Welcome

This is the first newsletter of the Iowa Neonatal Metabolic Screening Program (INMSP). The purpose of this newsletter is to raise the awareness of INMSP within the state of Iowa. We will be sharing information about our program, which includes the Metabolic Consultants, Central Screening Laboratory staff, Hemoglobinopathy Consultants, Endocrine Consultants, and the Birth Defects Institute (Center for Genetics) staff within the Iowa Department of Public Health (IDPH). We are a team working together to improve the quality of life for Iowa infants.

Our first newsletter will introduce you to the various staff for the INMSP program. The target audience for the newsletter includes nurse managers, lab managers, physicians, physician assistants, nurse practitioners, midwives, hospitals, and birthing centers.

It’s not just the PKU test...

The Iowa Neonatal Metabolic Screening Program (INMSP) has been referred to as the PKU test. The test is much more than PKU screening. The Iowa neonatal metabolic screen is a blood test for certain treatable metabolic and inherited disorders performed shortly after the infant’s birth. These disorders, some not apparent at birth, could lead to mental retardation or death if untreated. Research shows that very early treatment can prevent or reduce physical effects and brain damage.

Iowa law requires that all infants be screened for the following seven diseases: congenital hypothyroidism, phenylketonuria (PKU), galactosemia, hemoglobinopathies, congenital adrenal hyperplasia (CAH), medium chain acyl-CoA dehydrogenase (MCAD) deficiency, and biotinidase deficiency.

Central Screening Laboratory

The newborn screening section of the University Hygienic Laboratory (UHL) began providing PKU testing services in 1966. In 1980, pilot testing was initiated for galactosemia, maple syrup urine disease (MSUD) and hypothyroidism. In 1983, newborn screening became mandatory for these disorders in Iowa; the UHL was identified as the “Central Screening Laboratory.” In February 1988, hemoglobinopathy screening was initiated statewide. Congenital adrenal hyperplasia was added in 1992. We have been providing medium chain acyl-CoA dehydrogenase (MCAD) deficiency testing using tandem mass spectrometry (MS/MS) methodology since 1999. In July 2002, biotinidase deficiency screening was added to the panel.
UHL routinely provides information and consultation to the IDPH and to the Birth Defects Advisory Committee, IDPH’s advisory committee for genetic services, for improving the program and ensuring the state program continues to provide the most appropriate services for the state and the babies born here.

From left to right are: Laurie Hazelwood, Esther Blanchard, Mike Ramirez, Pam Busick, Dana Hartsock, Kim Patrick, Marcia Valbracht, and Pat Timmins. All laboratory-testing personnel are CLIA certified. Stan Berberich, Ph.D., Genetics, is the program manager responsible for overseeing the INMSP laboratory. Don Simmons, Ph.D., Analytical Chemistry, is a technical consultant responsible for the supervision of development, MS/MS testing, and training of staff. Laurie Hazelwood, BS, MT (ASCP) and Marcia Valbracht, BS, MHA are technical supervisors with a combined 30 years of experience in newborn screening and are co-leaders of the INMSP lab team. Esther Blanchard, AAA, AAS; Mike Ramirez, BS, Microbiology; Dana Hartsock; Kim Patrick, BS, MT (ASCP); and Pat Timmins, BA, are technical personnel with over 50 years of combined experience. Pam Busick has provided 20 years of data entry and secretarial skills to the program. All staff are trained in several testing areas and can rotate when needed.

**Expansion of Newborn Screening**

On April 17, 2003, the Birth Defects Advisory Committee (BDAC) unanimously voted to support expanding the newborn screening using tandem mass spectrometry (MS/MS). The BDAC is comprised of representatives from professional groups, agencies, legislators, consumers, and individuals with an interest in promoting genetic services. Recommendation for expanding the newborn screening was taken to the director of IDPH and approved June 2003.

Iowa currently screens for seven disorders. The expanded screening will use MS/MS to screen for more than 30 additional disorders detectable by this technology simultaneously from a single blood spot specimen.

In the fall of 2001, the Iowa Neonatal Metabolic Screening Program began a pilot project to screen all newborns in Iowa for additional disorders detectable by MS/MS. It has successfully identified two babies with rare metabolic disorders. One of the infants was diagnosed with 3-methylcrotonyl-CoA carboxylase deficiency (3-MCC); the other was diagnosed with very long chain acyl-CoA dehydrogenase deficiency (VLCAD). If the disorders had not been detected, these babies could have become very sick and may have died. Due to early diagnosis through the expanded newborn screening and appropriate management – a bad outcome was prevented!
Metabolic Consultants

This program is a part of the Medical Genetics Division at the University of Iowa Hospitals and Clinics, under the direction of Dr. Val Sheffield. Judy Miller and Pam DeBoer provide follow-up.

Cheryl Stimson and the genetics physicians provide care and management for children and adults with metabolic disorders. This includes direct patient care and care coordination for individuals with PKU, galactosemia and other inborn errors of metabolism. The Metabolic Genetics Clinic at the University of Iowa is the only metabolic clinic in the state of Iowa.

Exciting Change!

**Biotinidase deficiency becomes part of the Newborn Screening Panel**

The Iowa Department of Public Health authorized the Iowa Neonatal Metabolic Screening Program (INMSP) and the University Hygienic Laboratory to begin routine screening and reporting for biotinidase deficiency effective July 1, 2002.

The decision to provide routine reporting was based on recommendations made by the Birth Defects Advisory Committee. A six-month pilot study was conducted by the INMSP to develop testing competency and identify Iowa infants at risk. One infant was identified during the pilot study.

Biotinidase deficiency is an inherited disorder caused by the lack of biotinidase enzyme, which normally enables the body to recycle biotin. Biotin is essential for normal metabolism and, because humans do not synthesize biotin, the ability to use and reuse all available biotin is critical. The onset of symptoms occurs anywhere from two weeks to two years of age. Symptoms include seizures, hypotonia, hearing loss, developmental delay, optic atrophy, laryngeal stridor, ketolactic acidosis, and recurrent infections.

Individuals with biotinidase deficiency are successfully treated with prescribed unbound biotin supplementation. Usually no biotinidase related medical problems occur once supplementation is started. Identified children should be evaluated periodically for vision and hearing problems. It is estimated that screening will identify one to two children every two years, although the incidence could be higher.
Assistance in paying for metabolic formula
Effective November 1, 2002 Iowa added a $3.00 surcharge per infant to the newborn screening fee. The surcharge is to cover the metabolic formula of individuals who have extinguished all other sources of payment and who are financially unable to pay for all of the expenses themselves.

Hemoglobinopathy Consultants
Hemoglobinopathies are disorders of red blood cells. Hemoglobin is the protein present in red blood cells that carries oxygen and gives blood its color. There are different types of hemoglobinopathies of which sickle cell anemia is the most common. There are many medical problems associated with hemoglobinopathies, dependent upon the type and degree of disease.

Dr. Thomas Loew is the director of the Iowa Hemoglobinopathy Newborn Screening and Comprehensive Care Program. He has been director of the program since his arrival at the University of Iowa in 1997. He previously was director of the Hemoglobinopathy Program at Southern Illinois University located in Springfield, Illinois. Pam DeBoer, RN and Myrl Holida PA-C assist him.

DeBoer has been a Pediatric Nurse for 16 years with experience in the Neonatal Intensive Care Unit, private practice and Genetics. She is responsible to ensure that the patients identified by the newborn screening program are confirmed and referred for proper treatment. She is also active in other aspects of the program. Holida has been a physician assistant for 15 years in Pediatric Hematology/Oncology/Bone Marrow Transplantation. He will assist Dr. Loew with the comprehensive and direct patient care aspect of the Hemoglobinopathy Program. Holida will also provide monitoring of the chronic transfusion program and other areas of patient care required to meet the complex needs of this patient population. The program works closely with the physicians located at Blank Children’s Hospital in Des Moines, Iowa, who provide comprehensive care for those patients living in that region of the state.

The Iowa Hemoglobinopathy Newborn Screening and Comprehensive Care Program Consultants at the University of Iowa, from left to right: Dr. Thomas Loew, MD; Pam DeBoer, RN; and Myrl Holida, PA-C.
**Endocrine Consultants**

In addition to providing consultation to the INMSP, the endocrine consultants provide care to approximately 2,000 patients with endocrine disorders and 600 patients with diabetes at the Children’s Hospital of Iowa. The consultants are happy to serve as a resource for the INMSP, and welcome the opportunity to interact with Iowa health care providers.

**Quarterly Reports on INMSP**

Quarterly reports on the newborn metabolic screening will be issued to inform facilities of the follow-up for infants identified at the facility as not receiving a newborn metabolic screen. It will show how the facility compares to the state statistics to ensure the infants are being screened.

The reports will be sent to each facility and will include the following: number of newborn metabolic screening specimens submitted to the central laboratory; number of rejected specimens; number of specimens collected prior to 24 hours; it will compare the date of the specimen collection and date specimen received in lab; number of disorders confirmed; number of newborns identified not to have a screen at the facility and why (i.e. deceased, transferred, missed, signed a waiver).

**INMSP Training**

If you would like training or education from the central screening laboratory, metabolic consultants, hemoglobinopathy consultants, endocrine consultants, or the Center for Genetics staff - contact them and they will tailor the training to your group.

Contact information for each entity is found within the newsletter. CEU’s may be available at the training.
HIPAA

Health Insurance Portability and Accountability Act of 1996
The Iowa Neonatal Metabolic Screening Program (INMSP) is a program of the Iowa Department of Public Health. The University Hygienic Laboratory (Central Laboratory) is the designated newborn screening laboratory. Personnel within the University of Iowa Department of Pediatrics, Divisions of Genetics, Endocrine and Hematology provide consultation and follow-up for INMSP.

INMSP provides comprehensive newborn screening services for hereditary and congenital disorders pursuant to Iowa Code chapter 136A and 641 Iowa Administrative Code (IAC) Section 4.3. These provisions of law are not preempted by HIPAA. A hospital, clinic, or health care provider is not required to obtain consent or authorization from a patient or parent prior to releasing information to Iowa Neonatal Metabolic Screening Program Personnel.

The table below lists current INMSP personnel. The program personnel are diligent in upholding the highest confidentiality practices.

<table>
<thead>
<tr>
<th>University Hygienic Laboratory</th>
<th>Endocrinology Follow-Up Consultants</th>
</tr>
</thead>
<tbody>
<tr>
<td>Newborn Screening Laboratory</td>
<td>Consultants</td>
</tr>
<tr>
<td>P.O. Box 1803 Des Moines, IA</td>
<td>University of Iowa</td>
</tr>
<tr>
<td>50306-1803 (515) 243-0141 Fax:</td>
<td>Division of Pediatric</td>
</tr>
<tr>
<td>(515) 243-307 Esther Blanchard,</td>
<td>Endocrinology Room 2832 JCP</td>
</tr>
<tr>
<td>Kim Patrick, Pam Busick, Pat</td>
<td>Iowa City, IA 52242 (319) 356-2838</td>
</tr>
<tr>
<td>Timmins, Dana Hartsock, Mike</td>
<td>Mavis Rike, Eva Tsalikian,</td>
</tr>
<tr>
<td>Ramirez, Laurie Hazelwood,</td>
<td>Patricia Donohoue, Michael</td>
</tr>
<tr>
<td>Marcia Valbracht</td>
<td>Tansey</td>
</tr>
</tbody>
</table>

| Iowa Dept of Public Health      | Metabolic Follow-Up Consultants  |
| Center for Genetics 321 12th    | University of Iowa Division of   |
| Street Lucas State Office       | Medical Genetics                 |
| Des Moines, IA 50319 Dawn Mouw  | Room 2605 JCP                    |
| (515) 242-5593 Tammy O’Hollelarn| Iowa City, IA 52242 (319) 356-2674 or (319) 353-6131 |
| (515) 281-7642 Tonya Diehn      | Judy Miller, Pam DeBoer, Adam    |
| (515) 281-7584                   | Kanis, Marcia Willing, Kim      |
|                                 | Keppler, Val Sheffield           |

| Hematology Follow-Up Consultants| |
| University of Iowa Division of  |
| Pediatric Hematology Room 2518  |
| JCP Iowa City, IA 52242 (319)   |
| 356-1400 Pam DeBoer, Myr1        |
| Holida, Thomas Loew              |

Did you know...
Newborn screening is a state public health activity. The federal government requires each state to be responsible for planning and implementing their newborn screening program. State newborn screening programs vary from four to 36 disorders that are screened within the state. Most states screen for eight or fewer disorders.
Newborn Screening Quiz

The newborn screening quiz is an opportunity to test your knowledge about newborn screening. The answers to the quiz are found on page 8.

1. **In order for the PKU results to be accurate, a baby must be at least 24 hours old.**  True  False

2. **Antibiotics interfere with the PKU results.**  True  False

3. **A baby’s newborn screen should be obtained prior to a transfusion, even if they are less than 24 hours old.**  True  False

4. **Certain medications taken during pregnancy may cause an inaccurate result for the congenital adrenal hyperplasia (CAH) test.**  True  False

5. **Results of an initial neonatal screen reveal a borderline elevated TSH level. The infant had received a blood transfusion shortly after the initial neonatal screen was collected. You must wait 8 weeks before repeating the neonatal screen.**  True  False

6. **Sickle cell anemia is the most common type of hemoglobinopathy.**  True  False

7. **Hemoglobinopathies are ONLY found in certain racial or ethnic groups.**  True  False

8. **Specimens must be dried in a horizontal position.**  True  False

9. **Blood transfusions can cause serious disorders to be missed.**  True  False

10. **Specimens should be closely inspected after drying, and recollected if the specimen is of questionable quality.**  True  False

11. **Store collection forms flat, preferably beneath a stack of books to keep them from curling, prior to use.**  True  False

12. **Specimens should be batched and held to save postage.**  True  False

13. **Specimens must be mailed within 24 hours of collection.**  True  False

14. **The lancet used in the heel stick method should be 3.0 mm in length or less.**  True  False

15. **Newborn screening is an optional test.**  True  False

www.idph.state.us/genetics

The Birth Defects Institute (Center for Genetics) web site is packed full of information for parents, consumers and health professionals. The web site includes a practitioner’s manual, parent page, administrative rules, resources, educational materials, and contacts. Visit it often!
### Quiz Answers

1. **True.** In order for the PKU results to be accurate, a baby must be at least 24 hours old.

2. **False.** Antibiotics do not interfere with the PKU results.

3. **True.** A baby’s newborn screen should be obtained prior to a transfusion, even if they are less than 24 hours old.

4. **True.** Steroid therapy administered to the mother during pregnancy can interfere with CAH test results.

5. **False.** Other than the risk of hemodilution in a low birth weight infant, there is no reason to delay repeat testing for congenital hypothyroidism.

6. **True.** Sickle cell anemia is the most common type of hemoglobinopathy.

7. **False.** Hemoglobinopathies are found in all racial or ethnic groups.

8. **True.** Specimens must be dried in a horizontal position.

9. **True.** Blood transfusions can interfere with the correct interpretation of certain disorders, i.e., Galactosemia, Biotinidase and Hemoglobinopathies.

10. **False.** Specimens should be closely inspected after drying and recollected if the specimen is of questionable or inadequate quality.

11. **False.** Always store collection forms on their side in the original packages. Be careful to never allow the paper to be compressed.

12. **False.** Specimens should NEVER be held for any reason. They should ALWAYS be mailed within 24 hours of collection.

13. **True.** Specimens MUST be mailed within 24 hours of collection.

14. **False.** The lancet should be less than 2.0 mm according to NCCLS guidelines.

15. **False.** Newborn screening is a mandatory test according to state law.

### Fun Fact

Newborn Screening Collection Form: Do not use pens with gel or glitter ink.

**Problem** – The glitter will contaminate the specimen.

**Cause** - When the ink dries the glitter becomes loose.

**Result** - If glitter is on the blood specimen the specimen is unsatisfactory for testing.

### Center for Genetics

The Birth Defects Institute (Center for Genetics) was established within the Iowa Department of Public Health in 1976. The Iowa Neonatal Metabolic Screening Program, a program within the Center for Genetics, provides administrative oversight for the statewide newborn screening laboratory, follow-up, consultative and educational services. The program is a partnership between the Iowa Department of Public Health, University Hygienic Laboratory, and the University of Iowa Hospitals and Clinics.

The center currently employs four staff: Tonya Diehn, State Coordinator for Genetic Services; Tammy O’Hollearn, Community Health Consultant; Sherry Smith, Secretary, and Dawn Mouw, Program Planner. Dawn currently matches all Iowa birth certificates to the newborn screening results. She performs the necessary follow-up with the birthing facilities’ when a potential missed screen is identified. Early detection allows for the prevention or reduction of symptoms and ultimately saves babies’ lives.