI) to provide educational services to families of individuals with sickle cell diseases, other hemoglobinopathies or traits. Please call 312-345-1100 to learn more about SCDAI services.

University of Illinois at Chicago, Division of Specialized Care for Children (DSCC)

- DSCC provides payment for the initial diagnostic services for infants with certain abnormal metabolic newborn screening results provided these services are coordinated by specialists jointly approved by the Department and DSCC. The specific disorders for which these services are available are determined by DSCC.
    - DSCC provides these services in conjunction with other third-party payers, and remains the payer of last resort.
    - DSCC covers the ongoing medical care for infants and children diagnosed with certain metabolic disorders, and in those cases in which the family meets certain eligibility requirements.
    - DSCC does not provide initial diagnostic services or ongoing medical care for infants and children with sickle cell disease and other hemoglobinopathies, those who are carriers of a hemoglobin disorder, or for children with endocrine disorders.

Local Public Health Departments

- The Illinois Department of Public Health works in cooperation with local public health departments to provide follow-up services to the families of infants with abnormal newborn screening test results and infants diagnosed with newborn screening disorders. In some cases, the assistance of a local public health nurse may be requested in order to locate and assist the family in obtaining necessary follow-up services.
    - Newborn Screening Program staff and/or the infant's pediatrician may have difficulty locating the families of infants with abnormal test results, or parents may not understand the importance of seeking additional medical care.
    - Following the diagnosis of an endocrine, metabolic or hemoglobin disorder, the pediatric medical specialist may recommend community support services for the families of children with these serious disorders.
    - On occasion as families relocate or change medical care providers, the medical specialist may lose contact with the parents of children diagnosed with endocrine, metabolic and hemoglobin disorders. The assistance of a local health department public health nurse may be requested to locate these families and assure continuity of long-term care for the children.
    - A list of local public health department contacts may be requested by calling 217-785-8101.

Newborn Screening Disorders

Amino Acid/Urea Cycle Disorders

These disorders are inherited as autosomal recessive defects of amino acid metabolism. Each amino acid disorder is associated with a specific enzyme defect. Affected infants cannot properly metabolize certain amino acids, resulting in elevated levels of the amino acid or metabolites in body fluids. Accumulation of amino acids or metabolites may become neurotoxic, causing damage to organs and resulting in developmental delays, mental retardation or death. Clinical findings may include poor feeding, vomiting, lethargy or irritability, seizures, coma, respiratory distress and liver damage. MSUD may cause metabolic decompensation, and infants with this condition may require peritoneal dialysis or hyperalimentation without branched-chain amino acids (leucine/isoleucine and valine). See the Department's Web site, www.idph.state.il.us, for newborn screening fact sheets with additional information about these disorders and visit the American College of Medical Geneticists Web site at www.acmg.net to review the ACMG "ACT" sheets.

Urea cycle disorders involve defects in the breakdown of proteins and the conversion of ammonia and bicarbonate to urea for elimination of waste nitrogen. The resulting accumulation of ammonia in blood and tissues is neurotoxic and requires immediate detection and medical intervention. Urea cycle disorders may result in severe hyperammonemia. Infants with this condition require prompt medical intervention that may include hemodialysis. See the Department's Web site for newborn screening fact sheets with additional information about urea cycle disorders and visit the American College of Medical Geneticists Web site to review the ACMG "ACT" sheets.

In Illinois, newborn screening includes screening for the following amino acid disorders:

Phenylketonuria (PKU) also see section on Phenylketonuria
Maple syrup urine disease (MSUD)
Tyrosinemia* type 1, and possibly types 2 and 3
Homocystinuria (HCU)
5-oxoprolinuria
Urea cycle disorders: Citrullinemia (ASAS), Argininosuccinic aciduria (ASAL) and Argininemia

- Incidence **PKU** and hyperphenylalaninemia - one in 10,000 births
  MSUD - one in 200,000 births
  Tyrosinemia - one in 500,000 births (one in 12,500 births among French Canadian populations)
  Homocystinuria - one in 200,000 births
  5-oxoprolinuria - extremely rare, actual incidence unknown
  Urea Cycle Disorders - one in 200,000 to 300,000 births

- Analytes Measured in Screening Measurement of specific analytes by tandem mass spectrometry (MS/MS) reported in micro-Moles/Liter (uM/L)

- **Analytes Measured in Screening**** **Possible Disorder****

  Phenylalanine — Phenylketonuria (PKU)

Amino Acid/Urea Cycle Disorders - Continued

• **Analytes Measured in Screening****	**Possible Disorder****
Tyrosine	Tyrosinemia
Methionine	Homocystinuria (HCU)
(Iso)leucine and Valine	Maple syrup urine disease (MSUD)
Citrulline	Citrullinemia (ASAL) or Argininosuccinic aciduria (ASAS)
Arginine	Argininemia
5-oxoproline	5-oxoprolinuria

• **Reporting Ranges***** Abnormal results reported by phone, letter, fax to physician of record or hospital contact unless otherwise specified.	**Follow-up Referrals and Testing**
Presumptive Positive	Immediate referral to pediatric metabolic disease specialist
Suspect Borderline Abnormal	Medical evaluation and repeat newborn screening specimen within one to two days; mark retest box on specimen card.
Suspect Amino Acid Abnormal due to TPN (reported by fax and letter)	If baby is still in NICU or on TPN, repeat repeat newborn screen when off TPN 48 hours, at day 14 of life, or prior to discharge, whichever comes first. If baby has been discharged from hospital or was not on TPN at time of specimen collection, evaluate infant's status and repeat newborn screen within one to two days.
• Feeding Effect	Hyperalimentation and TPN may affect results. Unless otherwise instructed, repeat specimens are best collected 48 hours after TPN is discontinued.
• Transfusion Effect	Specimen collection prior to transfusion is always recommended. If post-transfusion specimen collection is necessary, collection is best performed 48 hours post-transfusion.

Amino Acid/Urea Cycle Disorders - Continued

• Timing Effect	If specimen is collected at less than 24 hours of age, submit second sample during second day of life.
• Specialist	Metabolic disease specialist (Department/DSCC designated), see Appendix for the contact information.
• Treatment	Treatment is disorder specific and may include specialized prescription medical formula, special diet limited in specific proteins and, in some cases, supplements and medications. The Department provides special medically necessary formula without charge to Illinois residents who are under comprehensive medical management provided by a metabolic disease specialist designated by the Department.

Comments

*Newborn screening may not detect all cases of tyrosinemia as tyrosine levels may not increase to detectable levels until after the fifth day of life.

**Additional amino acid levels may be reported when significant abnormalities are detected as by-products of tandem mass spectrometry, and in most cases the laboratory recommendation will suggest immediate consult with a pediatric metabolic specialist. These abnormalities may include:

Elevated Glycine – possibly indicative of Non-ketotic hyperglycinemia
Low Citrulline – possibly indicative of Ornithine transcarbamylase deficiency
Low Citrulline/elevated Glutamine – possibly indicative of Carbamoylphosphate synthetase deficiency
Elevated Ornithine and elevated Citrulline – possibly indicative of Hyperammonemia/ Ornithinemia/Citrullinemia (HHH)

***For questions about the most current laboratory reporting values for these disorders, contact the Department's Division of Laboratories at 312-793-4752.

Biotinidase Deficiency

This is an autosomal recessive disorder of biotin recycling that leads to multiple carboxylase deficiencies. Individuals with biotinidase deficiency cannot recycle biotin (vitamin B), and cannot process dietary protein-bound biotin. Early detection and treatment is essential to prevent permanent neurological damage. See the Department's Web site, www.idph.state.il.us, for newborn screening fact sheets with additional information about this disorder and visit the American College of Medical Geneticists Web site at www.acmg.net to review the ACMG "ACT" sheets.

• Incidence	One in 180,000 births
• Analyte Measured in Screening	Biotinidase enzyme activity is determined by colorimetric analysis.
• **Reporting Ranges** Abnormal results reported by phone, letter and fax to physician of record or hospital contact	**Follow-up Referrals and Testing**
Biotinidase activity absent (initial specimen)	Medical evaluation and collect repeat newborn screen within one to two days; mark retest box on specimen card. If retest sample does not have detectable biotinidase activity, refer to Department designated metabolic specialist for diagnostic testing.
• Feeding Effect	None
• Transfusion Effect	Screening is affected.* Specimen should always be collected prior to transfusion, even if infant is younger than 24 hours of age. If initial specimen collected post-transfusion a second specimen is required 48 hours post- transfusion and third specimen is required three months following the last transfusion.
• Timing Effect	If specimen is collected at less than 24 hours of age, submit second sample during second day of life.
• Specialist	Metabolic disease specialist (Department/DSCC designated), see Appendix for the contact information
• Treatment	Daily prescription dosage of biotin supplement

Comments

Improper collection and care of specimens may cause biotinidase enzyme degradation.
Exposure of specimens to excessive heat and/or delayed submission may result in false positive screening results.

*Transfusions may have long-term effects due to biotinidase activity of transfused red blood cells and may result in false negative screens for biotinidase deficiency.

Congenital Adrenal Hyperplasia

This is an autosomal recessive disorder of steroid hormone synthesis; 90 percent of individuals with congenital adrenal hyperplasia (CAH) cannot produce adequate amounts of the enzyme 21-hydroxylase, which is necessary for synthesis of cortisol. In Illinois, newborn screening includes testing for CAH due to 21-hydroxylase deficiency. In 50 percent to 75 percent of 21-hydroxylase deficiency cases, in addition to cortisol deficiency, the infant cannot synthesize adequate amounts of aldosterone, resulting in salt-wasting CAH. In utero, the developing fetus with CAH is exposed to excessive levels of androgen, and female infants may have varying degrees of virilization of external genitalia. Male infants usually appear normal at birth. Both males and females are susceptible to acute adrenal insufficiency. Infants with salt-wasting CAH are very susceptible to electrolyte imbalance and dehydration. Early detection and treatment of CAH is essential to prevent adrenal crisis, dehydration and sudden death in the first few weeks of life. See the Department's Web site, www.idph.state.il.us, for newborn screening fact sheets with additional information about this disorder and visit the American College of Medical Geneticists Web site, www.acmg.net, to review the ACMG "ACT" sheets.

• Incidence	One in 20,000 births
• Analyte Measured in Screening	17-OH (hydroxy) progesterone (17-OHP) level is measured by fluorometric assay, reported in *nanograms per milliliter(ng/mL)**

Due to the effects of pre-maturity and physiological stress on 17-OHP levels, a tiered of reporting for abnormal results has been developed based on birthweight and/or gestational age of newborns by the Department of Newborn Screening Laboratory.

Pre-term infants are defined as gestational age less than or equal to 36 wks.
Low birth weight is defined as birth weight less than 2000 grams.
Very low birth weight is defined as less than or equal to 1500 grams.

• **Reporting Ranges**** Abnormal results reported by phone, letter and fax to physician of record or hospital contact unless otherwise specified.	**Follow-up Referrals and Testing**
Presumptive positive abnormal Full-term infant $\geq$55 ng/mL	Immediate consult /referral to pediatric endocrinologist and serum electrolytes and 17-OHP
Pre-term infant/low birth weight $\geq$80 ng/mL	Consultation with pediatric endocrinologist; evaluation of risk for CAH; serum electrolytes and 17-OHP or repeat newborn screen. If retest remains abnormal, refer to pediatric endocrinologist.

Note: While neonates in the NICU*** with abnormal CAH screens need additional testing to rule out CAH, the neonatologist may use discretion monitoring in these cases.

Congenital Adrenal Hyperplasia - Continued

• **Reporting Ranges****	**Follow-up Referrals and Testing**
Suspect borderline abnormal (reported by letter only) Full-term infant 30 - 54 ng/mL	Medical evaluation and repeat newborn screen within one to two days, unless baby was admitted to NICU and will be tested at a later date.***If retest remains abnormal, refer to pediatric endocrinologist.
Pre-term, low birth weight 55 - 79 ng/mL	Medical evaluation and repeat newborn screen within one to two days, unless baby was admitted to NICU and will be tested at a later date.*** If retest remains abnormal refer to pediatric endocrinologist.
• Feeding Effect	None
• Transfusion Effect	If post-transfusion specimen collection is required, sample should be obtained 48 hours post-transfusion; enter date of last transfusion prior to specimen collection on specimen card.
• Timing Effect	If specimen is collected at less than 24 hours of age, submit second sample during second day of life.
• Specialist	Pediatric endocrinologist (Department/DSCC designated), see Appendix for the contact information.
• Treatment Simple virilizing CAH	Daily cortisol replacement therapy
Salt-wasting CAH	Daily cortisol and aldosterone replacement therapy, dietary salt supplements

Comments

Factors such as specimen collection prior to 24 hours of age, pre-term, low birth weight and illness may cause elevation of 17-OH progesterone levels. However, follow-up screening and/or diagnostic testing are necessary to rule out this life-threatening disorder.

Infants with any symptoms of vomiting, dehydration, weight loss, poor feeding, electrolyte imbalance and/or lethargy, require immediate medical attention, emergency care and referral to a pediatric endocrinologist.

Treatment with glucocorticoids (cortisone, dexamethasone) may cause <u>false negative</u> results.

Congenital Adrenal Hyperplasia - Continued

***Most reference laboratories measure 17-OHP levels in nanograms per deciliter (ng/dL)** and caution should be used when comparing newborn screening results (ng/mL) with reference laboratory results; please consult with a pediatric endocrinologist when in doubt about diagnostic testing results for possible CAH.

**Laboratory reporting cut-off values are procedure dependent and are subject to periodic revision. For questions about the most current laboratory cut-off values for this disorder, contact the Department's Division of Laboratories at 312-793-4752.

***Administrative Rules for Newborn Screening require that a routine repeat specimen be collected from all infants admitted to a NICU or Special Care Nursery. Second specimen should be collected on day 14 of life or prior to discharge, if baby is discharged before 2 weeks of age.

Congenital Hypothyroidism

Congenital hypothyroidism results from an inability of the thyroid gland to produce adequate amounts of the hormone, thyroxine. Congenital hypothyroidism is usually due to a failure of the thyroid gland to develop properly in utero. Less frequently, the disorder can result from an autosomal recessive defect in thyroid hormone synthesis. Primary congenital hypothyroidism usually requires lifetime treatment. Occasionally cases of transient hypothyroidism occur as a result of maternal anti-thyroid medications or temporary thyroid suppression in the infant due to exposure to iodine, prematurity or other causes. Consultation with a pediatric endocrinologist is usually recommended in these cases. See the Department's Web site, www.idph.state.il.us, for newborn screening fact sheets with additional information about this disorder and visit the American College of Medical Geneticists Web site at www.acmg.net to review the ACMG "ACT" sheets.

• Incidence	One in 2,000 births
• Analytes Measured in Screening	Thyroid stimulating hormone (TSH) and thyroxine (T4). If TSH is elevated (within top 4 percentile for each day), T4 level is determined. Levels are tested by fluorometric assay.
• **Reporting Ranges*** All abnormal results reported by phone, letter and fax, unless otherwise specified.	**Follow-up Referrals and Testing**
Presumptive positive abnormal TSH $\geq$ 54 uIU/mL T4 < 5 ug/dL	Immediate referral to pediatric endocrinologist, serum TSH and free T4
Suspect borderline abnormal TSH $\geq$36-54 uIU/mL T4 $\geq$5-8 ug/dL (reported by letter only)	Medical evaluation and repeat newborn screen within one to two days; mark retest on specimen card. If retest remains abnormal, refer to pediatric endocrinologist.
• Feeding Effect	None
• Transfusion Effect	If post-transfusion specimen collection is required, collection is best performed 48 hours post-transfusion; enter date of last transfusion prior to specimen collection on the specimen card.
• Timing Effect	If specimen is collected at less than 24 hours of age, submit second sample during second day of life. In pre-term infants, production of TSH may be delayed in first few days of life.

Congenital Hypothyroidism – Continued

• Specialist	Pediatric endocrinologist (Department/DSCC designated), see Appendix for contact information.
• Treatment	Daily thyroid supplement

Comments

TSH increases dramatically in first few hours after birth and gradually returns to normal levels in about 72 hours. This normal TSH elevation will be detected if the specimen is collected before the infant is 24 hours of age.

Very low birth weight infants and infants with cardiac defects, congenital craniofacial anomalies, and Down syndrome may be at increased risk of late onset hypothyroidism, and consult with pediatric endocrinology and/or diagnostic testing is advisable regardless of newborn screening results.

Although newborn screening can detect congenital hypothyroidism with a high degree of accuracy, other forms of hypothyroidism may develop within the first few weeks of life. The physician therefore must remain alert to clinical symptoms in older infants despite normal newborn screening results. Repeat the screening or refer to pediatric endocrinologist if any suspicions exist about possible hypothyroidism, regardless of newborn screening results.

Family history of thyroid disorders may indicate the need for diagnostic testing or pediatric endocrinology consult regardless of newborn screening results.

*Laboratory reporting cut-off values are procedure dependent and are subject to periodic revision. For questions about the most current laboratory cut-off values for this disorder, contact the Department's Division of Laboratories at 312-793-4752.

Cystic Fibrosis

Cystic fibrosis (CF) is an autosomal recessive disorder that results in production of a defective form of cystic fibrosis transmembrane conductance regulator (CFTR) protein. CFTR protein is an important chloride channel within epithelial cells of multiple organs, and regulates movement of salt and water into and out of the cells. In individuals with CF, the cells lining passageways of the lungs, pancreas and other organs produce thick, sticky mucus. Clinical signs and disease progression vary among affected individuals, but may include progressive lung disease, pancreatic insufficiency, male infertility and elevated sweat chloride levels. Early detection and diagnosis with adequate nutritional support and aggressive therapies to reduce risks of respiratory exacerbations have been shown to improve clinical outcomes. See the Department Web site, www.idph.state.il.us, for newborn screening fact sheets with additional information about this disorder and visit the American College of Medical Geneticists Web site at www.acmg.net to review the ACMG "ACT" sheet.

• Incidence	Varies with race and ethnicity One in 3,200 Caucasian births One in 9,000 Hispanic births One in 15,000 African-American births One in 30,000 Asian births
• Analyte Measured in Screening	Measurement of immunoreactive trypsinogen (IRT)* level by fluorometric assay reported as nanograms per milliliter (ng/mL). If the IRT is in the top 4 percentile for the day, DNA mutation analysis for a panel of CFTR mutations is also performed.
• **Reporting Ranges***	**Follow-up Referrals and Testing**
Abnormal results reported by phone, letter, fax to physician of record or hospital contact.	
Presumptive Positive	
ITR** in top 4 percentile, two CFTR mutations	Immediate referral*** to CF specialist for diagnostic sweat testing and genetic counseling services.
IRT** in top 4 percentile, one CFTR mutation	
IRT $\geq$ 170ng/mL IRT **	
• Feeding Effect	None

Cystic Fibrosis - Continued

• Transfusion Effect	Screening may be affected. Specimen should always be collected prior to transfusion, even if infant is younger than 24 hours of age. If initial specimen was collected post-transfusion, a second specimen is required 48 hours post-transfusion and third specimen is required three months following the last transfusion.
• Timing Effect	If specimen is collected at less than 24 hours of age, submit second sample during second day of life.
• Treatment	Dietary and vitamin supplements, respiratory therapies and frequent visits for evaluation and support are usually provided.
• Specialist	Pediatric pulmonologist and/or pediatric gastroenterologist specializing in care of children with CF, (Department/DSCC designated), see Appendix for contact information.

Comments

*Laboratory reporting cut-off values are procedure dependent and are subject to periodic revision. For questions about the most current laboratory cut-off values for this disorder, contact the Department's Division of Laboratories at 312-793-4752.

**Meconium ileus has been clinically associated with the diagnosis of cystic fibrosis in newborn infants, and any infant with this condition should have diagnostic evaluation through a CF specialist, regardless of the infant's newborn screening results. In some cases, normal IRT levels causing false negative newborn screens for CF have been reported in babies with meconium ileus.

***All babies with abnormal CF newborn screening results should be referred to a CF specialist for confirmatory pilocarpine iontophoresis sweat testing and genetic counseling services. Sweat testing for newborns should only be performed under the direction of a CF specialist at laboratories in compliance with Clinical Laboratory Standards Institute (CLSI) guidelines.

Fatty Acid Oxidation Disorders

Fatty acid oxidation (FAO) disorders are autosomal recessive inherited metabolic conditions. Each FAO disorder is associated with a specific enzyme defect in the fatty acid metabolic pathway, and affects utilization of dietary and stored fats. These disorders lead to an accumulation of fatty acids in the body, or an inability to breakdown dietary or stored fats, with a decrease in cell energy metabolism. Many of the FAO disorders cause a significant risk of death during the first clinical episode. In most cases, the first episode arises following illness or fasting, and occurs in infancy or early childhood. FAO disorders can cause recurrent episodes of hypoglycemia. Clinical findings may include lethargy, hypotonia, failure to thrive, persistent vomiting and hepatomegaly, rhabdomyolysis, and Reye syndrome-like episodes. Significant disability may result from prolonged episodes of hypoglycemia. See the Department's Web site, www.idph.state.il.us, for newborn screening fact sheets with additional information about these disorders and visit the American College of Medical Geneticists Web site at www.acmg.net to review the ACMG "ACT" sheets.

In Illinois, newborn screening includes testing for a panel of acylcarnitines. Following is a list of the FAO disorders that may be detected:

Short chain acyl-CoA dehydrogenase deficiency (SCADD)
Medium chain acyl-CoA dehydrogenase deficiency (MCADD)
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
Trifunctional protein deficiency (TFP)
Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)
Carnitine palmitoyl transferase deficiency type 2 (CPT 2)
Carnitine/acylcarnitine translocase deficiency (CACT)
Carnitine palmitoyl transferase deficiency type 1 (CPT 1 or CPT 1A)
Glutaric aciduria type 2 (GA 2)/Multiple acyl-CoA dehydrogenase deficiency (MADD)
Carnitine uptake defect (CUD)
Medium/Short chain L-3-hydroxyacyl-CoA dehydrogenase deficiency (M/SCHAD)
Isobutyryl-CoA dehydrogenase deficiency (IBCD)*

- Incidence — MCADD - one in 18,000 births
  LCHAD - one in 400,000 births
  VLCAD - one in 100,000 births
  SCADD - one in 20,000 births
  Other Disorders - one in 500,000 births

- Analytes Measured in Screening — Specific acylcarnitines** measured by tandem mass spectrometry (MS/MS), reported as micro-Moles per Liter (μM/L)

Analytes	**Possible Disorder**
Butanoyl carnitine (C4) is the primary analyte	SCADD, IBCD*, GA2/MADD, MSCHAD
Multiple medium chain acylcarnitines Octanoyl carnitine (C8) is the primary analyte (C8, C10, C10:1 and C6)***	MCADD
Multiple long chain acylcarnitines Hydroxy-hexadecanoyl carnitine (C16:OH) is the primary analyte (C16-OH, C18:1-OH)***	LCHAD/TFPD

Fatty Acid Oxidation Disorders – Continued

Analytes	**Possible Disorder**
Multiple long chain acylcarnitines Tetradecenoyl carnitine (C14:1) is the primary analyte (C14:1, C14 and C16)***	VLCAD
Multiple long chain acylcarnitines Hexadecanoyl carnitine (C16) or Octadecenoyl carnitine (C18:1) is the primary analyte (C16, C18:1 and C18:2)***	CPT 2/CACT
Free carnitine (elevated level) is the primary analyte (C0, and C0/C16+C18 ratio elevated)	CPT1/CPT1A
Multiple acylcarnitines (C4, C5, C8, C12, C14, C16 and C5-DC)***	GA 2/MADD
Free carnitine (decreased level) is the primary analyte (C0, and C0/C16+C18 ratio decreased)	CUD
Acylcarnitine 3-hydroxy-butyryl carnitine (C4-OH) is the primary analyte	M/SCHAD

• **Reporting Ranges** Abnormal results reported by phone, letter, fax to physician of record or hospital contact unless otherwise specified.	**Follow-up Referrals and Testing**
Presumptive Positive	Immediate referral to pediatric metabolic specialist
Suspect Borderline Abnormal	Medical evaluation and repeat newborn specimen within one to two days; mark retest on specimen card. If retest remains abnormal, refer to metabolic specialist.
Suspect Acylcarnitine Abnormal due to TPN (reported by fax and letter)	**If baby is still in NICU or on TPN, repeat** newborn screen when off TPN 48 hours, at day 14 of life, or prior to discharge, whichever comes first. If baby has been discharged from hospital or was not on TPN at time of specimen collection, repeat newborn screen within one to two days. If retest remains abnormal, refer to metabolic specialist.

Fatty Acid Oxidation Disorders – Continued

• Feeding Effect	Hyperalimentation, TPN and certain medications may affect results. Unless otherwise instructed, repeat specimens are best collected 48 hours after TPN is discontinued.
• Transfusion Effect	Specimen collection prior to transfusion is always recommended. If post-transfusion specimen collection is required, collection is best performed 48 hours post-transfusion.
• Timing Effect	If specimen is collected at less than 24 hours of age, submit second sample during second day of life. If repeat screening is requested, collect and submit new specimen within one to two days. **Acylcarnitine levels tend to normalize very quickly in newborns once the baby begins to feed well**; specimen collection as soon as possible after 24 hours of age is optimal and repeat specimens must be collected quickly to avoid false negative results for FAO disorders.
• Specialist	Pediatric metabolic disease specialist, see Appendix for a list of Department /DSCC designated medical specialists.
• Treatment	**Treatment is disorder specific** and usually includes frequent feeding and avoidance of fasting, high carbohydrate, low-fat diet and, in some cases, specialized medical formula, or supplements and medications. Special precautions, such as glucose monitoring and, in some cases, intravenous therapy may be required during intercurrent illnesses. If indicated, the Department provides special medically necessary formula without charge to Illinois residents who are under comprehensive medical management provided by a metabolic disease specialist designated by the Department.

Comments

*IBCD is categorized as an organic acid disorder in some reference materials.

**For substituted carnitines, a notation of (Cx) is used where (x) denotes the number of carbons in the fatty acid radical. Free carnitine is designated as (C0), acetyl carnitine as (C2), and propanoyl carnitine as (C3), etc. Hydroxylation is designated by (-OH), dicarboxylic acid is designated by (-DC), and unsaturation is designated by (:1). See Appendix for a complete listing of acylcarnitine analytes measured by the Department's laboratory tandem mass spectrometry.

***A particular acylcarnitine pattern, or group of abnormal acylcarnitines may be detected by MS/MS screening; while not diagnostic, these patterns may be suggestive of a certain type of fatty acid oxidation disorder.

****For questions about the most current laboratory reporting values for these disorders, contact the Department's Division of Laboratories at 312-793-4752.

Galactosemia

An autosomal recessive disorder of galactose metabolism, galactosemia is due to insufficient enzyme activity, usually of galactose-1-phosphate uridyl transferase (GALT) or, in some cases, deficiency of galactokinase or uridine diphosphate galactose-4-epimerase. In Illinois, newborn screening for galactosemia is designed to detect classical galactosemia due to GALT enzyme deficiency. These enzymes are necessary to convert galactose to glucose for energy and cellular growth. The main dietary source of galactose is lactose, the principle carbohydrate found in all forms of milk. Early detection and treatment of classical galactosemia is essential to prevent severe liver disease and complications including bleeding, overwhelming sepsis and death in the early neonatal period. See the Departments Web site, www.idph.state.il.us, for newborn screening fact sheets with additional information about these disorders and visit the American College of Medical Geneticists Web site at www.acmg.net to review the ACMG "ACT" sheets.

• Incidence	One in 75,000 births (classical galactosemia)
• Analyte Measured in Screening	Total galactose (free galactose and galactose-1-phosphate) and GALT enzyme activity are determined by fluorometric assay
• **Reporting Ranges*** Abnormal results reported by phone, letter, fax to physician of record or hospital contact.	**Follow-up Referrals and Testing**
Presumptive Positive Total galactose > 6.5mg/dL with No GALT activity or Total galactose ≥19.5mg/dL with Normal GALT activity	Immediate referral to metabolic disease specialist for diagnostic testing and **change feeding to soy formula**; encourage breast feeding mothers to temporarily avoid nursing and use breast pump to maintain milk supply until metabolic specialist advises otherwise.
Suspect Borderline Abnormal Total galactose ≥ 6.5mg/dL with Reduced GALT activity	Evaluate baby's condition, collect repeat newborn screening specimen within one to two days; mark retest box and indicate the baby's feeding type on the specimen card. If retest sample remains abnormal, refer to metabolic specialist.
• Feeding Effect	Screening will be affected. Mark specimen collection form to indicate type of feeding.
• Transfusion Effect	Screening is affected.** Specimen should always be collected prior to transfusion, even if infant is younger than 24 hours of age. If initial specimen was collected post- transfusion a second specimen is required 48 hours post-transfusion and third specimen is required three months following the last transfusion.

Galactosemia - Continued

• Timing Effect	If specimen is collected at less than 24 hours of age, submit second sample during second day of life.
• Treatment	Soy or galactose-free formula and lactose free diet. Variant forms of galactosemia may or may not require dietary restrictions.
• Specialist	Pediatric metabolic disease specialist (Department/DSCC designated), see Appendix for the contact information.

Comments

Improper specimen collection or inappropriate shipping and handling, including exposure of the specimen to excessive heat, humidity and/or delayed submission may cause GALT enzyme degradation.

*The Department laboratory cut-off values are procedure dependent and are subject to periodic revision. For questions about the most current laboratory cut-off values for this disorder, contact the Department's Division of Laboratories at 312-793-4752.

**Transfusions may have long-term effects due to GALT enzyme activity of transfused red blood cells and may result in false negative screening for classical galactosemia.

Organic Acid Disorders

Organic acid disorders are autosomal recessive inherited metabolic conditions. Each organic acid disorder is associated with a specific enzyme deficiency that causes the accumulation of organic acids in blood and urine. The accumulated compounds or their metabolites are toxic, and may result in metabolic acidosis, hyperammonemia or ketotic hyperglycinemia. Usually infants with organic acid disorders appear normal at birth, but may develop vomiting, poor feeding, hypoglycemia, seizures, hypotonia and lethargy progressing to coma. Common features may include ketotic hyperglycinemia, metabolic acidosis and sometimes, an unusual odor. There is significant risk of death in infancy due to organic acid disorders; early diagnosis and treatment can greatly improve disease outcome. Minimization of ketoacidotic episodes improves prognosis and, during such episodes, aggressive treatment including administration of glucose may be warranted. See the Department's Web site, www.idph.state.il.us, for newborn screening fact sheets with additional information about these disorders and visit the American College of Medical Geneticists Web site at www.acmg.net to review the ACMG "ACT" sheets.

In Illinois, newborn screening includes testing for a panel of acylcarnitines. Following is a list of the organic acid disorders that may be detected:

Propionic acidemia (PA)
Methylmalonic acidemia (MMA)
Isovaleric acidemia (IVA)
3-hydroxy-3-methylglutaryl-CoA lyase deficiency (3HMG)
3-methylglutaconic aciduria (3MGA)
3-methylcrotonyl CoA carboxylase deficiency (3-MCC)
Glutaric acidemia, type 1 (GA 1)
2-methylbutyryl-CoA dehydrogenase deficiency (2MBCD)
Malonic aciduria (MA)
Betaketothiolase deficiency (BKT)
Multiple carboxylase deficiency (MCD)

• Incidence	IVA - one in 50,000 births 3MCC - one in 50,000 births MMA - one in 100,000 births GA1 - one in 110,000 births PA, 3HMG, 3MGA, BKT, MCD, MA - all very rare, estimated at one in 100,000 to 500,000 births
• Analytes Measured in Screening	Specific acylcarnitines* measured by tandem mass spectrometry (MS/MS) in micro-Moles per Liter (uM/L)

Acylcarnitine	**Possible Disorder**
3-hydroxy-isovaleryl carnitine (C5-OH) is the primary analyte	3MCC, 3HMG, 3MGA, MCD
Pentanoyl carnitine (C5) is the primary analyte	IVA/2MBCD
Propanoyl carnitine (C3) is the primary analyte	MMA/PA

Organic Acid Disorders - Continued

Analytes	**Disorder**
Pentenoyl carnitine (C5:1) is the primary analyte	BKT
Glutaryl carnitine (C5-DC) is the primary analyte	GA1
Malonoyl carnitine (C3-DC) is the primary analyte	MA
• **Reporting Ranges**** Abnormal results reported by phone, letter and fax to physician of record or hospital contact unless otherwise specified.	**Follow-up Referrals and Testing**
Presumptive Positive	Immediate referral to pediatric metabolic specialist
Suspect Borderline Abnormal	Medical evaluation and repeat newborn screening specimen within one to two days; mark retest on specimen card. If retest sample remains abnormal, refer to metabolic specialist.
Suspect Amino Acid Abnormal due to TPN (reported by fax and letter)	If baby is still in NICU or on TPN repeat newborn screen when off TPN 48 hours, at day 14 of life, or prior to discharge, whichever comes first. If baby has been discharged from hospital or was not on TPN at time of specimen collection, repeat newborn screen within one to two days. If retest sample remains abnormal, refer to metabolic specialist.
• Feeding Effect	Hyperalimentation and TPN may affect results. Unless otherwise instructed, repeat specimens are best collected 48 hours after TPN is discontinued.
• Transfusion Effect	Specimen collection prior to transfusion is always recommended. If post-transfusion specimen collection is required, collection is best performed 48 hours post-transfusion.

Organic Acid Disorders - Continued

• Timing Effect	If specimen is collected at less than 24 hours of age, submit second sample during second day of life. If repeat screening is requested, collect and submit new specimen within one to two days.
• Specialist	Pediatric metabolic disease specialist (Department/DSCC designated), see Appendix for the contact information.
• Treatment	**Treatment is disorder specific** and may include low-protein diet and avoidance of fasting and, in some cases, specialized medical formula, or supplements and medications. Special precautions, such as close monitoring and in some cases, intravenous therapy may be required during intercurrent illness and introduction of new foods. If indicated, the Department provides special medical formula without charge to Illinois residents who are under comprehensive medical management provided by a metabolic disease specialist designated by the Department.

Comments

*For substituted carnitines, a notation of (Cx) is used where (x) denotes the number of carbons in the fatty acid radical. Free carnitine is designated as (C0), acetyl carnitine as (C2), propanoyl carnitine as (C3), etc. Hydroxylation is designated by (-OH), dicarboxylic acid is designated by (-DC), and unsaturation is designated by (:1). See Appendix for a complete listing of acylcarnitine analytes measured by the Department laboratory tandem mass spectrometry.

**For questions about the most current laboratory reporting values for these disorders, contact the Department's Division of Laboratories at 312-793-4752.

Phenylketonuria

Phenylketonuria (PKU) is an autosomal recessive disorder of amino acid metabolism resulting in excess levels of phenylalanine in body fluids. Hyperphenylalaninemia is usually due to a deficiency of the phenylalanine hydroxylase enzyme or, in some cases, impaired synthesis or recycling of biopterin cofactor. Elevated levels of phenylalanine can become neurotoxic; early detection and treatment of hyperphenylalaninemia is necessary to prevent severe mental retardation. Phenylalanine is an essential amino acid, and individuals with PKU require careful dietary management and monitoring for life. Women of childbearing age who are diagnosed with PKU or hyperphenylalaninemia require strict dietary control prior to conception and throughout pregnancy to reduce their risk of pregnancy complications, including miscarriage, or of having an infant with severe birth defects due to high maternal levels of phenylalanine. See the Department's Web site, www.idph.state.il.us, for newborn screening fact sheets with additional information about this disorder and visit the American College of Medical Geneticists Web site at www.acmg.net to review the ACMG "ACT" sheets.

• Incidence	One in 10,000 births (Classical PKU)
• Analytes Measured in Screening	Phenylalanine level and phenylalanine/tyrosine ratio measured by tandem mass spectrometry (MS/MS) in micro-Moles per Liter (uM/L).
• **Reporting Ranges*** Abnormal results reported by phone, hospital contact unless otherwise specified.	**Follow-up Referrals and Testing**
Presumptive Positive* Seriously elevated phenylalanine level and/or elevated phenylalanine/ tyrosine ratio	Immediate referral to PKU metabolic disease specialist
Suspect Borderline Abnormal Slightly elevated phenylalanine level	Medical evaluation and repeat newborn screening specimen within one to two days; mark retest on specimen card. If retest sample remains abnormal, refer to metabolic specialist.
<u>Suspect Amino Acid Abnormal due to TPN</u> (reported by letter and fax) Slightly elevated phenylalanine level	If baby is still in NICU or on TPN, repeat newborn screen when off TPN 48 hours, at day 14 of life, or prior to discharge, usually associated with TPN effects whichever comes first. If baby has been discharged from hospital or was not on TPN at time of specimen collection, repeat newborn screen within one to two days. If retest sample remains abnormal, refer to metabolic specialist.

Phenylketonuria - Continued

• Feeding Effect	Laboratory detection of phenylalanine is not necessarily diet dependent. Screening is sensitive to catabolic activity occurring shortly after birth. TPN and hyperalimentation may cause false positive screening results.
• Transfusion Effect	Specimen collection prior to transfusion is always recommended. If post-transfusion specimen collection is required, collection is best performed 48 hours post-transfusion.
• Timing Effect	If specimen is collected at less than 24 hours of age, submit second sample during second day of life.
• Specialist	Pediatric metabolic disease specialist (Department/DSCC designated), see Appendix for the contact information.
• Treatment	Low phenylalanine diet consisting of specialized medically necessary formula in combination with foods that are low in phenylalanine. The Department provides PKU medically necessary formula without charge to Illinois residents who are under comprehensive medical management provided by a metabolic specialist designated by the Department/DSCC.

Comments

*For questions about the most current laboratory reporting values for these disorders, contact the Department's Division of Laboratories at 312-793-4752.

Sickling Hemoglobin Disorders, Alpha and Beta Thalassemias (FS, FSC, FSa, FNA, Bart's)

This group of autosomal recessive hemoglobin disorders is characterized by production of abnormal forms of hemoglobin and no adult, or normal hemoglobin. In sickling diseases, this abnormal hemoglobin may be less stable, and may cause red blood cells to sickle after repeated de-oxygenation. Sickled cells may block blood vessels causing pain, stroke and other complications. The severity of the disorder varies greatly, but infants with sickle cell disease, sickle hemoglobin C disease, sickle beta thalassemia and beta thalassemia major are very susceptible to anemia, life-threatening infections and other complications. **Prophylactic penicillin by 2 months of age and adequate immunizations have been shown to greatly reduce morbidity and mortality associated with sickling hemoglobinopathies.** Infants and children diagnosed with alpha thalassemia or beta thalassemia major may experience severe anemia requiring blood transfusions and ongoing medical care best provided by pediatric hematologists or through the Comprehensive Thalassemia Center. See the Department Web site, www.idph.state.il.us, for newborn screening fact sheets with additional information about these disorders and visit the American College of Medical Geneticists Web site at www.acmg.net to review the ACMG "ACT" sheet.

• Incidence	Sickle cell disease occurs in one in 375 African Americans. Sickling hemoglobinopathies occur in all races and ethnic groups. Hemoglobin S is more common among individuals with West African, Middle Eastern, Mediterranean or Central Indian heritage. Hemoglobin C is more common among individuals with West African heritage. Hemoglobin E is more common among individuals with Mediterranean, African or Southeast Asian heritage. Thalassemias are more common among individuals with Mediterranean, African or Southeast Asian heritage, but do occur worldwide.
• Analyte Measured in	Identification of types of hemoglobin present in the Screening sample is performed by high performance liquid chromatography (HPLC). This screening is not quantitative.
• **Reporting Classifications** Abnormal results reported letter, phone and fax to physician of record or hospital contact	**Follow-up Referrals/Testing** Confirmatory diagnostic testing along with genetic counseling and education are necessary. Referral to pediatric hematologist for testing and genetic counseling services is highly recommended.

Sickling Hemoglobin Disorders, Alpha and Beta Thalassemias (FS, FSC, FSa, FNA, Bart's) - Continued

- Reporting Classifications
  FS - Fetal and sickle hemoglobins - probable SS sickle cell disease
  FSC - Fetal and hemoglobin C - probable sickle hemoglobin C disease
  FSa - Fetal, sickle and beta thalassemia hemoglobins - probable sickle/beta thalassemia
  FNA - Fetal and no adult hemoglobin - probable beta thalassemia major
  F-Bart's - Fetal and Bart's hemoglobin- probable alpha thalassemia/hemoglobin H disease

- Feeding Effect — None

- Transfusion Effect — Screening **will be** affected by transfusions. If initial specimen is collected post transfusion, repeat screening will be necessary three months after the last transfusion. Donor red blood cells may mask hemoglobin disorders due to presence of transfused adult hemoglobin. Specimens from transfused infants with disease conditions may have false negative results or results that falsely indicate carrier (trait) status.

- Timing Effect — Usually no effect in first two weeks of newborn period, although older infants will have a gradual decrease in fetal hemoglobin.

  Bart's hemoglobin levels decline rapidly during the neonatal period, and newborns with positive screens for possible alpha thalassemia/hemoglobin H disease require immediate diagnostic testing.*

- Specialist — Pediatric hematologist, see Appendix for contact information. *Referral to the Comprehensive Thalassemia Center at Children's Memorial Hospital is recommended for abnormal screens indicative of possible alpha thalassemia/hemoglobin H disease.

- Treatment — Prophylactic penicillin is recommended for children with sickling disorders from ages 2 months to 5 years. In addition to all regular childhood immunizations, pneumococcal conjugate vaccine also is recommended to help prevent pneumococcal infections. While prophylactic penicillin is **not** usually indicated for treatment of thalassemias, these disorders are associated with severe anemia usually requiring medical management by a pediatric hematologist. Services are also available through the Comprehensive Thalassemia Center at Children's Memorial Hospital.

Sickling Hemoglobin Disorders, Alpha and Beta Thalassemias (FS, FSC, FSa, FNA, Bart's) - continued

Comments

Family education, genetic counseling and diagnostic services are available to families of infants with sickling hemoglobin disorders. The Illinois Department of Public Health, through grants to university-based medical clinics, provides diagnostic and treatment services for infants and children identified with hemoglobin disorders or traits. In addition, family counseling and educational services about sickling disorders are offered through the Sickle Cell Disease Association of Illinois.

Other Hemoglobinopathies and Traits

Hemoglobin traits and other less serious hemoglobin diseases are autosomal recessive disorders of hemoglobin production that usually do not require treatment. Individuals with these conditions produce adequate amounts of functional hemoglobin and usually do not have complications associated with the conditions. Visit the American College of Medical Geneticists Web site at www.acmg.net to review the ACMG "ACT" sheet for additional information.

Low fetal hemoglobin usually indicates that the infant was older at the time of specimen collection or, in newborns, that the infant was transfused. If the infant was transfused prior to collection of the initial newborn screening specimen, another specimen is required three months after the last transfusion, when the effects of donor red blood cells have dissipated.

• Incidence	Sickle cell trait occurs in one in 10 African Americans. Hemoglobinopathies do occur in all races and ethnic groups. Hemoglobin S is more common among individuals with West African, Middle Eastern, Mediterranean, or Central Indian heritage. Hemoglobin C is more common among individuals with West African heritage. Hemoglobin E is more common among individuals with Mediterranean, African or Southeast Asian heritage. Thalassemias are more common among individuals with Mediterranean, African or Southeast Asian heritage, but do occur worldwide.
• Analyte Measured	Identification of types of hemoglobin present in the sample is performed by high performance liquid chromatography (HPLC). This screening is not quantitative.
• **Reporting Classifications** (Reported by letter to physician of record or hospital contact)	**Follow-up Referrals/Testing** Confirmatory testing, family education and genetic counseling services highly recommended.

Hemoglobin Disease Conditions not usually requiring treatment
FD - Fetal and D hemoglobins – probable hemoglobin D disease*
FE - Fetal and E hemoglobins – probable hemoglobin E disease*
FX - Fetal and anamolous, unidentified hemoglobin – unidentified hemoglobin disorder*

Other Hemoglobinopathies and Traits – Continued

- Reporting Classifications

  Carrier Status
  FAS - Fetal, adult and sickle hemoglobins - probable sickle cell carrier status
  FAC - Fetal, adult hemoglobin C - probable hemoglobin C carrier status
  FAD - Fetal, adult hemoglobin D - probable hemoglobin D carrier status
  FAE - Fetal, adult and E hemoglobins - probable E hemoglobin carrier status

  Other Hemoglobin Results
  LFA - Low fetal and elevated adult hemoglobins - probable transfusion or older infant)

- Feeding Effect — None

- Transfusion Effect — Screening **will be affected** by transfusions. If initial specimen is collected post-transfusion, repeat screening is necessary three months after the last transfusion.

- Timing Effect — No effect in first two weeks of life, although as the infant grows, the amount of fetal hemoglobin usually decreases.

- Specialist — Pediatric hematologist, see Appendix for contact information

- Treatment — Carrier status (hemoglobin traits) usually are considered benign with no treatment necessary

  *Hemoglobin E and Hemoglobin D disease states should be verified by further diagnostic testing. Referral to the Comprehensive Thalassemia Center at Children's Memorial Hospital for diagnostic testing is highly recommended for newborns with results indicating possible thalassemia disorders.

  *Additional diagnostic testing through a pediatric hematologist is recommended for infants with results indicating an unidentified abnormal hemoglobin type.

Comments

Family education, genetic counseling and diagnostic services are available to all families of infants with sickling hemoglobin disorders or traits. The Illinois Department of Public Health, through grants to university-based medical clinics, provides diagnostic and treatment services for infants and children identified with hemoglobin disorders or traits. See Appendix for a listing of pediatric hematology specialists for information about these services. The Sickle Cell Disease Association of Illinois also will provide educational services to families of individuals with sickle cell diseases or carrier status.

Lysosomal Storage Diseases

(Statewide screening of newborns for lysosomal storage disorders is scheduled to begin in 2012.)

Lysosomal storage diseases are inherited metabolic conditions. Each disease is caused by deficiency of a specific enzyme related to the metabolism of lipids or glycogen. Of the lysosomal storage diseases included in the Illinois newborn screening panel, each disease causes accumulation of metabolites, primarily sphingolipids, or in the case of Pompe disease, harmful amounts of glycogen, in the lysosomes of cells. Over time, these accumulated lipids or glycogen deposits cause permanent cellular and tissue damage. Lysosomal disorders affect different organ systems including the brain and nervous system, liver, renal system, heart, skeletal muscles, lungs, spleen and bone marrow. Treatment options vary among the disorders, and are disease specific, but may include enzyme replacement therapy or stem cell transplantation. See the Department's Web site, www.idph.state.il.us, for newborn screening fact sheets with additional information about this disorder and visit the American College of Medical Geneticists Web site at www.acmg.net to review the ACMG "ACT" sheets.

In Illinois, newborn screening includes testing for enzyme activity levels of specific lysososmal storage diseases. Following is a list of the lysosomal storage disorders that may be detected:

Pompe
Fabry
Gaucher
Krabbe
Niemann-Pick

- Incidence — These are rare disorders, actual incidence unknown. Estimated incidence of all lysosomal storage diseases is around one in 40,000 births.

- Analytes Measured in Screening — Enzyme activity quantified by Enzyme activity microfluidic assay in micro-Moles per Liter per hour (μM/L/hour).
  Screening for Krabbe also includes DNA sequencing of the GALC gene.

Enzyme	**Associated Disease**
Alpha-glucosidase (GAA)	Pompe disease
Alpha-galactosidase (GLA)	Fabry disease
Acid beta-glucocerebrosidase (ABG)	Gaucher disease
Galactocerebrosidase (GALC)	Krabbe disease
Acid sphingomyelinase (ASM)	Niemann-Pick disease

Lysosomal Storage Diseases - Continued

• **Reporting Ranges*** Abnormal results reported by phone, and fax to physician of record or hospital contact unless otherwise specified.	**Follow-up Referrals and Testing** All positive screens require immediate referral to Department designated pediatric metabolic specialist for evaluation and diagnostic testing.
• Feeding Effect	None
• Transfusion Effect	Specimen collection prior to transfusion is always recommended. If post-transfusion specimen collection is required, collection is best performed 48 hours post-transfusion.
• Timing Effect	If specimen is collected at less than 24 hours of age, submit second sample during second day of life.
• Specialist	Pediatric metabolic disease specialist (Department/DSCC designated), see Appendix for the contact information.
• Treatment	Treatment is disease specific.
Pompe Disease	Supportive treatment includes physical therapy. Enzyme replacement therapy is available.
Fabry Disease	Not typically treated during early childhood. Enzyme replacement therapy is available.
Gaucher Disease	Supportive, symptomatic care and monitoring by pediatric specialists. Enzyme replacement therapy is available for Gaucher type 1.
Krabbe Disease**	Supportive, symptomatic care and monitoring by pediatric specialists. Some individuals may benefit from stem cell transplant within first 30 days of life.
Niemann-Pick Disease	Supportive, symptomatic care. Bone marrow transplants have been attempted and clinical trials of enzyme replacement therapy are under study.

Comments

*For questions about the most current laboratory reporting values for these disorders, contact the Department's Division of Laboratories at 312-793-4752.

**Positive newborn screens for possible Krabbe disease require immediate referral to pediatric metabolic specialists at children's hospitals with access to pediatric stem cell transplantation teams. See specialist list for Krabbe.

Appendices

Appendix A

Illinois Department of Public Health

Newborn Metabolic Screening

Specimen Collection Card

Form Name	Illinois NBS - 6" Form
Design ID	ILNB20101014006
Version	006
Design Date	10/14/10

Dotted Magenta lines signify perf lines.

Magenta circles signify line holes.

Front of Form (Flap Folded)

All measurements can vary +/- 1/16" (1.6mm)

Glue lines are between the stubs of parts 1, 2, 3, and 5, and in between parts 3 and 4

SEE DIRECTIONS ON REVERSE SIDE

INSTRUCTIONS TO HOSPITAL: Remove this top sheet and give to parent.

Illinois Department of Public Health
Genetics/Newborn Screening Program
535 W. Jefferson, 2nd Floor
Springfield, Illinois 62761
217-785-8101

PARENT INFORMATION SHEET

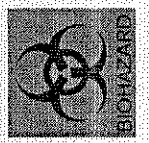
Dear Parent,

A blood sample has been taken from your baby's heel to test for rare, but serious disorders, which can cause mental retardation, poor growth, or death, if not treated. The pamphlet "A Baby's First Step in Life" describes the mandated Illinois newborn screening tests. If you have not received this pamphlet, ask your baby's physician, nurse, or other health care provider for a copy.

It is important that your hospital and your baby's doctor have your correct name and phone number in case your baby needs another test or to be seen by a specialist. If your baby's test is done before he or she is 24 hours old, ask your child's primary care provider to repeat the newborn screening test within 1-2 days.

The Illinois Department of Public Health Newborn Screening Program office will report test results to the hospital, doctor, midwife, or individual who submitted the test. If a repeat test is needed, the Program will notify the health care provider who submitted the sample. If the test is positive, the Program office may also notify a treatment center that may contact you directly. If you have questions, speak to your baby's doctor, contact the Department's Newborn Screening Program, or visit the Department's Web site at www.idph.state.il.us.

AL
EST
EBIRTH
TARY TIME)
TION
(MILITARY TIME)
ARE
ERT
ECTOR



This flap should not touch blood spot when wet.

Total Form Height (All Parts) 6" (152.4mm)

Total Form Length (flap folded) 10" (254.0mm)

ID Biological SYSTEMS ID0214_Rev1 30-Dec-2008

Form Name	Illinois NBS - 6" Form
Design ID	ILNB20101014006
Version	006
Design Date	10/14/10

Dotted Magenta lines signify perf lines.

○ ○ ○

Magenta circles signify line holes.

Face of Part 2 (no copy on back)

ILLINOIS DPH NEWBORN SCREENING
Public Health Laboratory
2121 W. Taylor St., Chicago, IL 60612

DO NOT WRITE IN THIS SPACE

BABY'S INFORMATION
LAST NAME
FIRST NAME
CHART NUMBER

MOTHER'S INFORMATION
SSN ___-___-____
LAST NAME
FIRST NAME
DOB MM/DD/YY
PHONE (___)___-____
ADDRESS
CITY
ST ZIP
COUNTY

GENDER: ☐ M ☐ F ☐ UNK

RACE OF BABY: ☐ White ☐ Black ☐ Native Amer. ☐ UNK ☐ Asian/Pacific Is.

ETHNICITY OF BABY: ☐ Non-Hispanic ☐ Hispanic ☐ UNK

☐ INITIAL ☐ RETEST ☐ HOMEBIRTH

FEEDING: ☐ BREAST ☐ SOY ☐ TPN ☐ CARNITINE ☐ NPO ☐ OTHER

BIRTH DATE MM/DD/YY

TIME OF BIRTH (MILITARY TIME) HH:MM

SPECIMEN DATE OF COLLECTION MM/DD/YY

TIME OF COLLECTION (MILITARY TIME) HH:MM

TRANSFUSED DATE MM/DD/YY

☐ NICU / SPECIAL CARE ☐ ANTIBIOTIC ☐ ERT

BIRTH WEIGHT (grams) | GESTATIONAL AGE | BIRTH # & ORDER ☐ SINGLE BIRTH ☐ MULTIPLE ___ (1,2,3) | COLLECTOR

BABY'S PHYSICIAN NAME & PHONE NUMBER
(___)___-____

HOSP SUBMITTER
CITY ID

ILW0044401x

Total Form Height (All Parts) 6" (152.4mm)

Part 2: 16# White CB, Red 185 ink and black ink face only, red lines screened at 70%, red text screened at 15%, Barcode 3 of 9 with Human Readable with Mod 43 Check Digit
8 1/2" (215.9mm)

ID Biological SYSTEMS ID0214_Rev1 30-Dec-2008

Appendix B

Illinois Department of Public Health

Division of Laboratories

Newborn Screening Specimen Report

Illinois Department of PUBLIC HEALTH

Newborn Screening Laboratory Report

2121 W. Taylor Street · Chicago, Illinois 60612 · (312) 793-4752 · www.idph.state.il.us

Accession #: 2010TEST
Date Received: 01/01/1900
Date Reported:

Illinois Department of Public Health
2121 West Taylor Street
Chicago, IL 60612

Submitter:

Newborn Information:
Name:
BirthDate:
Sex: Weight: g
Race:
BirthOrder: Antibiotics:
NICU: Transfused: N
BabyID: Hospital #:

Physician/Other:

Mother's Information:
Name:
Address:

Phone:

Specimen Information:
Collection Date:
Age At Coll: hrs Gest:
Feeding Type:

Disorder	Analyte	Interpretation	Comment
Biotinidase Deficiency	Biotinidase	Negative	
Congenital Adrenal Hyperplasia	17-OHP	Negative	
Congenital Hypothyroidism*	TSH/T4	Negative	
Cystic Fibrosis*	IRT/DNA	Negative	
Galactosemia	Total GAL/GALT	Negative	
Hemoglobinopathies	Hemoglobin	Negative	
Fatty Acid Disorders	Fatty Acids	Profile Negative	
Amino Acid Disorders	Amino Acids	Profile Negative	
Organic Acid Disorders	Acylcarnitines	Profile Negative	

**Secondary test is only performed when primary test is Borderline or Positive.*

The purpose of the Newborn Screening Program in Illinois is to identify infants at risk for certain congenital conditions and in need of more definitive testing. Abnormal results always require medical evaluation. Results can be affected by: age at time of collection, feeding status, prematurity, low birth weight, transfusion, TPN, illness, medications and collection and handling techniques. As with any laboratory test, false positive or false negative results are possible. A negative screening result does not rule out the possibility of an underlying metabolic/genetic disease. Newborn screening test results are insufficient information on which to base diagnosis or treatment.

Analyte	Results	Interpretation	Normal Range
Biotinidase	Activity Present	Negative	Enzyme Activity Present
TSH	11.1 µU/mL	Negative	<=20 µU/mL
T4	No Results	No Results	Not Applicable
17-OHP	5.8 ng/mL	Negative	***
IRT	13.6 ng/mL	Negative	<170 ng/mL
DNA (CF)	No Results	No Results	Not Applicable
TGAL	2.3 mg/dL	Negative	<6.5 mg/dL
Galt/Uridyltransferase	No Results	No Results	Not Applicable
Hemoglobin	FA	FA	FA Hemoglobin Detected
Arg	5.810 µmol/L	Negative	<40.0 µmol/L
Asa	0.010 µmol/L	Negative	<0.08 µmol/L
Cit	9.810 µmol/L	Negative	>=3.0 and <30.0 µmol/L
Gly	403.930 µmol/L	Negative	<850.0 µmol/L
Leu	90.150 µmol/L	Negative	<260.0 µmol/L
Met	18.390 µmol/L	Negative	<60.0 µmol/L
Orn	40.550 µmol/L	Negative	<150.0 µmol/L
Phe	52.650 µmol/L	Negative	<123.0 µmol/L
Tyr	48.440 µmol/L	Negative	<260.0 µmol/L
Val	114.710 µmol/L	Negative	<255.0 µmol/L
5Oxp	82.92 µmol/L	Negative	<265.0 µmol/L
C0	17.960 µmol/L	Negative	>=7.5 and <66.4 µmol/L
C3	1.170 µmol/L	Negative	<8.0 µmol/L
C3DC (C8OH)	0.050 µmol/L	Negative	<0.15 µmol/L
C4	0.350 µmol/L	Negative	<1.0 µmol/L
C4DC	0.150 µmol/L	Negative	<0.64 µmol/L
C4OH	0.260 µmol/L	Negative	<0.7 µmol/L
C5	0.090 µmol/L	Negative	<0.5 µmol/L
C5:1	0.020 µmol/L	Negative	<0.1 µmol/L
C5DC (C10OH)	0.060 µmol/L	Negative	<0.18 µmol/L
C5OH	0.040 µmol/L	Negative	<0.6 µmol/L
C6	0.0700 µmol/L	Negative	<0.17 µmol/L
C6OH	0.030 µmol/L	Negative	<0.15 µmol/L
C8	0.070 µmol/L	Negative	<0.3 µmol/L
C10	0.150 µmol/L	Negative	<0.4 µmol/L
C10:1	0.080 µmol/L	Negative	<0.2 µmol/L
C10:2	0.010 µmol/L	Negative	<0.1 µmol/L
C12	0.130 µmol/L	Negative	<0.6 µmol/L
C14	0.210 µmol/L	Negative	<0.7 µmol/L
C14:1	0.110 µmol/L	Negative	<0.6 µmol/L
C14:2	0.030 µmol/L	Negative	<0.1 µmol/L
C16	3.270 µmol/L	Negative	<8.0 µmol/L
C16:1OH	0.030 µmol/L	Negative	<0.15 µmol/L
C16OH	0.020 µmol/L	Negative	<0.08 µmol/L
C18	1.110 µmol/L	Negative	<2.2 µmol/L
C18:1	1.120 µmol/L	Negative	<3.0 µmol/L
C18:1OH	0.020 µmol/L	Negative	<0.08 µmol/L
C18OH	0.010 µmol/L	Negative	<0.08 µmol/L

***17-OHP Ranges

Gestation Age and Weight Concentration

<=36 WKS or >36 WKS and <2000 g <55 ng/mL Negative, <80 ng/mL Borderline, >=80 ng/mL Positive

>36 WKS and/or >=2000 g <30 ng/mL Negative, <55 ng/mL Borderline, >=55 ng/mL Positive

Illinois Department of Public Health, Newborn Screening Laboratory, 2121 W. Taylor St., Chicago, IL 60612

Appendix C

Illinois Department of Public Health

Newborn Metabolic Screening

Abnormal Screening Report Sample

Illinois Department of Public Health

NEWBORN SCREENING
OFFICE OF HEALTH PROMOTION
535 W. Jefferson St., 2nd Floor
Springfield, IL 62761
Phone: 217-785-8101
Fax: 217-557-5396

ABNORMAL NEWBORN SCREENING RESULT
Date Reported: 3/8/2011

Sent To:

Newborn Information:	**Laboratory Information:**	**Mother Information:**
Name:	Accession Number:	
A.K.A.:	Sex:	
Date of Birth:	Weight:	
Collection Date:	Transfused:	
Repeat Specimen:	Feeding Type:	
Medical Record #:	Accession Date:	
Facility: HOSPITAL	Gestational Age (weeks):	

ABNORMAL SCREENING RESULT - Tyrosinemia
Tyrosine = 600.805 uM/L (Normal Range: <260.0 uM/L)

POSSIBLE AMINO ACID DISORDER. PLEASE IMMEDIATELY REFER THIS BABY TO A PEDIATRIC METABOLIC SPECIALIST FROM THE ENCLOSED LIST FOR EVALUATION AND DIAGNOSTIC TESTING. The clinician should immediately check on the clinical status of the baby; if left untreated, these conditions may result in significant disability, and ultimately, death.

If you are unable to contact this family, please call (217) 785-8101 and a member of the Newborn Screening Program will assist you.

Resources:
www.acmg.net (ACMG ACT sheets and algorithms)
www.idph.state.il.us (newborn screening information and fact sheets)
www.region4genetics.org (medical home and newborn screening resources)

Appendix D

Illinois Department of Public Health

Division of Specialized Care for Children

Pediatric Subspecialists Available for Consultation

The Illinois Department of Public Health Newborn Screening Program

Medical Specialists for Amino Acid/Organic Acid/Fatty Acid Oxidation Disorders

CHICAGO LOCATIONS

CHILDREN'S MEMORIAL HOSPITAL
(773) 880-4462
2300 Children's Plaza, Box 59
Chicago, IL 60614
Barbara K. Burton, M.D.
Joel Charrow, M.D.

RUSH UNIVERSITY MEDICAL CENTER
(312) 942-6298
1750 W. Harrison St., Rm. 1507, Jelke
Chicago, IL 60612
Paul Wong, M.D., M.Sc.

THE UNIVERSITY OF ILLINOIS AT CHICAGO HOSPITAL
(312) 355-0732
840 S. Wood St., Rm. 1215, M/C 856
Chicago, IL 60612-7311
George Hoganson, M.D.
Allen Horwitz, M.D.

CHICAGO SUBURBAN LOCATIONS

ADVOCATE HOPE CHILDREN'S HOSPITAL
(312) 355-0732
4440 W. 95th St.
Oak Lawn, IL 60453
George Hoganson, M.D.

CHILDREN'S PEDIATRIC SPECIALTY SERVICES
(773) 880-4462
2301 Enterprise Dr.
Westchester, IL 60154
Barbara K. Burton, M.D.

NAPERVILLE CLINIC
(312) 355-0732
1020 E. Ogden Ave., Ste. 302
Naperville, IL 60563
George Hoganson, M.D.

PEORIA LOCATION

UNIVERSITY OF ILLINOIS AT PEORIA
(312) 355-0732
420 N.E. Glen Oak Ave., Ste. 401
Peoria, IL 61603
George Hoganson, M.D.

ROCKFORD LOCATION

ROCKFORD MEMORIAL HOSPITAL
(815) 971-5069
2400 N. Rockton Ave.
Rockford, IL 61103
George Hoganson, M.D.

ST. LOUIS LOCATIONS

CARDINAL GLENNON CHILDREN'S MEDICAL CENTER
(314) 577-5639
1465 S. Grand Blvd.
St. Louis, MO 63104-1095
Stephen Braddock, M.D.
Gary Gottesman, M.D.

ST. LOUIS CHILDREN'S HOSPITAL
(314) 454-6093
660 S. Euclid Ave.
Campus Box 8116
St. Louis, MO 63110
Dorothy K. Grange, M.D.
Marwan Shinawi, M.D.

The Division of Specialized Care for Children (DSCC) will cover diagnostic testing for any infant with a suspect newborn screening test for these disorders regardless of the family's financial eligibility if the testing is ordered by a physician on this list. DSCC requires other third party payers and insurers to be maximized when coverage is available.

November 2010

The Illinois Department of Public Health Newborn Screening Program

Medical Specialists for Biotinidase Deficiency/Galactosemia

CHICAGO LOCATIONS

CHILDREN'S MEMORIAL HOSPITAL
(773) 880-4462
2300 Children's Plaza, Box 59
Chicago, IL 60614
Barbara K. Burton, M.D.
Joel Charrow, M.D.

JOHN A. STROGER JR. HOSPITAL OF COOK COUNTY
(312) 864-4158
1900 W. Polk, Rm. 1159
Chicago, IL 60612
Susan C. Echiverri, M.D.

RUSH UNIVERSITY MEDICAL CENTER
(312) 942-6298
1750 W. Harrison St., Rm. 1507, Jelke
Chicago, IL 60612
Paul Wong, M.D., M.Sc.

THE UNIVERSITY OF CHICAGO HOSPITALS
(773) 834-9110
5841 S. Maryland Ave., Ste. L161
Chicago, IL 60637
Darrel J. Waggoner, M.D.

THE UNIVERSITY OF ILLINOIS AT CHICAGO HOSPITAL
(312) 355-0732
840 S. Wood St., Rm. 1217, M/C 856
Chicago, IL 60612-7311
George Hoganson, M.D.
Allen Horowitz, M.D.

CHICAGO SUBURBAN LOCATIONS

SAINT JOSEPH MEDICAL CENTER
(312) 942-6298
333 N. Madison St.
Joliet, IL 60435
Paul Wong, M.D., M.Sc.

POPLAR CREEK/HOFFMAN ESTATES
(847) 608-7517
1800 McDonough Rd., #203
Hoffman Estates, IL 60192
Paul Wong, M.D., M.Sc.

CHICAGO SUBURBAN LOCATIONS

ADVOCATE MEDICAL CENTERS LUTHERAN GENERAL CHILDREN'S MEDICAL CENTER
(847) 723-7705
1875 Dempster St., Ste. 310
Park Ridge, IL 60068
Carol Booth, M.D.
Debra Rita, M.D.

CHILDREN'S PEDIATRIC SPECIALTY SERVICES
(773) 880-4462
2301 Enterprise Dr.
Westchester, IL 60154
Barbara K. Burton, M.D.

COPLEY MEDICAL CENTER
(312) 942-6298
Fox Valley Medical Associates Bldg.
Rt. 3 at Fox Valley Center
Aurora, IL 60605
Paul Wong, M.D., M.Sc.

NAPERVILLE CLINIC
(312) 355-0732
1020 E. Ogden Ave., Ste. 302
Naperville, IL 60563
George Hoganson, M.D.

LOYOLA UNIVERSITY MEDICAL CENTER
(708) 327-9085
2160 S. 1st Ave.
Maywood, IL 60153
Carolyn H. Jones, M.D.

RIVERSIDE MEDICAL CENTER
(312) 942-6298
350 N. Wall St.
Kankakee, IL 60901
Paul Wong, M.D., M.Sc.

ADVOCATE HOPE CHILDREN'S HOSPITAL
(312) 355-0732
4440 W. 95th St.
Oak Lawn, IL 60453
George Hoganson, M.D.

ST. LOUIS LOCATIONS

CARDINAL GLENNON CHILDREN'S MEDICAL CENTER
(314) 577-5639
1465 S. Grand Blvd.
St. Louis, MO 63104-1095
Stephen Braddock, M.D.
Gary Gottesman, M.D.

ST. LOUIS CHILDREN'S HOSPITAL
(314) 454-6093
660 S. Euclid Ave.
Campus Box 8116
St. Louis, MO 63110
Dorothy K. Grange, M.D.
Marwan Shinawi, M.D.

PEORIA LOCATION

UNIVERSITY OF ILLINOIS AT PEORIA
(312) 355-0732
420 N.E. Glen Oak Ave., Ste. 401
Peoria, IL 61603
George Hoganson, M.D.

ROCKFORD LOCATION

ROCKFORD MEMORIAL HOSPITAL
(815) 971-5069
2400 N. Rockton Ave.
Rockford, IL 61103
George Hoganson, M.D.

SPRINGFIELD LOCATIONS

SIU MEDICAL SCHOOL/SIU PHYSICIANS & SURGEONS
(217) 545-4839
P.O. Box 19658
Springfield, IL 62794
Michael Schneider, M.D.

SANGAMON COUNTY DEPARTMENT OF PUBLIC HEALTH
(312) 355-0732
2833 S. Grand Ave. East
Springfield, IL 62703
George Hoganson, M.D.

The Division of Specialized Care for Children (DSCC) will cover diagnostic testing for any infant with a suspect newborn screening test for these disorders regardless of the family's financial eligibility if the test is orderd by a physician on this list. DSCC requires other third party payers and insurers to be maximized when coverage is available. **November 2010**

The Illinois Department of Public Health Newborn Screening Program

Medical Speciaists for Phenylketonuria

CHICAGO LOCATIONS

CHILDREN'S MEMORIAL HOSPITAL
(773) 880-4462
2300 Children's Plaza, Box 59
Chicago, IL 60614
Barbara K. Burton, M.D.

RUSH UNIVERSITY MEDICAL CENTER
(312) 942-6298
1750 W. Harrison St., Rm. 1507, Jelke
Chicago, IL 60612
Paul Wong, M.D., M.Sc.

THE UNIVERSITY OF ILLINOIS AT CHICAGO HOSPITAL
(312) 355-0732
840 S. Wood St., Rm. 1215, M/C 856
Chicago, IL 60612-7311
George Hoganson, M.D.
Allen Horwitz, M.D.

CHICAGO SUBURBAN LOCATIONS

ADVOCATE HOPE CHILDREN'S HOSPITAL
(312) 355-0732
4440 W. 95th St.
Oak Lawn, IL 60453
George Hoganson, M.D.

CHILDREN'S PEDIATRIC SPECIALTY SERVICES
(773) 880-4462
2301 Enterprise Dr.
Westchester, IL 60154
Barbara K. Burton, M.D.

COPLEY MEDICAL CENTER
(312) 942-6298
Fox Valley Medical Associates Bldg.
Rt. 3 at Fox Valley Center
Aurora, IL 60605
Paul Wong, M.D., M.Sc.

CHICAGO SUB. LOCATIONS (cont.)

NAPERVILLE CLINIC
(312) 355-0732
1020 E. Ogden Ave., Ste. 302
Naperville, IL 60563
George Hoganson, M.D.

RIVERSIDE MEDICAL CENTER
(312) 942-6298
350 N. Wall St.
Kankakee, IL 60901
Specialist: Paul Wong, M.D., M.Sc.

SAINT JOSEPH MEDICAL CENTER
(312) 942-6298
333 N. Madison St.
Joliet, IL 60435
Paul Wong, M.D., M.Sc.

PEORIA LOCATION

UNIVERSITY OF ILLLINOIS AT PEORIA
(312) 355-0732
420 N.E. Glen Oak Ave., Ste. 401
Peoria, IL 61603
George Hoganson, M.D.

ROCKFORD LOCATION

ROCKFORD MEMORIAL HOSPITAL
(815) 971-5069
2400 N. Rockton Ave.
Rockford, IL 61103
George Hoganson, M.D.

ST. LOUIS LOCATIONS

CARDINAL GLENNON CHILDREN'S MEDICAL CENTER
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1465 S. Grand Blvd.
St. Louis, MO 63104-1095
Stephen Braddock, M.D.
Gary Gottesman, M.D.

ST. LOUIS CHILDREN'S HOSPITAL
(314) 454-6093
660 S. Euclid Ave.
Campus Box 8116
St. Louis, MO 63110
Dorothy K. Grange, M.D.
Marwan Shinawi, M.D.

SPRINGFIELD LOCATION

SANGAMON COUNTY DEPARTMENT OF PUBLIC HEALTH
(312) 355-0732
2833 S. Grand Ave. East
Springfield, IL 62703
George Hoganson, M.D.

The Division of Specialized Care for Children (DSCC) will cover diagnostic testing for any infant with a suspect newborn screening test for these disorders regardless of the financial eligibility of the family if tests are ordered by a physician on this list. DSCC requires other third party payers and insurers to be maximized when coverage is available.

November 2010

The Illinois Depatment of Public Health Newborn Screening Program Medical Specialists for Congenital Adrenal Hyperplasia and Congenital Hypothyroidism

CHICAGO LOCATIONS

CHILDREN'S MEMORIAL HOSPITAL
(773) 327-7740
2300 Children's Plaza
Chicago, IL 60614
Reema Habiby, M.D.
Mary Kreiter, M.D.

RUSH UNIVERSITY MEDICAL CENTER
(312) 942-8989
1653 W. Congress Parkway
Chicago, IL 60612
Richard A. Levy, M.D.

THE UNIVERSITY OF CHICAGO HOSPITALS
(773) 795-7993
5841 S. Maryland Ave.
Chicago, IL 60637
Dianne Deplewski, M.D.
Elizabeth Baumann-Littlejohn, M.D.

THE UNIVERSITY OF ILLINOIS AT CHICAGO HOSPITAL
(312) 996-1682 or (312) 996-1795
840 S. Wood St., M/C 856
Chicago, IL 60612-7311
Songya Pang, M.D.
Diego Ize-Ludlow, M.D.

JOHN H. STROGER JR. HOSPITAL OF COOK COUNTY
(312) 864-4172
Division of Genetics
1900 W. Polk St., Rm. 1159
Chicago, IL 60612
Vanessa Davis, M.D.

CHICAGO SUBURBAN LOCATIONS

ADVOCATE HOPE CHILDREN'S HOSPITAL
(708) 684-5670
4440 W. 95th St.
Oak Lawn, IL 60453
Fuad Ziai, M.D.

CHICAGO SUBURBAN LOCATIONS (cont.)

ADVOCATE MEDICAL CENTERS LUTHERAN GENERAL CHILDREN'S MEDICAL CENTER
(847) 723-9330
1775 Dempster St.
Park Ridge, IL 60068
Kanika Ghai, M.D.

CENTER PEDIATRIC SUBSPECIALTIES
(708) 684-5670
16151 Weber Road, Ste. 101
Crest Hill, IL 60435
Fuad Ziai, M.D.

CENTER PEDIATRIC SUBSPECIALTIES
(708) 684-5670
4151 Naperville Road, Ste. 204
Lisle, IL 60532
Fuad Ziai, M.D.

ELMHURST CLINIC
(630) 941-4710
1200 S. York Rd. #2000
Elmhurst, IL 60126-5634
Fuad Ziai, M.D.

ACADEMIC ENDOCRINE METABOLISM AND NUTRITION INC.
(630) 416-4501
2001 Gary Avenue, Ste. 240
Wheaton, IL 60187
W. Patrick Zeller, M.D.

ENH MEDICAL GROUP: OLD ORCHARD
(847)- 663-8508
997 Woods Dr.
Skokie, IL 60077
Stephen C. Duck, M.D.

LOYOLA UNIVERSITY MEDICAL CENTER
(708) 216-8563
2160 S. 1st Ave., Bldg. 105, Rm. 3330
Maywood, IL 60153
Carla Minnuti, M.D.

PEORIA LOCATION

PEDIATRIC SUBSPECIALTY CLINIC
(309) 624-9600
420 N.E. Glen Oak Ave.
Peoria, IL 61603
Sue Ellyn Sauder, M.D.
Joyce E. Wise, M.D.
Maurico Flores, M.D.

ST. LOUIS LOCATIONS

CARDINAL GLENNON CHILDREN'S MEDICAL CENTER
1465 S. Grand Blvd.
St. Louis, MO 63104-1095
Sherida Tollefsen, M.D.
David Paul Dempsher, M.D.
Susan E. Myers, M.D.

ST. LOUIS CHILDREN'S HOSPITAL
(314) 454-6051
#1 Children's Place
Campus Box 8116, 9th floor, NWT
St. Louis, MO 63110
Neil H. White, M.D.
Bess Adkins Marshall, M.D.
Abby Solomon Hollander, M.D.

November 2010

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Illinois Department of Public Health
Newborn Screening Program
Medical Specialists for Cystic Fibrosis

CHICAGO LOCATIONS

CHILDREN'S MEMORIAL HOSPITAL
773-880-4462
2300 Children's Plaza, Box # 43
Chicago, IL 60614
Susanna McColley, M.D.

RUSH UNIVERSITY MEDICAL CENTER
312-563-2270
1725 W. Harrison, Suite 710
Chicago, IL 60612
Girish Sharma, M.D.

UNIVERSITY OF CHICAGO CHILDREN'S HOSPITAL DEPARTMENT OF PEDIATRICS
773-702-6178
5841 S. Maryland Ave., MC 4064
Chicago, IL 60637
Lucille Lester, M.D.

UNIVERSITY OF ILLINOIS AT CHICAGO
312-996-6714
1801 Taylor St. , Clinic 2E
Chicago, IL 60637
Lucy Parks, M.D.

CHICAGO SUBURBAN LOCATIONS

EVANSTON HOSPITAL
847-998-3434
2650 Ridge Ave.
Evanston, IL 60201
Stephen Boas, M.D.

ST. ALEXIUS MEDICAL CENTER
847-998-3434
1555 Barrington Rd.
Hoffman Estates, IL 60169
Stephen Boas, M.D.

CHICAGO SUBURBAN LOCATIONS

ADVOCATE CHRIST HOPE CHILDREN'S HOSPITAL
708-684-5810 or 708-684-5685
4440 W. 95th St.
Oak Lawn, IL 60453
Javed Akhter, M.D.

ADVOCATE LUTHERAN GENERAL CHILDREN'S HOSPITAL
847-723-7705
1675 Dempster St.
Park Ridge, IL 60068
Gabriel Aljadeff, M.D.

PEORIA LOCATION

CHILDREN'S HOSPITAL OF ILLINOIS AT O.S.F. ST. FRANCIS MEDICAL CENTER
309-624-9632 or 309-624-9627
320 E. Armstrong Ave.
Peoria, IL 61603
Jaylayne Lapke, M.D.

CHAMPAIGN LOCATION

CARLE CLINIC ASSOCIATION, DEPARTMENT OF PEDIATRICS
217-383-3100
602 W. University Ave.
Urbana, IL 61801
Donald F. Davison, M.D.

LOYOLA UNIVERSITY MEDICAL CENTER
708-327-9056
2160 S. First Ave.
Maywood, IL 60153

SPRINGFIELD LOCATION

SOUTHERN ILLINOIS SCHOOL OF MEDICINE
217-545-0702
St. John's Pavilion
301 N. 8th St., P.O. Box 19658
Springfield, IL 62794
Mark Johnson, M.D.

ROCKFORD LOCATION

ROCKFORD MEMORIAL HOSPITAL
815-971-5069
Department of Medical Genetics
2400 N. Rockton Ave.
Rockford, IL 61103
David Shoberg, M.D.

ST. LOUIS LOCATIONS

CARDINAL GLENNNON CHILDREN'S HOSPITAL SAINT LOUIS UNIVERSITY SCHOOL OF MEDICINE
314-268-6439
1465 S. Grand Ave.
St. Louis, MO 63104
Blakeslee Noyes, M.D.

WASHINGTON UNIVERSITY SCHOOL OF MEDICINE, DEPARTMENT OF PEDIATRIC ALLERGY AND PULMONARY MEDICINE
314-454-2353
660 S. Euclid Ave., Campus Box 8116
St. Louis, MO 63110
Thomas Ferkol, M.D.

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The Illinois Department of Public Health Newborn Screening Program

Pediatric Hematologists

CHICAGO LOCATIONS

CHILDREN'S MEMORIAL HOSPITAL*
(773) 880-4125
2300 Children's Plaza, #30
Chicago, IL 60614
Alexis Thompson, M.D.

JOHN A. STROGER JR. HOSPITAL OF COOK COUNTY*
(312) 864-4168
1900 W. Polk, Rm. 1136
Chicago, IL 60612
Lily Matthew, M.D.
Lisa Giordano, M.D.
Radhika Peddinpi, M.D.

MT. SINAI HOSPITAL*
(773) 257-6892
2720 W. 15th St., Rm. F444
Chicago, IL 60612
Bruce Sharon, M.D.

RUSH UNIVERSITY MEDICAL CENTER*
(312) 942-3034
1753 W. Congress Parkway
Chicago, IL 60612
Allen Korenblit, M.D., F.A.A.P.
Paul Kent, M.D.

THE UNIVERSITY OF CHICAGO/ LARABIDA CHILDREN'S HOSPITAL SICKLE CELL PROGRAM*
(773) 256-5759 (Ask for Patricia Bailey)
East 65th St @Lake Michigan
Chicago, IL 60649
Uma Subramanian, M.D.

THE UNIVERSITY OF ILLINOIS AT CHICAGO HOSPITAL*
(312) 996-6143
840 S. Wood St., MC 856
Chicago, IL 60612-7324
MaryLou Schmidt, M.D.
Bruce Sharon, M.D.
Ruth Seeler, M.D.

CHICAGO SUBURBAN LOCATIONS

ADVOCATE HOPE CHILDREN'S HOSPITAL*
(708) 684-4094
4440 W. 95th St.
Oak Lawn, IL 60453
Sharad Salvi, M.D.
Jason Canner, M.D.
Ammar Hayani, M.D.

ADVOCATE MEDICAL CENTERS LUTHERAN GENERAL CHILDREN'S MEDICAL CENTER
(847) 318-9330
1675 Dempster St.
Park Ridge, IL 60068
Jong-Hyo Kwon, M.D.
William Goodell, M.D.

LOYOLA UNIVERSITY MEDICAL CENTER
(708) 216-8563
2160 S. 1st Ave.
Maywood, IL 60153
Ricarchito Manera, M.D.
Charles Hemenway, M.D.
Marie-Ellen Sarvida, M.D.

CENTRAL DUPAGE HOSPITAL
(630) 933-6631
25 N. Winfield Rd.
Winfield, IL 60190
Ammar Hayani, M.D.
Jason Canner, M.D.

PEORIA LOCATION

CHILDREN'S HOSPITAL OF ILLINOIS AT O.S.F. ST. FRANCIS MEDICAL CENTER*
(309) 624-4945
530 N.E. Glen Oak Ave.
Peoria, IL 61637
Kay L. Saving, M.D.
Stephen D. Smith, M.D.
Mohamad Al-Rahawan, M.D.

EAST ST. LOUIS LOCATION

SOUTHERN IL HEALTH CARE FOUNDATION MOTHER CHILD CENTER*
(618) 332-27[illegible] (ask for Sickle Cell Nurse)
6000 Bond Avenue
Centreville, IL 62207
William Ferguson, M.D.

ST. LOUIS LOCATIONS

CARDINAL GLENNON CHILDREN'S MEDICAL CENTER*
(314) 577-5600
1465 S. Grand Blvd.
St. Louis, MO 63104-1095
William Ferguson, M.D.
Deepika Bhatla, M.D.
Karen Gauvain, M.D.
Christopher Hugge, M.D.
Brandon Triplett, M.D.

ST. LOUIS CHILDREN'S HOSPITAL*
(314) 454-6018
#1 Children's Place
Campus Box 8116
St. Louis, MO 63110
Elliott Gellman, M.D.
Allison King, M.D.

CHAMPAIGN LOCATION

CARLE CLINIC
(217) 365-6202
1701 W. Curtis Road
Champaign, IL 61822
Mark Musselman, M.D., M.A.

SPRINGFIELD LOCATION

SIU MEDICAL SCHOOL/SIU PHYSICIANS & SURGEONS
(217) 545-7377
415 N. 9th St.
P.O. Box 19678
Springfield, IL 62794-9678
Gregory Brandt, M.D.
Daniel Niebrugge, M.D.

*Centers receiving funding from the Illinois Department of Public Health

November 2010

Illinois Department of Public Health Newborn Screening Program

Medical Specialists for Lysosomal Storage Disorders
Pompe, Fabry, Gaucher and Niemann-Pick Diseases

CHICAGO LOCATIONS

CHILDREN'S MEMORIAL HOSPITAL
(773) 880-4462
2300 Children's Plaza, Box 59
Chicago, IL 60614
Barbara K. Burton, M.D.
Joel Charrow, M.D.

RUSH UNIVERSITY MEDICAL CENTER
(312) 942-6298
1750 W. Harrison St., Rm. 1507, Jelke
Chicago, IL 60612
Paul Wong, M.D., M.Sc.

THE UNIVERSITY OF CHICAGO HOSPITALS
(773) 834-9110
5841 S. Maryland Ave., Ste.L161, M/C 0077
Chicago, IL 60637
Specialist: Darrel J. Waggoner, M.D.

THE UNIVERSITY OF ILLINOIS AT CHICAGO HOSPITAL
(312) 355-0732
840 S. Wood St., Rm. 1215, M/C 856
Chicago, IL 60612-7311
George Hoganson, M.D.

CHICAGO SUBURBAN LOCATIONS

ADVOCATE HOPE CHILDREN'S HOSPITAL
(312) 355-0732
4440 W. 95th St.
Oak Lawn, IL 60453
George Hoganson, M.D.

CHILDREN'S PEDIATRIC SPECIALTY SERVICES
(773) 880-4462
2301 Enterprise Dr.
Westchester, IL 60154
Barbara K. Burton, M.D.

NAPERVILLE CLINIC
(312) 355-0732
1020 E. Ogden Ave., Ste. 302
Naperville, IL 60563
George Hoganson, M.D.

PEORIA LOCATION

UNIVERSITY OF ILLINOIS AT PEORIA
(312) 355-0732
420 N.E. Glen Oak Ave., Ste. 401
Peoria, IL 61603
George Hoganson, M.D.

ROCKFORD LOCATION

ROCKFORD MEMORIAL HOSPITAL
(815) 971-5069
2400 N. Rockton Ave.
Rockford, IL 61103
George Hoganson, M.D.

ST. LOUIS LOCATIONS

CARDINAL GLENNON CHILDREN'S MEDICAL CENTER
(314) 577-5639
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St. Louis, MO 63104-1095
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Gary Gottesman, M.D.

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Campus Box 8116
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November 2010

Illinois Department of Public Health Newborn Screening Program

Medical Specialists for Lysosomal Storage Disorders

Krabbe Disease

CHICAGO LOCATIONS

CHILDREN'S MEMORIAL HOSPITAL
(773) 880-4462
2300 Children's Plaza, Box 59
Chicago, IL 60614
Barbara K. Burton, M.D.
Joel Charrow, M.D.

THE UNIVERSITY OF CHICAGO HOSPITALS
(773) 834-9110
5841 S. Maryland Ave., Ste.L161, M/C 0077
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Darrel J. Waggoner, M.D.

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660 S. Euclid Ave.
Campus Box 8116
St. Louis, MO 63110
Dorothy K. Grange, M.D.
Marwan Shinawi, M.D.

CARDINAL GLENNON CHILDREN'S MEDICAL CENTER
(314) 577-5639
1465 S. Grand Blvd.
St. Louis, MO 63104-1095
Stephen Braddock, M.D.
Gary Gottesman, M.D.

November 2010

Appendix E

Illinois Local Public Health Department Contacts

ILLINOIS DEPARTMENT OF PUBLIC HEALTH
LOCAL HEALTH DEPARTMENTS
PROVIDING NEWBORN SCREENING FOLLOW-UP SERVICES

Adams County Health Department Candee Musgrove, R.N. 330 Vermont St. Qunicy, IL 62301 Phone: 217.222.8440 ext 121 Fax: 217.222.8508	**Bond County Health Department** Judi Markell, RN, BSN MCH Coordinator 503 S. Prairie Greenville, IL 62246 Phone: 618.664.1442, x 136 Fax: 618.664.1744	**Boone County Health Department** Chris McKibben, RN 1204 Logan Avenue Belvidere, IL 61008-4031 Phone: 815.544.2951 Fax: 815.544.2050
Brown County Health Department Nancy Kropp, R.N. 111 W. Washington Mount Sterling, IL 62353 Phone: 217.773.2714 Fax: 217.773.2425	**Bureau County Health Department** Deb Piper, R.N. 526 Bureau Valley Pkwy., Perry Plaza Princeton, IL 61356 Phone: 815.872.5091 Fax: 815.872-5092	**Calhoun County Health Department** Sandy Teichmann, R.N. P. O. Box 158 Hardin, IL 62047 Phone: 618.576.2428 Fax: 618.576.9808
Carroll County Health Department Sally Marken, R.N. FCM Coordinator/APORS 822 S. Mill Street Mt. Carroll, IL 61053 Phone: 815.244.8855 Fax: 815.244.5010	**Cass County Health Department** Jamie Epping, RN, MCH Director 331 S. Main Street Virginia, IL 62691 Phone: 217.452.2182 Fax: 217.452.7245	**Champaign-Urbana Public Health District** Cathy Ito, R.N., MCH Division 201 W. Kenyon Road Champaign, IL 61820 Phone: 217.531.4311 Fax: 217.352.0126 Andrea Taylor, RN Amy Walker, RN – followup for SIDS Phone : 531-4283 Fax : 531-4297
Chicago Department of Public Health (covering City of Chicago) Cecelia Libunao, RN Central Contact DePaul University Center 333 S. State St., Second Floor Chicago, IL 60604 Phone: 312-747-9690 Fax: 312-747-9694	**Christian County Health Department** Tracie Riggs, R.N. MCH Coordinator 902 W. Springfield Road Taylorville, IL 62568 Phone: 217.824.4113x114 Fax: 217.824.4380	**Clark County Health Department** Barbara Reedy, R.N. 1001 North York Street Martinsville, IL 62442 Phone: 217.382.4207 Fax: 217.382.4226
Clay County Health Department Nancy Wiley, R.N. P. O. Box 579, 601 E. 12th Street Flora, IL 62839 Phone: 618.662.4406 Fax: 618.662.2801	**Clinton County Health Department** Janice Albers, R.N. 930-A Fairfax Street Carlyle, IL 62231 Phone: 618.594.2723 Fax: 618.594.3225	**Coles County Health Department** **Jill Temple, R.N.** P. O. Box 1064, 825 18th St. Charleston, IL 61920-9391 Phone: 217.348.0530 Fax: 217.348.5322
Cook County Department of Public Health Jeannie Taverne, R.N. 1701 S. First Ave., 1st floor Maywood, IL 60153 Phone: 708.786.4054 Fax: 708-786-4001	**(Cook County con't)** **Evanston Health Department** **Will be covered by Cook County as of 7/1/07**	**(Cook County Con't)** **Oak Park Health Department** Catherine Amato, R.N.B.S.N. 1 Village Hall Plaza Oak Park, IL 60302 Phone: 708.358.5428 Fax: 708.358.5552

(Cook County Con't) **Skokie Health Department** Susan A Reisberg, R.N. 5127 Oakton St., P. O. Box 309 Skokie, IL 60077 Phone: 847.933.8252 x 4237 Fax: 847.673.8606	**(Cook County cont)** **Stickney Township Public Health District** Pat Englehart, R.N. 5635 State Road Burbank, IL 60459 Phone: 708.424-9200 x 36 Fax: 708-499-5427	**Crawford County Health Department** Darla Tracy, Public Health Adm. 202 N. Bline Blvd. Robinson, IL 62454 Phone: 618-544-8798 Fax: 618-544-9398
Cumberland County Health Department Jessica Ryder, R.N. MCH Coordinator P. O. Box 130, N.E. Corner of Square Toeldo, IL 62468 Phone: 217-849-3211 Fax: 217-849-3121	**DeKalb County Health Department** Kay Chase, R.N. FCM/WIC Program Coordinator 2550 North Annie GliddenRoad DeKalb, IL 60115 Phone: 815.748-2438 Fax: 815.748.2447	**DeWitt-Piatt Bi-County Health Department** Ann Barnett, R.N., B.S.N. DeWitt-Piatt Bi-County Health Department P. O. Box 518, 910 Route 54 East Clinton, IL 61727 Phone: 217.935.3427 Fax: 217.935.4037
Douglas County Health Department Susan Hays, R.N. Director of Nursing Douglas County Health Department 1250 E. U.S. Highway 36 Tuscola, IL 61953 Phone: 217.253.4137 Fax: 217.253.3421.	**DuPage County Health Department** Doris Feery R.N. 111 N. County Farm Road Wheaton, IL. 60187 630-682-7979 Ext. 7128 Fax: 630-462-9085	**East Side Health District (ESHD)** **(Portions of St. Clair County)** Lynn Shelton, R.N. 636 N. 20th St. East St. Louis, IL 62206 Phone: 618.8740.4713 x 239 Fax: 618.874.4737 **(**E. St. Louis, Washington Park, Fairmount City, SW area of Caseyville, Cahokia, Alorton, Centreville, Sauget, Brooklyn, Lovejoy)
Edgar County Health Department Carol Cline 502 Shaw Avenue Paris, IL 61944 Phone : 217.465.2212, x 236 Fax: 217.465.1121	**Effingham County Health Department** Shawn Bourland, R.N. P. O. Box 685, 901 W. Virginia Effingham, IL 62401 Phone: 217.342.9237 Fax: 217.342-9324	**Egyptian County Health Department** **(Gallatin, Saline & White Counties)** Casey Carlile, R.N. 1412 U.S. 45 North Eldorado, IL 62930-9324 Phone : 618.273.3326 Fax : 618.273.2808
Evanston Health Department **See Cook County**	**Fayette County Health Department** Becky Pryor (Thompson), R.N. 416 W. Edwards St. Vandalia, IL 62471 Phone : 618.283.1044 Fax : 618.283.5038	**Ford-Iroquois Public Health Department** Marilyn Schaumburg, R.N. Director of Nursing 114 N. Third Street Watseka, IL 60970 Phone: 815.432.2483x127 Fax: 815.432.2198
Franklin-Williamson Bi-County Health Department Lisa Sorenson, R.N. Williamson Co. Airport, 120 Express Drive Marion, IL 62959-9808 Phone: 618.993.8111 x 226 Fax: 618.993.6455	**Fulton County Health Department** Louise Hiett, RN, BSN Maternal Child Health Programs Manager 700 E Oak Canton, IL 61520 Phone : 309.647.1134 x 271 Fax: 309.647.9545	**Greene County Health Department** Ruth Ann Flowers, R.N., B.S., Public Health Administrator 310 Fifth St. Carrollton, IL 62016 Phone: 217.942.6961or 6962 Fax: 217.942.3904

Grundy County Health Department Kay Lynn Shumaker, R.N. Director of Nursing 1320 Union St. Morris, IL 60450 Phone: 815.941.3117 Fax: 815.941.2389	**Hamilton County Health Department** LaDonna Lasater, R.N. County Courthouse, Room 5 McLeansboro, IL 62859 Phone: 618.643.3522 Fax: 618.643.2390	**Hancock County Health Department** Vicki Hendrickson, R.N./Jennifer Sherman, R.N. P. O. Box 357, 671 Wabash Carthage, IL 62321 Phone: 217.357.2171 Fax: 217.357.3562
Henderson County Health Department (Also covers Warren County) Mary Lynne Haase, R.N., Family Case Mgt P. O. Box 220 Gladstone, IL 61437-0220 Phone : 309.627.2812 Fax : 309.627.2793	**Henry County Health Department (also covers Stark County)** Sandy Sommer, R.N. 4424 U.S. Highway 34 Kewanee, IL 61433 Phone: 309.852.0197 Fax: 309.852.0595	**Jackson County Health Department** Marilyn Twitty, RN, Genetic Coordinator 618-684-3143x111 Christy Guy, RN (For SIDS cases) 415 Health Department Road P. O. Box 307 Murphysboro, IL 62966 Phone: 618684.3143 x 145 Fax: 618.687.1255
Jasper County Health Department (also covers Richland County) Deborah Riddle, R.N. 106 Edwards St. Newton, IL 62448 Phone: 618.783.4436 Fax: 618.783.4146	**Jefferson County Health Department** Becky Brooks, R.N., Director of Nurses #1 Doctors Park Road, Suite F Mt. Vernon, IL 62864-6213 Phone: 618.244.7134 Fax: 618.244.2640	**Jersey County Health Department** Geri Daniels, R.N., B.S.N. 1307 State Highway 109 Jerseyville, IL 62052 Phone: 618.498.9565 Fax: 618.498.6291
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LaSalle County Health Dept Colleen Gibson, Genetics Coordinator 717 Etna Road Ottawa, IL 61350 Phone: 815.433.3366 Fax: 815.433.9522 SIDS: Cathy Larsen, R.N	**Lawrence County Health Department** Phyllis Wells, R.N., M.S.N.,C.N.S. R.R. #3, Box 414 POB 516 Lawrenceville, IL 62439 Phone: 618.943.3302 Cell : 618-943-9995 Fax: 618.943.7589	**Lee County Health Department** Kathy Schutz, R.N. 309 S. Galena Avenue, Suite 100 Dixon, IL 61021-9185 Phone : 815.284.3371 x 116 Fax : 815.288.1811

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Macoupin County Health Department Karen Hazzard, R.N. 805 N. Broad St. Carlinville, IL 62626 Phone: 217.854.3223, x 234 Fax: 217.854.3225	**Madison County Health Dept.** Becci Weirich, R.N. 101 E. Edwardsville Wood River, IL 62095 Phone: 618.296.6062 Fax: 618.251.9482	**Marion County Health Department** Dena Kemp., R.N. 1013 N. Poplar St Centralia, IL 62801 Phone: 618-532-6518 x 2227 Fax : 618.532.6543
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Rock Island County Health Dept. Franki Cunningham, R.N., B.S.N. Director of Family Health 2112 Twenty-Fifth St. Rock Island, IL 61201 Phone: 309.558.2850 Fax : 309-794-7091	**St. Clair County Health Department** Karoline Stock, R.N., Case Management Supervisor #19 Public Square, Suite 150 Belleville, IL 62220-1624 Phone : 618.233.6170 x 4483 Fax : 618.236.0821 (Caseyville, Dupo, Collinsville and St. Clair County excludes Canteen, Centreville, E. St. Louis, Stites, Cahokia Washington Park, Alorton) –See ESHD	**Sangamon County Health Department** Stacy Darland, R.N. 2501 N. Dirksen Parkway Springfield, IL 62702 Phone: 217.535.3100 x3794 Fax: 217.535.3104
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Stephenson County Health Dept. Julia Marynus, R.N. Case Management Coordinator 10 W. Linden Street Freeport, IL 61032 Phone: 815.235.8271 x 394 Fax : 815.232.7160	**Tazewell County Health Department** Pam Bowen, R.N., Supervisor, Family Case Management 21306 Illinois Route 9 Tremont, IL 61568-9252 Phone: 309.477.2223 x 249 Fax: 309.925.4381	**Vermilion County Health Department** Pam Hull, R.N. 200 S. College Danville, IL 61832 Phone: 217.431.2662 Fax : 217.431.7485 NBS & SIDS cases will be handled by Champaign county for now 5/26/10

Wabash County Health Department (also covers Edwards County) Kendra Grounds, R.N. 130 W. Seventh St. Mount Carmel, IL 62863 Phone: 618.263.3873 Fax: 618.262.4215	**Warren County covered by Stark County**	**Washington County Health Department** Joyce Carson, R.N. 177 S. Washington St. Nashville, IL 62263 Phone: 618.327.3644 Fax: 618.327.4229
Wayne County Health Department Pat Weber, R.N., Coordinator Family Case Management P. O. Box 645, 405 N. Basin Road Fairfield, IL 62837 Phone: 618.842.5166 Fax: 618.842.3305	**Whiteside County Health Department** Pam VanderVinne, R.N. 1300 W 2nd Street Rock Falls, IL 61071 Phone: 815.626.2230 x 313 Fax: 815.626.2231	**Will County Health Department** Sharon Wesel, R.N. 323 Quandrangle Dr. Bolingbrook, IL 60440 Phone: 630.679.7001 Fax: 630-679-7015
Williamson **Covered by Franklin-Williamson**	**Winnebago County Health Department** Carol Deutsh-Schmidt/Gen Coord Phone: 815-720-4319 SIDS: Cathie Heilman, MSW/SIDS 401 Division St. Rockford, IL 61104 Phone: 815.720.4320 Fax: 815.962.4203	**Woodford County Health Department** Linda McKeown, R.N. 109 S. Major Eureka, IL 61530 Phone: 309.467.3064 Fax : 309.467.5104

November 2010

Appendix F

Illinois Department of Public Health Newborn Screening Laboratory

Tandem Mass Spectrometry

Acylcarnitine/Amino Acid Analytes and Associated Disorders

Tandem Mass Spectrometry
Newborn Screening

Tandem mass spectrometry (MS/MS) analysis detects amino acids and acylcarnitines in dried blood newborn screening samples, and the presence of these analytes may indicate certain amino acid, organic acid and fatty acid oxidation disorders. The following is a summary of the disorders that may be associated with elevations of certain amino acids and acylcarnitines.

Amino acid disorders*	**Analytes (Amino acids)****
Phenylketonuria (PKU) or Hyperphenylalaninemia	Phenylalanine
Maple syrup urine disease (MSUD)	Leucine/Isoleucine, Valine
Homocystinuria (cystathionine synthase deficiency) or Hypermethioninemia	Methionine
Tyrosinemia, type 1 and possibly type II or III	Tyrosine - elevations may not be detectable on filter paper in first days of life
5-oxoprolinuria (glutathione synthetase deficiency)*	5-oxoproline
Urea cycle disorders*	
Citrullinemia	Citrulline
Argininosuccinic aciduria (ASA)	Citrulline, Argininosuccinic acid
Argininemia*	Arginine
Fatty acid oxidation disorders*	**Analytes (Acylcarnitines)****
Short chain acyl-CoA dehydrogenase deficiency (SCAD)	**C4**
Isobutyryl-CoA dehydrogenase deficiency (IBCD)	**C4**
Glutaric aciduria, type 2 (GAII) or Multiple acyl-CoA dehydrogenase deficiency (MADD)	**C4, C5, C8:1**, C8, C12, C14, C16, C5-DC
Medium/Short chain L-3-hydroxyacyl-CoA dehydrogenase deficiency (M/SCHAD)*	**C4-OH**
Medium chain acyl-CoA dehydrogenase deficiency (MCAD)	C6, **C8**, C10, C10:1
Long chain 3 hydroxyacyl-CoA dehydrogenase def. (LCHAD)	**C16-OH**, **C18:1-OH**
Trifunctional protein deficiency (TFPD)*	**C16-OH**, **C18:1-OH**
Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)	**C14:1**, C14, C16
Carnitine palmitoyl transferase deficiency, type 2 (CPTII)*	**C16**, **C18:1**, **C18**
Carnitine palmitoyl transferase deficiency, type 1A (CPT1A)*	**C0 elevated,** low C16, C18
Carnitine/acylcarnitine translocase deficiency (CACT)*	**C16**, **C18:1**, **C18**
Carnitine uptake defect (CUD)	**Low C0** - may not be detected in first few days of life
Organic acid disorders*	**Analytes (Acylcarnitines)****
Propionic acidemia (PA)*	**C3**
Methylmalonic acidemia (MMA)*	**C3**
Malonic aciduria (MA)*	**C3-DC**
Multiple carboxylase deficiency (MCD)	**C5-OH**
3-hydroxy 3-methylglutaryl-CoA lyase deficiency (3HMG)	**C5-OH**
3-methylcrotonyl-CoA carboxylase deficiency (3MCC)	**C5-OH**
3-methylglutaconic aciduria (3MGA)	**C5-OH**
2-methylbutyryl-CoA dehydrogenase deficiency (2MBD)	**C5**
Isovaleric acidemia (IVA)	**C5**
Glutaric acidemia, type 1 (GAI)	**C5-DC**
Beta-ketothiolase deficiency (BKT)*	**C5:1**, C5-OH

Notes: *Some forms (genotypes) of these disorders may not be detected by neonatal screening, may not be detected in a newborn dried blood spot, or are extremely rare (1: >250,000).

Primary MS/MS analyte(s) written in **bold type.

MS/MS analytes are measured in micromoles per liter (uM/L).

For substituted carnitines, a notation of (Cx) is used, in which (x) denotes the number of carbons in the fatty acid radical.

Hydroxylation is designated by (-OH), dicarboxylic acids are designated by (-DC), and unsaturation of the fatty acid chain is designated by (:1).

Printed by Authority of the State of Illinois
P.O.#3711742 2M 12/10