State of Illinois
Rod R. Blagojevich, Governor

Department of Public Health
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Newborn Screening Practitioner's Manual

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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 proven 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 most 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 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 (IDPH) 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 IDPH 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 B.

Overview

The Phenylketonuria Testing 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 Phenylketonuria Testing Act and the newborn screening administrative rules may be viewed at the IDPH Web site, http://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 or mail service to

the IDPH Newborn Screening Laboratory in Chicago for testing. When testing is completed, a report of all test results is issued by the IDPH Newborn Screening Laboratory to the specimen submitter (usually the birth center). Birth centers are expected to retain the 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, all abnormal, unsatisfactory and invalid 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 the baby's primary care provider of the abnormal test results, and to facilitate any recommended follow-up activities, including 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 IDPH 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 serious abnormal results requiring immediate referral to a pediatric medical specialist, a list of IDPH 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), http://www.acmg.net, provides detailed action plans for follow-up of each suspected newborn screening disorder. The University of Illinois at Chicago, Division of Specialized Care for Children (DSCC), http://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, http://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 sickling hemoglobinopathies

Amino Acid Disorders
- Homocystinuria (HCU)/Hypermethioninemia
- Maple syrup urine disease (MSUD)
- Phenylketonuria (PKU)/Hyperphenylalaninemia
- Tyrosinemia, type 1 and possibly type 2 or type 3 - levels may not be sufficiently elevated for detection
- 5-Oxoprolinuria (5OXP) - may not be reliably detected in first few days of life

Urea Cycle Disorders
- Argininemia - extremely rare
- Argininosuccinic aciduria (argininosuccinic acid synthetase deficiency - AS)
- Citrullinemia (argininosuccinic acid lyase deficiency - AL)

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 (MCAD)
- Medium/Short chain L-3-hydroxyacyl-CoA dehydrogenase deficiency (M/SCHAD)
- Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
- Short chain acyl-CoA dehydrogenase deficiency (SCAD)
- Trifunctional protein deficiency (TFPD)
- Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)

Other Disorders
- Biotinidase deficiency
- Galactosemia (Classical)
- Cystic fibrosis (CF)

Contact Information

Newborn Screening Follow-up Program
Illinois Department of Public Health
Genetics/Newborn Screening Program
500 E. Monroe St., First Floor
Springfield, IL 62701
Phone 217-785-8101
FAX 217-557-5396
TTY(hearing impaired use only) 800-547-0466

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
IDPH Genetics/Newborn Screening Program
http://www.idph.state.il.us/HealthWellness/genetics.htm

IDPH Newborn Screening Laboratory
http://www.idph.state.il.us/a-zlist.htm#N

****Note:* *Newborn Screening Shipping Labels***
For courier deliveries through the United States Parcel Services (UPS) or other courier services, shipping labels should be addressed to:
Illinois Department of Public Health
Newborn Screening Laboratory
2121 W. Taylor St.
Chicago, IL 60612

For mail deliveries through the United States Postal Service (USPS), shipping labels should be addressed to:
Illinois Department of Public Health
Newborn Screening Laboratory
P.O. Box 12279
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* brochure is available through IDPH Genetics/Newborn Screening Program. Call 217-785-8101 or fax 217-557-5396 to order copies of this brochure.
  - Documentation that a newborn screening specimen was collected and a copy of the IDPH 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, http://medhomes.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 IDPH 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 IDPH 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.** IDPH highly recommends that the physician's office contact the medical specialist and provide the screening results.

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 should 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 IDPH Newborn Screening Laboratory. The physician should obtain IDPH 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 IDPH. Specimens should be collected and submitted as soon as possible after 24 hours of age. Newborn screening of infants over 6 months of age is not recommended as IDPH normal values are based on normal analytes, distributions and controls for newborns. In these cases, consult with a pediatric sub-specialist is recommended.

- **Infants born in Illinois whose mothers reside in another state must have a specimen sent to the IDPH Newborn Screening Laboratory.** Illinois provides screening for all 29 disorders currently recommended by the American College of Medical Geneticists and the 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 IDPH filter paper specimen collection forms are accepted by the IDPH Newborn Screening Laboratory.** See Appendix A to view a copy of the IDPH collection form.

- **Newborn screening filter paper specimen collection forms and mailing envelopes may be requested by contacting IDPH Newborn Screening Laboratory 312-793-4752 (phone) or 312-793-1054 (fax).**

- **Filter paper specimen cards should be stored in the original wrapping and stacked in a vertical position to avoid compression of the filter paper.** When properly stored in a cool, dry place, specimen cards have a shelf life of two years. If in doubt about the age of the cards, please contact the IDPH Newborn Screening Laboratory to verify age by lot number, and re-order cards if necessary.

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

- **Specimen cards are considered legal documents, and accurate personal health information is crucial to valid and reliable testing.** This information also may be vital to the physician for use in locating the infant and contacting the parents, should abnormalities be detected in the blood sample.

Timing of Specimen Collection

- **The ideal time for newborn screening specimen collection from healthy**

newborns is 24 hours after birth. Specimens should <u>not</u> be collected earlier than 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 within the second day of life.*
  - **Transfers:** If the infant requires transfer to another facility, if at all possible, specimen should be collected prior to transfer, regardless of infant's age. If a specimen cannot be collected prior to transfer, the transferring facility is responsible for informing the admitting facility of the need for specimen collection.
  - **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, <u>repeat specimen collection is necessary as soon as possible during the second day of life</u>.***

Note: This is a new requirement due to changes in the Newborn Screening Administrative Rule; visit the IDPH Web site, www.idph.state.il.us, to view the most current Administrative Rule.

<u>Special Considerations</u>

- **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.

Note: This is a new requirement due to changes in the Newborn Screening Administrative Rule; visit the IDPH Web site, www.idph.state.il.us, to view the most current Administrative Rule.*

- **Infants receiving special feedings:** Infants requiring soy formula, hyperalimentation or total parenteral nutrition (TPN), and those not yet receiving milk (galactose) feedings 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 IDPH laboratory staff to assure reliable testing.
  - Soy formula or lack of milk feeding **<u>will affect</u>** screening for galactosemia.
  - Hyperalimentation and TPN **<u>may affect</u>** 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.
    - If indicated by the feeding type information provided, IDPH laboratory staff will perform additional testing, the galactose-1-phosphate uridyl transferase (GALT) enzyme assay to screen for classical galactosemia on specimens from infants receiving soy formula, and those infants who have not yet received an oral feeding (NPO), when the specimen card is marked accordingly.

- **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 specimen 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 <u>last transfusion date prior to the specimen collection</u> on the filter paper collection form.
    - Transfusions **<u>will invalidate</u>** screening for classical galactosemia.
    - Transfusions **<u>will invalidate</u>** screening for biotinidase deficiency.
    - Transfusions **<u>will invalidate</u>** screening for hemoglobinopathies.
    - Specimen collection immediately after transfusion **<u>will affect</u>** all newborn screening results. If a post-transfusion specimen is necessary, collection should be delayed for 48 hours post-transfusion.
    - **If the infant's <u>initial specimen</u> 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.***

****Note: This is a new requirement due to changes in the Newborn Screening Administrative Rule; visit the IDPH Web site, www.idph.state.il.us, to view the most current Administrative Rule.***

<u>Tips for specimen collection</u>

- **Copies of the Whatman newborn screening specimen collection posters "Neonatal Screening: Blood Specimen Collection and Handling Procedure" and "Simple Spot Check"** are available for download and printing at the Whatman company Web site, www.whatman.com, see "Neonatal Screen" and "Spot Check." These posters provide an excellent training resource.

- **IDPH Genetics/Newborn Screening Program (217-785-8101) also provides (on loan) the Clinical Laboratory Standards Institute (CLSI) document, "Blood Collection on Filter Paper for Neonatal Screening Programs; Approved Standard"(LA4-A4).**
- **A small number of CLSI neonatal blood collection videos, "Newborn Screening - The First Step," are 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 CLSI.**
  - If heel stick is not a viable option, see CLSI recommended procedures for specimen collection, Approved Standard LA4-4A.
  - 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 IDPH or CLSI.**
  - Heparinized capillary tubes should only be used when heel stick or collection by syringe is not possible.
  - Extreme caution in applying blood samples from capillary tubes is required to avoid damage to the filter paper and allow uniform application of the sample to avoid "layering."
  - 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 IDPH when results on 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" on page 8.

- **Submitters should mark the retest specimen 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 and the 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 are air dried for a minimum of three hours and are**

submitted <u>within 24 hours of collection</u>, using the courier service designated by IDPH or United States Postal Services first class mail.
For details about the latest recommendations for specimen submission contact:

Illinois Department of Public Health
Newborn Screening Laboratory
2121 W. Taylor St.
Chicago, IL 60612
312-793-4752

- **Currently, all Illinois birthing hospitals may utilize a IDPH 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 or the IDPH laboratory 312-793-4752 for more information about this service.

<u>Timeliness</u>

- **Specimens should be submitted to IDPH Division of Laboratories on a <u>daily basis</u>, during regular business days, Monday through Friday.** Saturday UPS courier service pickup of specimens for next business day delivery is also available to birthing hospitals. Batching of specimens from multiple collection days is <u>unacceptable</u>.

- **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.

- **Tracking delivery of specimens to the IDPH laboratory by the courier service is the responsibility of birthing hospitals utilizing this service.** 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 birthing hospital and health care facility submitting specimens to IDPH are reviewed every three months and copies are available through the Newborn Screening Program.**

Reporting of Screening Results

Normal Results

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

Abnormal Results

- **Newborn screening is not diagnostic.** Abnormal screening results are designated by IDPH 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 of all results is also sent to the submitting facility or submitting agent by IDPH Newborn Screening Laboratory for every specimen received.

- **Suspect borderline abnormal:** Indicates that the screening was slightly abnormal and that 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 of all results also is sent to the submitting facility or submitting agent by IDPH Newborn Screening Laboratory for every specimen received.

Unsatisfactory Specimens

- **Indicates that the specimen was improperly collected, handled or submitted, as determined by IDPH Division of Laboratories.** Specimens must be of good quality to assure reliable, valid newborn screening; unsatisfactory specimens require collection and submission of a new specimen 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 IDPH laboratory report sent to the submitting facility or submitting agent.

- **Unsatisfactory specimens delay newborn screening and cause frustration and added expenses for families and hospital staff.**

- **Reports on the number and type of unsatisfactory specimens submitted by each birthing hospital and health care provider are reviewed every three months; copies are sent to the facilities by the Newborn Screening Program.**

Referrals to Pediatric Medical Specialists and Other Agencies

Pediatric Medical Specialists

- **Lists of IDPH 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 B 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 IDPH recommendations offered 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 B for a listing of medical centers providing pediatric hematology services, or call 217-785-8101 for a listing of IDPH designated pediatric hematologists.
  - In addition, by Memorandum of Understanding, IDPH 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 any infant with certain abnormal metabolic newborn screening results, provided these services are coordinated by specialists jointly approved by IDPH and 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 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.

- IDPH program staff and 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.
- An updated 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 IDPH Web site, http: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 http://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 IDPH 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 (AL), Argininosuccinic aciduria (AS) and Argininemia

• **Incidence**	**<u>PKU</u> and hyperphenylalaninemia - one in 10,000 births** **<u>MSUD</u> - one in 200,000 births** **<u>Tyrosinemia</u> - one in >500,000 births (one in 12,500 births among French Canadian populations)** **<u>Homocystinuria</u> - one in 200,000 births** **5-oxoprolinuria - extremely rare, actual incidence unknown** **<u>Urea Cycle Disorders</u> - one in 200,000 to 300,000 births**
• **Analytes Measured in Screening**	**Measurement of specific analytes by tandem mass spectrometry (MS/MS) in micro-Moles/Liter (uM/L)**

<u>Amino Acid/Urea Cycle Disorders - Continued</u>

• **Analytes Measured in Screening**	**<u>Possible Disorder</u>**
Phenylalanine	**Phenylketonuria (PKU)**
Tyrosine	**Tyrosinemia**

Methionine	**Homocystinuria (HCU)**
(Iso)leucine and Valine	**Maple syrup urine disease (MSUD)**
Citrulline	**Citrullinemia (AL) or Argininosuccinic aciduria (AS)**
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.

<u>Presumptive Positive</u>	**Immediate referral to pediatric metabolic disease specialist**
<u>Suspect Borderline Abnormal</u>	**Medical evaluation and repeat newborn screening specimen within one to two days; mark retest box on specimen card.**
<u>Suspect Amino Acid Abnormal due to TPN</u> (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.**

- **Feeding Effect** — **Hyperalimentation and TPN may affect results. Unless otherwise instructed, repeat specimens are best collected 48 hours after TPN is discontinued.**

<u>Amino Acid/Urea Cycle Disorders - Continued</u>

- **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**	**Metabolic disease specialist (IDPH/DSCC designated), see Appendix B 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. IDPH provides special medically necessary formula without charge to Illinois residents who are under comprehensive medical management provided by a metabolic disease specialist designated by IDPH.**

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

****For questions about the most current laboratory reporting values for these disorders, contact IDPH 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 IDPH Web site, http: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 http://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/testing for abnormal test results**
Biotinidase activity absent on initial specimen	**Medical evaluation and collect repeat newborn screen within one to two days; mark retest box on specimen card.**
Biotinidase activity absent on second specimen	**Immediate referral to IDPH 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 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.**
• **Specialist**	**Metabolic disease specialist (IDPH/DSCC designated), see Appendix B for the contact information.**

Biotinidase Deficiency - Continued

- **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 IDPH Web site, http: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, http://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 in nanograms per milliliter (ng/mL)***
• **Reporting Ranges** **Abnormal results reported by phone, letter and fax to physician of record or hospital contact unless otherwise specified.**	**Follow-up referrals/testing for abnormal results**
Due to the effects of pre-maturity and physiological stress on 17-OHP levels, a tiered system of reporting for abnormal results has been developed for pre-term, low birth weight and NICU infants by the IDPH Newborn Screening Laboratory.	**Pre-term infants are defined as gestational age of ≤ 36 wks.** **Low birth weight is defined as birth weight < 2000 gms.** **Very low birth weight is defined as ≤1500 gms.**
Presumptive positive abnormal **Full-term infant ≥100 ng/mL** **Pre-term infant, low birth weight not in NICU ≥130ng/mL**	**Immediate consult /referral with pediatric endocrinologist and serum electrolytes and 17-OHP**
Pre-term infant, low birth weight in NICU ≥160ng/mL	**Consult with pediatric endocrinologist with evaluation of risk for CAH, serum electrolytes and 17-OHP**

Congenital Adrenal Hyperplasia - Continued

• **Reporting Ranges****	**Follow-up referrals/testing for abnormal results**
Suspect borderline abnormal (reported by letter only) Full-term infant ≥48-100 ng/mL	**Medical evaluation and repeat newborn screen within one to two days**
Pre-term, low birth weight not in NICU ≥100-130 ng/mL	**Medical evaluation and repeat newborn screen within one to two days; mark retest on specimen card.**
Pre-term, low birth weight in NICU ≥100-160 ng/mL	**Medical evaluation and repeat newborn screen at day 14 of life, or prior to discharge, whichever comes first**
Very low birth weight, in NICU with an initial abnormal 17-OHP (reported by letter only)	**NICU guidelines*** require a repeat screen at day 14 of life, unless the infant has symptoms of CAH which would require consult with a pediatric endocrinologist.**
• **Feeding Effect**	**None**
• **Transfusion Effect collection obtained enter date prior to specimen the specimen card.**	**If post-transfusion specimen is required, sample should be 48 hours post-transfusion; of last transfusion collection on**
• **Timing Effect**	**If specimen is collected at less than 24 hours of age, submit second sample during second day of life.**
• **Specialist**	**Pediatric endocrinologist (IDPH/DSCC designated), see Appendix B for the contact information.**
• **Treatment**	
Simple virilizing CAH	**Daily cortisol replacement therapy**
Salt-wasting CAH	**Daily cortisol and aldosterone replacement therapy, dietary salt supplements**

Congenital Adrenal Hyperplasia - Continued

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

Treatment with glucocorticoids (cortisone, dexamethasone) may cause false negative results.

***Most reference laboratories measure 17-OHP levels in nanograms per deciliter (ng/dL) and caution should be used when comparing newborn screening results with reference laboratory results; please consult 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 IDPH Division of Laboratories at 312-793-4752.**

*****New 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 two weeks of age.**

Congenital Hypothyroidism

Congenital hypothyroidism results from an inability of the thyroid gland to produce adequate amounts of thyroid hormone, thyroxine. Congenital hypothyroidism usually results from 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 IDPH Web site, http: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 http://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, T4 is measured. Levels are measured by fluorometric assay.**
• **Reporting Ranges*** **All abnormal results reported by phone, letter and fax, unless otherwise specified.**	**Follow-up referrals/testing for abnormal screening results**
<u>Presumptive positive abnormal</u> **TSH ≥ 54 uIU/mL** **T4 < 5 ug/dL**	**Immediate referral to pediatric endocrinologist, serum TSH and free T4**
<u>Suspect borderline abnormal</u> **TSH ≥36-54 uIU/mL** **T4 ≥5-8 ug/dL** **(reported by letter only)**	**Medical evaluation and repeat newborn screen within one to two days; mark retest on specimen card.**
• **Feeding Effect**	**None**
• **Transfusion Effect** **collection** **transfusion;**	**If post-transfusion specimen is required, collection is best performed 48 hours post-** **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 (IDPH/DSCC designated), see Appendix B 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 IDPH 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 IDPH Web site, http: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 http://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. If IRT is elevated, DNA mutation analysis is performed for a panel of common CF mutations.**
• **Reporting Ranges** **Abnormal results reported by phone, letter, fax to physician of record or hospital contact.**	**Follow-up referrals/testing for abnormal screening results**
Presumptive Positive **-ITR** elevated with two CFTR mutations** **-IRT** elevated with one CFTR mutation** **-Seriously elevated IRT **** **specialist**	**Immediate referral*** to CF** **for diagnostic sweat testing and genetic counseling**
• **Feeding Effect**	**None**
• **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.

Cystic Fibrosis - Continued

• **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. Complete list not available at time of printing, contact IDPH/DSCC for contact information.**

Comments

***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.**

****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 IDPH Division of Laboratories at 312-793-4752.**

*****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.**

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, and 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 IDPH Web site, http: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 http://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 (SCAD)
Medium chain acyl-CoA dehydrogenase deficiency (MCAD)
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD) or
Trifunctional protein deficiency (TFPD)
Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)
Carnitine palmitoyl transferase deficiency type 2 (CPT 2) or
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)

- **Incidence** — **MCAD - one in 18,000 births**
  LCHAD - one in 400,000 births
  VLCAD - one in 100,000 births
  SCAD - 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) in micro-Moles per Liter (uM/L)**

Analytes	Possible Disorder
Butanoyl carnitine (C4) is the primary analyte	SCAD, IBCD**, also GA2/MADD
Multiple medium chain acylcarnitines Octanoyl carnitine (C8) is the primary analyte (C8, C10, C10:1 and C6)***	MCAD
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.**

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 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 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.**

Fatty Acid Oxidation Disorders - Continued

• **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.**
• **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.**
to	**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 avoid false negative results for FAO disorders.**
• **Specialist**	**Metabolic disease specialist, see Appendix B for a list of IDPH /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, intra-venous therapy may be required during intercurrent illnesses. If indicated, IDPH provides special medically necessary formula without charge to Illinois residents who are under comprehensive medical management provided by a metabolic**

disease specialist designated by IDPH.

Fatty Acid Oxidation Disorders - Continued

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 D for a complete listing of acylcarnitine analytes measured by IDPH laboratory tandem mass spectrometry.**

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

*****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 IDPH 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 galactosemia is essential to prevent severe liver disease and complications including bleeding, overwhelming sepsis and death in the early neonatal period. See the IDPH Web site, http: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 http://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) is determined by fluorometric assay. If galactose level is elevated, GALT enzyme activity is determined by Beutler assay.**
• **Reporting Ranges*** **Abnormal results reported by phone, letter, fax to physician of record or hospital contact.**	**Follow-up referrals/testing for abnormal screening results**
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 to use breast pump to maintain milk supply until diagnosis is confirmed or ruled out.**
Suspect Borderline Abnormal **Total galactose ≥ 6.5mg/dL with Reduced GALT activity present**	**Evaluate baby's condition, collect second newborn screening specimen, within one to two days; mark retest box and indicate the baby's feeding type on the specimen card.**
• **Feeding Effect**	**Screening will be affected. Mark specimen collection form to indicate type of feeding (breast, soy, other, NPO). This informs IDPH laboratory staff that GALT enzyme activity testing may be necessary although galactose level is normal.**

Galactosemia - Continued

• **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.**
• **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.**
• **Specialist**	**Metabolic disease specialist (IDPH/DSCC designated), see Appendix B 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.

When infants are on soy formula feeding, or have not had an oral galactose feeding, galactose values may not be meaningful or valid, and screening for GALT enzyme activity (Beutler assay) should be requested.

***IDPH 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 IDPH 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 IDPH Web site, http: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 http://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 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

Organic Acid Disorders - Continued

Analytes	Possible Disorder
Propanoyl carnitine (C3) is the primary analyte	**MMA/PA**
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.

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 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 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.**

- **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**	**Metabolic disease specialist (IDPH/DSCC designated), see Appendix B 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, intra-venous therapy may be required during intercurrent illness and introduction of new foods.**
	If indicated, IDPH provides special medical formula without charge to Illinois residents who are under comprehensive medical management provided by a metabolic disease specialist designated by IDPH.

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 D for a complete listing of acylcarnitine analytes measured by IDPH laboratory tandem mass spectrometry.**

****For questions about the most current laboratory reporting values for these disorders, contact IDPH 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 IDPH Web site, http: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 http://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, letter, and fax to physician of record or hospital contact unless otherwise specified.**	**Follow-up referrals/testing for abnormal screening results**
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.**
Suspect Amino Acid Abnormal due to TPN (reported by letter and fax) **Slightly elevated phenylalanine level usually associated with TPN effects**	**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.**

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 (IDPH/DSCC designated), see Appendix B for the contact information.**
• **Treatment**	**Low phenylalanine diet consisting of specialized medically necessary formula in combination with foods that are low in phenylalanine. IDPH provides PKU medically necessary formula without charge to Illinois residents who are under comprehensive medical management provided by a metabolic specialist designated by IDPH/DSCC.**

Comments

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

Sickling Hemoglobin Disorders and Beta Thalassemia (FS, FSC, FSa, FNA)

This group of autosomal recessive hemoglobin disorders is characterized by production of abnormal forms of hemoglobin and no adult, or normal hemoglobin. 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. See the IDPH Web site, http: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 http://www.acmg.net to review the ACMG "ACT" sheet.

- **Incidence**

  all races

  Sickle cell disease occurs in one in 375 African-Americans. Sickling hemoglobinopathies occur in 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 Screening**

  Identification of types of hemoglobin present in the sample is performed by high performance liquid chromatography (HPLC). This screening is not quantitative.

- **Reporting Classifications**
  Abnormal results reported by phone, letter and fax to physician of record or hospital contact

  FS - Fetal and sickle hemoglobins (probable sickle cell disease)

Sickling Hemoglobin Disorders and Beta Thalassemia (FS, FSC, FSa, FNA) - Continued

• **Reporting Classifications**	**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)**
• **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 sickling disorders due to transfused adult hemoglobin. Upon screening, specimens from transfused infants with disease conditions may present as carrier (trait) conditions.**
• **Timing Effect**	**Usually no effect in first two weeks of newborn period, although older infants will have a gradual decrease in fetal hemoglobin.**
• **Follow-up Referrals/Testing for Abnormal Screening Results**	**Diagnostic testing, including family counseling and education is necessary. Referral to pediatric hematologist for diagnosis and counseling is highly recommended.**
• **Specialist**	**Pediatric hematologist, see Appendix B for contact information.**
• **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.**

Sickling Hemoglobin Disorders and Beta Thalassemia (FS, FSC, FSa, FNA) - Continued

• **Treatment**	**Prophylactic penicillin is not usually indicated for treatment of beta-thalassemia; this disorder is associated with severe anemia usually requiring medical management by a pediatric hematologist.**

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 are available through the Sickle Cell Disease Association of Illinois. See page 13 of this manual for contact information.

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 http://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 in Screening**	**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)**	**FAS - Fetal, adult and sickle hemoglobins (probable sickle cell trait)** **FAC - probable hemoglobin C trait)** **FAD – probable hemoglobin D trait)**

Other Hemoglobinopathies and Traits - Continued

• **Reporting Classifications**	**FAE – Fetal , adult and E hemoglobins (probable E hemoglobin trait)** **FD – Fetal and D hemoglobins (probable hemoglobin D disease)*** **FE – Fetal and E hemoglobins (probable hemoglobin E disease)*** **LFA – Low fetal and elevated adult hemoglobins (probable transfusion or older child)**
• **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 fetal hemoglobin level usually decreases.**
• **Specialist**	**Pediatric hematologist, see appendix B for contact information.**
• **Treatment**	**Carrier states (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 a pediatric hematologist is recommended.** **Some types of thalassemia may cause serious anemia, in some cases requiring transfusion therapy; referral to a pediatric hematologist is recommended.**

Other Hemoglobinopathies and Traits - Continued

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 B for a listing of pediatric hematology specialists for information about these services.

Sickle Cell Disease Association of Illinois also will provide educational services to families of individuals with sickle cell diseases or traits. See page 13 of this manual for contact information.

***Referral to a pediatric hematologist for additional diagnostic testing is recommended for these conditions.**

Appendix A

Filter Paper Collection Form

Image of the Illinois Newborn Screening ILP Series Filter Paper Collection Form
(Front view)

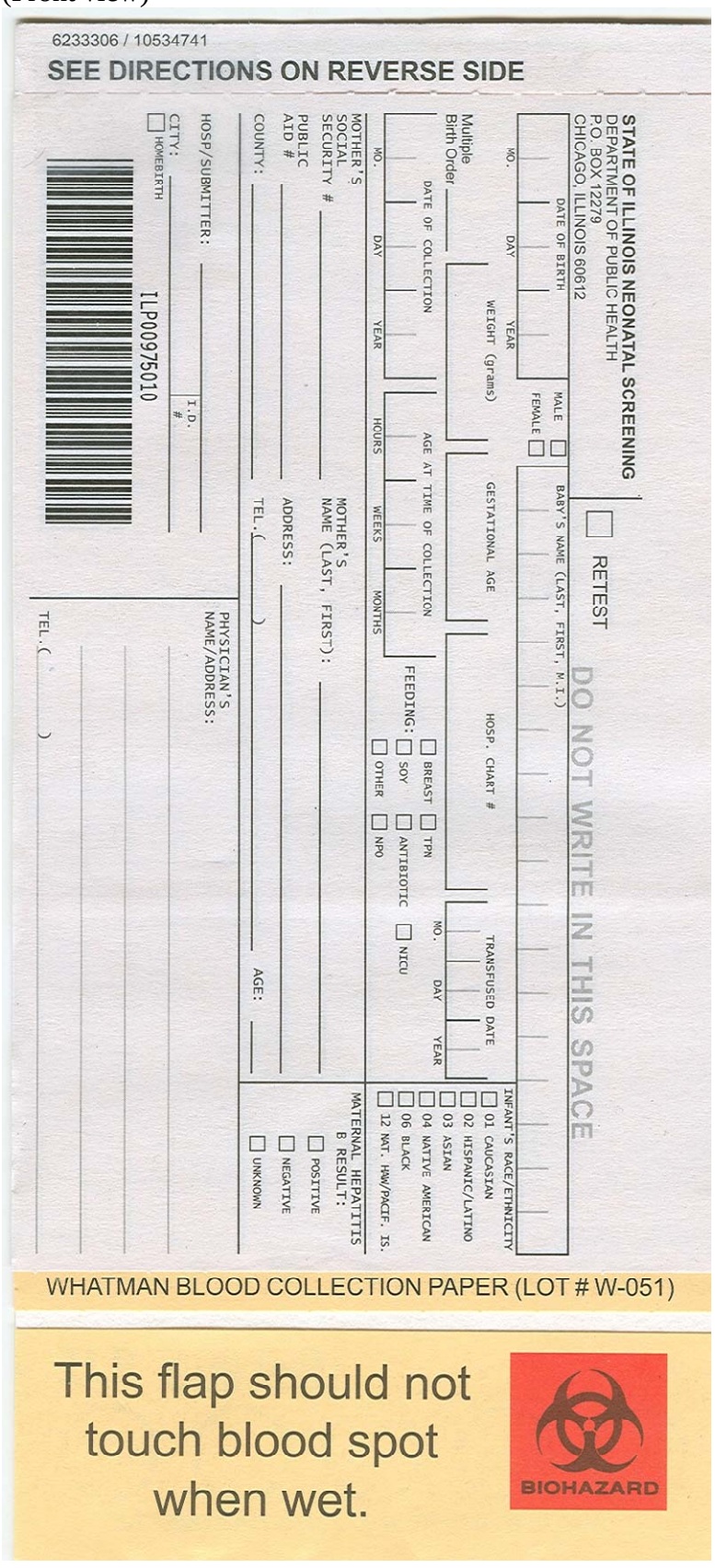
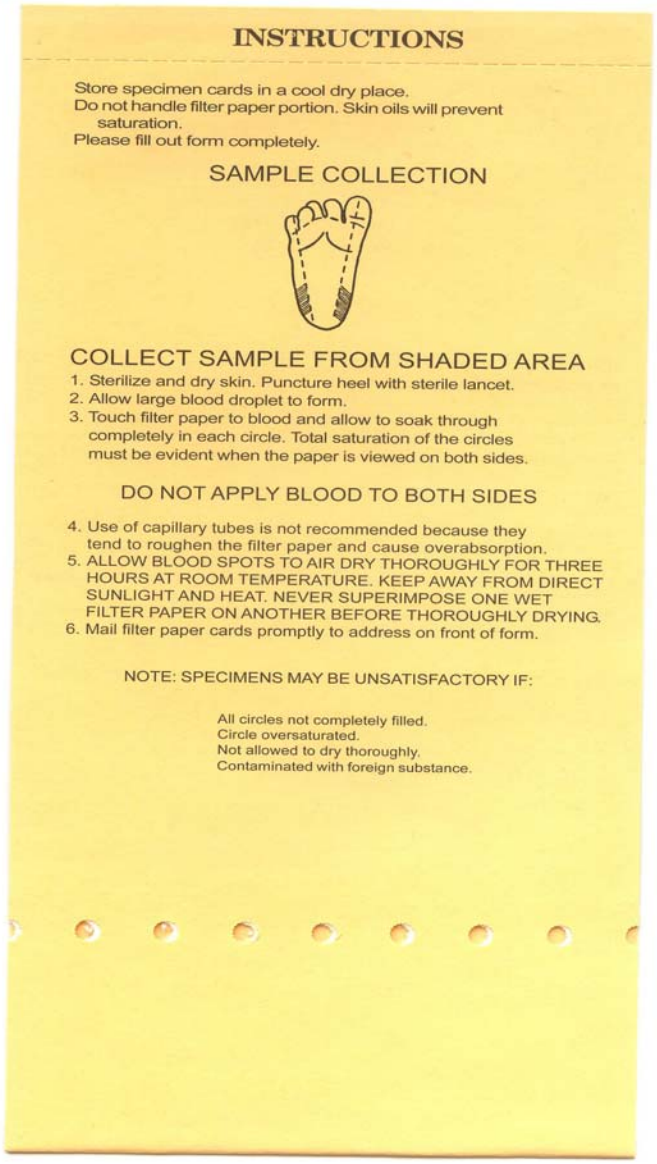


Image of the Illinois Newborn Screening ILP Series Filter Paper Collection Form
(Back view)



INSTRUCTIONS

Store specimen cards in a cool dry place.
Do not handle filter paper portion. Skin oils will prevent saturation.
Please fill out form completely.

SAMPLE COLLECTION

COLLECT SAMPLE FROM SHADED AREA

1. Sterilize and dry skin. Puncture heel with sterile lancet.
2. Allow large blood droplet to form.
3. Touch filter paper to blood and allow to soak through completely in each circle. Total saturation of the circles must be evident when the paper is viewed on both sides.

DO NOT APPLY BLOOD TO BOTH SIDES

4. Use of capillary tubes is not recommended because they tend to roughen the filter paper and cause overabsorption.
5. ALLOW BLOOD SPOTS TO AIR DRY THOROUGHLY FOR THREE HOURS AT ROOM TEMPERATURE. KEEP AWAY FROM DIRECT SUNLIGHT AND HEAT. NEVER SUPERIMPOSE ONE WET FILTER PAPER ON ANOTHER BEFORE THOROUGHLY DRYING.
6. Mail filter paper cards promptly to address on front of form.

NOTE: SPECIMENS MAY BE UNSATISFACTORY IF:

All circles not completely filled.
Circle oversaturated.
Not allowed to dry thoroughly.
Contaminated with foreign substance.

Appendix B

Pediatric Medical Specialists

Illinois Department of Public Health
Newborn Screening Program
Pediatric Medical Specialists for
Amino Acid, Organic Acid and Fatty Acid Oxidation Disorder

Chicago and Suburban Areas

Children's Memorial Hospital
Division of Genetics
2300 Children's Plaza, Box 59
Chicago, IL 60614
773-880-4462
Barbara K. Burton, M.D.
Joel Charrow, M.D.

Children's Pediatric Specialty Services
2301 Enterprise Drive
Westchester, IL 60154
For appointment call: 773-880-4462
Barbara K. Burton, M.D.

University of Illinois at Chicago
PKU Clinics
Division of Genetics
840 S. Wood St., Room 1215, M/C 856
Chicago, IL 60612-7311
312-355-0732
George Hoganson, M.D.

Division of Genetics
Department of Pediatrics
840 S. Wood St., M/C 856
312-355-0732
Allen L. Horwitz, M.D., Ph.D.

Advocate Christ Hospital/Hope Children's Hospital
4440 W. 95th St.
Oak Lawn, IL 60453
For appointment call: 312-355-0732
George Hoganson, M.D.

Hindsdale Hospital
120 Oak St.
Hinsdale, IL 60521
For appointment call: 312-355-0732
George Hoganson, M.D.

Rush University Medical Center
Genetics Section
1750 W. Harrison St., Room 1507, Jelke Bldg.
Chicago, IL 60612
312-942-6298
Paul Wong M.D., M.Sc.

Peoria Area

University of Illinois at Peoria
507 E. Armstrong Ave.
Peoria, IL 61603
For appointment call: 312-355-0732
George Hoganson, M.D.

Rockford Area

Rockford Memorial Hospital
2400 N. Rockton Ave.
Rockford, IL 61103
For appointment call: 312-355-0732
George Hoganson, M.D

St. Louis Area

St. Louis Children's Hospital
Division of Medical Genetics
Washington University School of Medicine
660 S. Euclid Ave., Campus Box 8116
St. Louis, MO 63110
314-454-6093
Dorothy K. Grange, M.D.
Tyler Reimschisel, M.D.

Cardinal Glennon Children's Hospital
Director, Division of Medical Genetics
Department of Pediatrics
Saint Louis University School of Medicine
1465 S. Grand Blvd.
St. Louis, MO 63104
314-577-5639
Gary S. Gottesman, M.D.

Illinois Department of Public Health
Newborn Screening Program
Pediatric Medical Specialists for
Biotinidase Deficiency and Galactosemia Disorders

Chicago and Suburban Areas

Children's Memorial Hospital
Division of Genetics
2300 Children's Plaza, Box 59
Chicago, IL 60614
773-880-4462
Joel Charrow, M.D.
Barbara K. Burton, M.D.

Children's Pediatric Specialty Services
2301 Enterprise Drive
Westchester, IL 60154
For appointment call: 773-880-4462
Barbara K. Burton, M.D.

University of Illinois at Chicago
PKU Clinics
Division of Genetics
840 S. Wood St.
Room 1217, M/C 856
Chicago, IL 60612-7311
312-355-0732
George Hoganson, M.D.
Allen L. Horwitz, M.D., Ph.D.

The University of Chicago
Dept. of Human Genetics and Pediatrics
5841 S. Maryland Ave., Suite L161, M/C 0077
Chicago, IL 60637
773-834-9110
Darrel J. Waggoner, M.D.

Rush University Medical Center
Genetics Section
1750 W. Harrison St., Room 1507, Jelke Bldg.
Chicago, IL 60612
312-942-6298
Paul Wong, M.D., M.Sc.
David S. Kang, M.D., Ph.D.

Copley Medical Center
Fox Valley Medical Associates Bldg.
Route 3 at Fox Valley Center
Aurora, IL 60505
For appointment call: 312-942-6298
Paul Wong, M.D., M.Sc.

Riverside Medical Center
350 N. Wall St.
Kankakee, IL 60901
For appointment call: 312-942-6298
Paul Wong, M.D., M.Sc.

Saint Joseph Medical Center
333 N. Madison St.
Joliet, IL 60435
For appointment call: 312-942-6298
Paul Wong, M.D., M.Sc.

Poplar Creek/Hoffman Estates
1800 McDonough Road, #203
Hoffman Estates, IL 60192
847-608-7517
Paul Wong, M.D., M.Sc.

Loyola University Medical Center
Dept. of Pediatrics/Section of Genetics
2160 S. First Ave.
Maywood, IL 60153
708-327-9085
Carolyn H. Jones, M.D.

Advocate Lutheran General Hospital
Parkside Center
1875 Dempster St., Suite 310
Park Ridge, IL 60068
847-723-7705
Carol Booth, M.D.
Debra Rita, M.D.

(Continued)

Pediatric Medical Specialists for
Biotinidase Deficiency and
Galactosemia Disorders (Continued)

Chicago and Suburban Areas

John A. Stroger Jr. Hospital of Cook County
Division of Genetics & Metabolics
1900 W. Polk St., Room 1159
Chicago, IL 60612
312-864-4158
Susan C. Echiverri, M.D.

Advocate Christ Hospital/Hope Children's Hospital
4440 W. 95th St.
Oak Lawn, IL 60453
For appointment call: 312-355-0732
George Hoganson, M.D.

Hinsdale Hospital
120 Oak St.
Hinsdale, IL 60521
For appointment call: 312-355-0732
George Hoganson, M.D.

Peoria Area

University of Illinois at Peoria
507 E. Armstrong Ave.
Peoria, IL 61603
For appointment call: 312-355-0732
George Hoganson, M.D.

Rockford Area

Rockford Memorial Hospital
2400 N. Rockton Ave.
Rockford, IL 61103
For appointment call: 312-355-0732
George Hoganson, M.D.

Springfield Area

Southern Illinois School of Medicine
Deparment of Pediatrics
P.O. Box 19658
Springfield, IL 62794
217-545-4839
Michael Schneider, M.D.

Sangamon County Department of Public Health
1415 E. Jefferson St.
Springfield, IL 62703
For appointment call: 312-355-0732
George Hoganson, M.D.

St. Louis Area

St. Louis Children's Hospital
Division of Medical Genetics
Washington University School of Medicine
660 S. Euclid Ave., Campus Box 8116
St Louis, MO 63110
314-454-6093
Dorothy K. Grange, M.D.
Tyler Reimschisel, M.D.

Cardinal Glennon Children's Hospital
Director, Division of Medical Genetics
Department of Pediatrics
Saint Louis University School of Medicine
Cardinal Glennon Children's Hospital
1465 S. Grand Blvd.
St. Louis, MO 63104
314-577-5639
Gary S. Gottesman, M.D.

Illinois Department of Public Health
Newborn Screening Program
Pediatric Medical Specialists for
Phenylketonuria

Chicago and Suburban Areas

Children's Memorial Hospital
Division of Genetics
2300 Children's Plaza, Box 59
Chicago, IL 60614
773-880-4462
Barbara K. Burton, M.D.

Children's Pediatric Specialty Services
2301 Enterprise Drive
Westchester, IL 60154
773-880-4462
Barbara K. Burton, M.D.

Advocate Christ Hospital/Hope Children's Hospital
4440 W. 95th St.
Oak Lawn, IL 60453
For appointment call: 312-355-0732
George Hoganson, M.D.

University of Illinois at Chicago
PKU Clinics
Division of Genetics
840 S. Wood St., Room 1215, M/C 856
Chicago, IL 60612-7311
312-355-0732
George Hoganson, M.D.

Hinsdale Hospital
120 Oak St.
Hinsdale, IL 60521
For appointment call: 312-355-0732
George Hoganson, M.D.

Rush University Medical Center.
Division, Genetics Section
1750 W. Harrison St., Room 1507, Jelke Bldg.
Chicago, IL 60612
312-942-6298
Paul Wong, M.D., M.Sc.

Saint Joseph Medical Center
333 N. Madison St.
Joliet, IL 60435
For appointment call: 312-942-6298
Paul Wong, M.D., M.Sc.

Copley Medical Center
Fox Valley Medical Associates Bldg.
Route 3 at Fox Valley Center
Aurora, IL 60505
For appointment call: 312-942-6298
Paul Wong, M.D., M.Sc.

Riverside Medical Center
350 N. Wall St.
Kankakee, IL 60901
For appointment call: 312-942-6298
Paul Wong, M.D., M.Sc.

Peoria Area

University of Illinois at Peoria
507 E. Armstrong Ave.
Peoria, IL 61603
For appointment call: 312-355-0732
George Hoganson, M.D.

(Continued)

Pediatric Medical Specialists for Phenylketonuria (Continued)

Rockford Area

Rockford Memorial Hospital
2400 N. Rockton Ave.
Rockford, IL 61103
For appointment call: 312-355-0732
George Hoganson, M.D.

Springfield Area

Sangamon County Department of Public Health
1415 E. Jefferson St.
Springfield, IL 62703
For appointment call: 312-355-0732
George Hoganson, M.D.

St. Louis Area

St. Louis Children's Hospital
Division of Medical Genetics
Washington University School of Medicine
660 S. Euclid Ave., Campus Box 8116
St. Louis, MO 63110
314-454-6093
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Cardinal Glennon Children's Hospital
Director, Division of Medical Genetics
Department of Pediatrics
Saint Louis University School of Medicine
Cardinal Glennon Children's Hospital
1465 S. Grand Blvd.
St. Louis, MO 63104
314-577-5639
Gary S. Gottesman, M.D.

Illinois Department of Public Health
Newborn Screening Program
Pediatric Medical Specialists for
Congenital Adrenal Hyperplasia and Congenital Hypothyroidism

Chicago and Suburban Areas

Children's Memorial Hospital
2300 Children's Plaza
Chicago, IL 60614
773-327-7740
Reema Habiby, M.D.
Mary Kreiter, M.D.

Rush University Medical Center
Department of Pediatrics
1653 W. Congress Parkway
Chicago, IL 60612
1725 W. Harrison St., Suite 938
312-942-8989
Richard A. Levy, M.D.

University of Illinois at Chicago
Department of Endocrinology and Pediatrics
840 S. Wood St., M/C 856
Chicago, IL 60612
312-996-7714 clinic
312-996-1795 office
Songya Pang, M.D.

The University of Chicago
Pediatric Endocrinology
5841 S. Maryland Ave.
Chicago, IL 60637
773-702-6432
Robert Rosenfield, M.D.
Dianne Deplewski, M.D.
Elizabeth Baumann-Littlejohn, M.D.

ENH Medical Group:
Old Orchard Pediatric Endocrinology
9977 Woods Drive
Skokie, IL 60077
847-663-8508
Stephen C. Duck, M.D.

Advocate Christ Hospital
Oak Lawn, IL 60453
708-684-5670
Fuad Ziai, M.D.

Loyola University Medical Center
2160 S. First Ave., Bldg. 105, Room 3330
Maywood, IL 60153
For appointment call: 708-216-8563 or
708-327-9095

120 Spalding, Suite. 401
Naperville, IL 60540
630-416-4501
W. Patrick Zeller, M.D.

Advocate Lutheran General Hospital
1775 Dempster St.
Park Ridge, IL 60068
847-723-8409
Kanika Ghai, M.D.

Peoria Area

University of Illinois at Peoria
Pediatric Sub-Specialty Clinic
320 E. Armstrong Ave.
Peoria, IL 61603-3103
309-624-9680
Sue Ellyn Sauder, M.D.
Joyce E. Wise, M.D.
William F. Maurer, M.D.

Department of Pediatrics
530 N.E. Glen Oak Ave.
Peoria, IL 61637
309-624-9680
Rodney A. Lorenz, M.D.

(Continued)

Pediatric Medical Specialists for
Congenital Adrenal Hyperplasia and
Congenital Hypothyroidism (Continued)

St. Louis Area

Cardinal Glennon Children's Hospital
St. Louis University Medical School
1465 S. Grand Blvd.
St. Louis, MO 63104
314-577-5648
Sherida Tollefsen, M.D.
David Paul Dempsher, M.D.
Susan E. Myers, M.D.

St. Louis Children's Hospital
Washington University School of Medicine
1 Children's Place
Campus Box 8116, Room 4530
St. Louis, MO 63110
314-454-6051
Neil H. White, M.D.
Bess Adkins Marshall, M.D.
Louis Joseph Muglia, M.D., Ph.D.
Abby Solomon Hollander, M.D.

Illinois Department of Public Health
Newborn Screening Program
Pediatric Medical Specialists for
Hemoglobinopathies/Sickle Cell Disease

Chicago and Suburban Areas

Children's Memorial Hospital-Northwestern University*
Division of Hematology/Oncology
Comprehensive Sickle Cell Program
2300 Children's Plaza, #30
Chicago, IL 60614
773-880-4125
Alexis Thompson, M.D.

Advocate Christ/Hope Children's Hospital
Pediatric Hematology/Oncology
4440 W. 95th St.
Oak Lawn, IL 60453
708-346-4094
Sharad Salvi, M.D.

John A. Stroger, Jr. Hospital of Cook County*
Division of Hematology/Oncology
1900 W. Polk, Room 1155
Chicago, IL 60612
312-864-4166
Lilly Mathew, M.D.

Rush University Medical Center*
1753 W. Congress Parkway
Chicago, IL 60612
312-942-5983
Allen Korenblit, M.D., F.A.A.P.

Mt. Sinai Hospital*
Department of Pediatrics
California Ave. at 15th St., Room F444
Chicago, IL 60608
773-257-6395 (diagnostic scheduling)
Sudha Rao, M.D.

Loyola University Medical Center
Department of Pediatrics
Hematology/Oncology Section
2160 S. First Ave.
Maywood, IL 60153
708-327-9136
Ricarchito Manera, M.D.

Advocate Lutheran General Children's Hospital
Department of Pediatrics
1675 Dempster St.
Park Ridge, IL 60068
847-318-9330
Jong-Hyo Kwon, M.D.
William Goodell, M.D.

The University of Chicago Hospitals*
5841 S. Maryland Ave.
WCHC430, MC 4060
Chicago, IL 60637
Patricia Bailey, R.N., Sickle Cell Clinic
773-256-5759
Uma Subramanian, M.D.

University of Illinois at Chicago*
Division of Pediatric Hematology/Oncology
840 S. Wood St., MC 856
Chicago, IL 60612-7324
Richard J. Labotka, M.D.
312-996-6143

(Continued)

Pediatric Medical Specialists for Hemoglobinopathies/ Sickle Cell Diseases (Continued)

Champaign Area

Carle Clinic*
Department of Pediatrics
602 W. University St.
Urbana, IL 61801
217-383-3100
Mark Musselman, M.D., M.A.

Peoria Area

St. Francis Medical Center*
Department of Pediatrics
507 E. Armstrong Ave.
Peoria, IL 61603
309-624-4945
Kay Saving, M.D.
309-624-9856

Springfield Area

Southern Illinois University School of Medicine
Carol Jo Vecchie
Women and Children's Center
415 N. 9th St.
P.O. Box 19678
Springfield, IL 62794-9678
217-545-5817
Gregory Brandt, M.D.

East St. Louis/St. Louis Area

Southern Illinois Health Care Foundation Mother Child Center*
6000 Bond Ave.
Centreville, IL 62207
618-332-2740 (ask for sickle cell nurse)
William Ferguson, M.D.

St. Louis Children's Hospital*
Washington University School of Medicine
1 Children's Place
St. Louis, MO 63110
314-454-6018 or 314-454-2193
Michael DeBaun, M.D.

Cardinal Glennon Children's Hospital*
St. Louis University School of Medicine
Department of Pediatrics
Division of Hematology/Oncology
1465 South Grand Blvd.
St. Louis, MO 63104-1095
314-577-5600
William Ferguson, M.D.

***Centers receiving grant funding from the Illinois Department of Public Health**

Appendix C

Local Public Health Departments

Illinois Department of Public Health
Newborn Screening Program
Local Public Health Department Contacts

Adams County Health Department 333 N. Sixth St. Quincy, IL 62301 Candee Musgrove, R.N. 217- 222-8440	**Bond County Health Department** 503 S. Prairie Greenville, IL 62246 Judi Markell, R.N. 618-664-1442	**Boone County Health Department** 1204 Logan Ave. Belvidere, IL 61008-4031 Pam Hart, R.N. 815-544-2951 or 9730
Brown County Health Department 111½ W. Washington St. Mount Sterling, IL 62353 Nancy Kropp, R.N. 217-773-2714	**Bureau County Health Department** 526 Bureau Valley Parkway Princeton, IL 61356 Colleen Sailer, R.N., B.S.N. 815-872-5091	**Calhoun County Health Department** P.O. Box 158, 210 French St. Hardin, IL 62047 Judy Zipprich, R.N. 618-576-2428
Cass County Health Department 331 S. Main St. Virginia, IL 62691 Marsha Kirchner, R.N., B.S.N. 217-452-3057 or 217-323-2182 Beardstown Office	**Champaign-Urbana Public Health District** 710 N. Neil St. Champaign, IL 61820-1488 Ellen Weise, R.N. 217-352-4289	**Chicago Department of Health** DePaul University Center 333 S. State St., Room. 2105 Chicago, IL 60604 Pam Hegbe, R.N. 312-747-9690
Christian County Health Department 902 W. Springfield Road Taylorville, IL 62568 Marcie Hurley, R.N. 217-824-4113	**Clark County Health Department** 1001 N. York St. Martinsville, IL 62442 JoBeth Gilbert, R.N. 217-382-4207	**Clay County Health Department** 601 E. 12th St. Flora, IL 62839 Nancy Wiley, R.N. 618-662-4406
Clinton County Health Department 930A Fairfax St. Carlyle, IL 62231 Janice Albers, R.N. 618-594-8942	**Coles County Health Department** P.O. Box 1064, 825 18th St. Charleston, IL 61920-9391 Jill Temple, R.N. 217-348-0530	**Cook County Department of Public Health** 1010 Lake St., Suite 104 Oak Park, IL 60301 Pat Rewers, R.N. 708-450-4787
Crawford Co. Health Department 202 N. Bline Blvd. Robinson, IL 62454 Debbie Pryor, R.N. 618-544-8798	**Cumberland Coounty Health Department** P.O. Box 130, N.E. Corner of Square Toledo, IL 62468 Connie Kerner, R.N. 217-849-3211	**DeKalb County Health Department** 2550 N. Annie Glidden Road DeKalb, IL 60115 Liz Carney, R.N. 815-748-2447 or 815-758-6673
DeWitt-Piatt Bi-County Health Department P.O. Box 518, 910 Route 54 E. Clinton, IL 61727 Ann Barnett, R.N. 217-935-3427	**Douglas County Health Department** 1250 East U.S. Highway 36 Tuscola, IL 61953 Susan Hays, R.N. 217-253-4137	**DuPage County Health Department** 111 N. County Farm Road Wheaton, IL 60187 Ellen Finegan, R.N. 630-682-7979, ext. 7049
East Side Health District 638 N. 20th St. East St. Louis, IL 62205 Carolyn Martin, R.N., B.S.N. 618-874-4713 or 618-274-4888 Covering: E. St. Louis, Washington Park, Fairmount City, Southwest area of Caseyville, Cahokia, Alorton, Centreville, Sauget, Brooklyn and Lovejoy cities	**Edgar County Health Department** 502 Shaw Ave. Paris, IL 61944 Carol Cline, R.N. 217-465-2212	**Effingham County Health Department** 901 W. Virginia, P.O. Box 685 Effingham, IL 62401 Shawn Bourland, R.N., B.S.C.L.C 217-342-9237

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Local Health Department Contacts (Continued)

Egyptian County Health Department Route 3, Box 90A 1412 U.S. Highway 45 N. Eldorado, IL 62930-9234 Lynn Schanzle, R.N. 618-273-3326 Covering: Galatin, Saline, White counties	**Evanston Health Department** Evanston Civic Center 2100 Ridge Ave. Evanston, IL 60201 Evonda Thomas, R.N. 847-866-2957	**Fayette County Health Department** P.O. Box 340 509 W. Edwards St. Vandalia, IL 62471 Debby Lay, R.N. 618-283-1044
Ford-Iroquois Public Health Department 114 N. Third St. Watseka, IL 60970 Lyn Schaumburg, R.N. 815-432-2483	**Franklin-Williamson Bi-County Health Department** 120 Express Drive Marion, IL 62959-9808 Lisa Sorenson, R.N. 618-993-8111	**Fulton County Health Department** 700 E. Oak St. Canton, IL 61520 Louise Hiett, R.N. 309-647-1134
Greene County Health Department 310 Fifth St. Carrollton, IL 62016 Susan Thornton, R.N. 217-942-6961 or 217-942-6962	**Grundy County Health Department** 1320 Union Str. Morris, IL 60450 Kay Lynn Shumaker, R.N. 815-941-3400	**Hamilton County Health Department** Courthouse, Room 5 McLeansboro, IL 62859 Andrea Miller, R.N. 618-643-3522
Hancock County Health Department 73 S. Adam St. Carthage, IL 62321 Jennifer Sherman, R.N. 217-357-2171	**Henderson County Health Department** P.O. Box 220 Gladstone, IL 61437-0220 Mary Lynne Haas, R.N. 309-627-2812 Covering: Warren county	**Henry County Health Department** 4424 U.S. Highway 34 Kewanee, IL 61443 Sue Duncan, R.N. 309-852-0197
Jackson County Health Department Route 13 at Country Club Road P.O. Box 307 Murphysboro, IL 62966 Marilyn Twitty, R.N. 618-684-3143, ext. 111	**Jasper County Health Department** 106 E. Edwards St. Newton, IL 62448 Marilyn Cox, R.N. 618-783-4436 Covering Richland county	**Jefferson County Health Department** #1 Doctors Park Road, Suite F Mt. Vernon, IL 62864 Vickie Nollman, R.N. 618-244-7134
Jersey County Health Department 1307 State Highway 109 Jerseyville, IL 62052 Geri Daniels, R.N., B.S.N. 618-498-9565, ext. 313	**JoDaviess County Health Department** 9483 U.S. Route 20 W. P.O. Box 318 Galena, IL 61036 Lori Stanglet, R.N. 815-777-0263	**Kane County Health Department** 1240 N. Highland Ave., Suite 12 Aurora, IL 60506 Carolyn Hammar, R.N. 630-264-7679
Kankakee County Health Department 2390 W. Station St. Kankakee, IL 60901 Janie Semlar, R.N. 815-937-7892	**Kendall County Department of Health & Human Services** 500A Countryside Center Yorkville, IL 60560 Noreen Transier, R.N. 630-553-9100	**Knox County Health Department** 1361 W. Fremont St. Galesburg, IL 61401 Carol Winbigler, R.N. 309-344-2224
Lake County Health Department 3010 Grand Ave. Waukegan, IL 60085 Carmen Perez, R.N. 847-377-8128	**LaSalle County Health Department** 717 Etna Road Ottawa, IL 61350 Colleen Gibson, R.N. 815-433-3366	**Lawrence County Health Department** P.O. Box 516 Lawrenceville, IL 62439 Mary Copp, R.N. 618-943-3302

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Local Health Department Contacts (Continued)

Lee County Health Department 309 S. Galena Ave., Suite 100 Dixon, IL 61021-9185 Cyndi Kenney, R.N. 815-284-3371	**Livingston County Health Department** Livingston Co. Health & Education Bldg. P.O. Box 886, 310 E. Torrance Ave. Pontiac, IL 61764 Melinda Hillman, R.N. 815-844-7174	**Logan County Health Department** 109 Third St., P.O. Box 508 Lincoln, IL 62656 Sue Estes, R.N. 217-735-2317
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Rock Island County Health Department 2112 25th Ave. Rock Island, IL 61201 Franki Cunningham, R.N. 309-793-1955, ext. 260	**St. Clair County Health Department** #19 Public Square, Suite 150 Belleville, IL 62220 Karoline Stock, RN, BSN 618-233-7703, ext. 4483	**Sangamon County Department of Public Health** 2501 N. Dirksen Parkway Springfield, IL 62702 Andrea Evans, R.N. 217-535-3100
Schuyler County Health Department 127 S. Liberty St., P.O. Box 320 Rushville, IL 62681 Barbara Wrench, R.N. 217-322-4373	**Scott County Health Department** 32 E. Market St., P.O. Box 115 Winchester, IL 62690 Phyllis Jefferson, R.N. 217-742-8203	**Shelby County Health Department** R.R.2 , Box 54, 1810 W.S. Third St. Shelbyville, IL 62565 Carol Johnston, R.N. 217-774-2355

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Local Health Department Contacts (Continued)

Southern Seven Health Department 37 Rustic Campus Drive Ullin, IL 62992 Judi Duff, R.N. 618-634-2297, ext. 159 Covering: Alexander, Hardin, Johnson, Massac, Pope, Pulaski, Union Counties	**Skokie Health Department** 5127 Oakton St., P.O. Box 309 Skokie, IL 60077 Sue Reisbert, R.N. 847-933-8252	**Stark County Health Department** 4424 U.S. Highway 34 Kewanee, IL 61433 Sue Duncan, R.N. 309-852-3115
Stephenson County Health Department 10 W. Linden St. Freeport, IL 61032 Julia Marynus, R.N. 815-235-8394	**Tazewell County Health Department** 21306 Illinois Route 9 Tremont, IL 61568-9252 Sarah Buller Fenton, M.S., R.N.C. 309-925-5511, ext. 236	**Vermilion County Health Department** 200 S. College St. Danville, IL 61832 Pam Warner, R.N. 217-431-2662
Wabash County Health Department 130 W. Seventh St. Mount Carmel, IL 62863 Kendra Grounds, R.N. 618-263-3873	**Washington Co. Health Department** 177 S. Washington St. Nashville, IL 62263 Joyce Carson, R.N. 618-327-3644	**Wayne County Health Department** 405 N. Basin Road, P.O. Box 645 Fairfield, IL 62837 Pat Weber, R.N. 618-842-5166
Whiteside County Health Department 1300 W. Second St. Rock Falls, IL 61071 Pam Vanderinne 815-626-2230	**Will County Health Department** 501 Ella Ave. Joliet, IL 60433 Sharon Wesel, R.N. 630-679-7000	**Winnebago County Health Department** 401 Division St. Rockford, IL 61104 Paula Hart, R.N. 815-962-5092, ext. 273
Woodford County Health Department 109 S. Major St. Eureka, IL 61530 Nancy Allen, R.N. 309-467-2371		

Appendix D

Tandem Mass Spectrometry
Analytes

Tandem Mass Spectrometry (MS/MS) Analytes

Amino Acid Analytes

Arginine
Citrilline
Glycine
Iso-leucine/Leucine
Methionine
Phenylalanine
Tyrosine
Valine
5-oxoproline

Acylcarnitine Analytes

C0 – free carnitine
C2 – acetyl carnitine
C3 – propanoyl carnitine
C4 – butanoyl carnitine
C5 – pentanoyl carnitine
C5:1 – pentenoyl carnitine
C5-DC – glutaryl carnitine
C5-OH – 3-hydroxy-isovaleryl carnitine
C6 – hexanoyl carnitine
C6-OH – 3-hydroxyhexanoyl carnitine
C8 – octanoyl carnitine
C8-OH – 3-hydroxyoctanoyl carnitine
C10 – decanoyl carnitine
C10:1 decenoyl carnitine
C12 – dodecanoyl carnitine
C12:1 – dodecenoyl carnitine
C14 – tetradecenoyl carnitine
C14:1 – tetradecanoyl carnitine
C16 – hexadecanoyl carnitine
C16-OH – hydroxyl-octadecanoyl carnitine
C18 – octadecanoyl carnitine
C18:1 – octadecenoyl carnitine
C18:1-OH – hydroxyl-octadecenoyl carnitine

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).