Hello and welcome to the Heel Stick News. This edition includes information from Iowa as well as North Dakota. Our plan is to distribute this newsletter to EVERYONE including medical professionals, parents, families, communities and legislators. This will bring news, reports and resources from this Regional Newborn Screening Community. Our goal for this newsletter is to enhance awareness of “Newborn Screening.”

Since this newsletter has taken a “hiatus” of sorts, we decided to feature this newsletter as “Newborn Screening: Past, Present and Future.” We sure do welcome your suggestions and/or comments to the contents of this and future newsletters. Please contact me at bschweit@nd.gov.

Barb Schweitzer, RN

About Newborn Screening

Newborn screening is the practice of testing every newborn for certain harmful or potentially fatal disorders that aren't otherwise apparent at birth.

Many of these are metabolic disorders (often called "inborn errors of metabolism") that interfere with the body’s use of nutrients to maintain healthy tissues and produce energy. Other disorders that screening can detect include problems with hormones or the blood.

In general, metabolic and other inherited disorders can hinder an infant's normal physical and mental development in a variety of ways. And parents can pass along the gene for a certain disorder without even knowing that they're carriers.

With a simple blood test, doctors often can tell whether newborns have certain conditions that could eventually cause problems. Even though these conditions are considered rare and most babies are given a clean bill of health, early diagnosis and proper treatment can make the difference between lifelong impairment and healthy development.

Newborn Screening: Past, Present and Future

In the early 1960s, scientist Robert Guthrie, PhD, developed a blood test that could determine whether newborns had the metabolic disorder phenylketonuria (PKU). People with PKU lack an enzyme needed to process the amino acid phenylalanine, which is necessary for normal growth in kids and for normal protein use throughout life. However, if too much phenylalanine builds up, it damages the brain tissue and can eventually cause substantial developmental delay.

If kids born with PKU are put on a special diet right away, they can avoid the developmental delay the condition caused in past generations and lead normal lives.

Since the development of the PKU test, researchers have developed additional blood tests that can screen newborns for other disorders that, unless detected and treated early, can cause physical problems, developmental delay, and in some cases, death.

Some experts want more evidence that early detection of every disease tested for will actually offer babies long-term benefit. Equally important, parents may not want to know ahead of time that their child will develop a serious condition when there are no medical treatments or dietary changes that can improve the outcome. And some questions about who will pay (states, insurance companies or parents) for it have yet to be resolved.
Newborn Screening: Past, Present and Future

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The American Academy of Pediatrics (AAP) and the federal government's Health Resources and Services Administration formed a task force of experts to examine these issues and recommend next steps. Their report identified some flaws and inconsistencies in the current state-driven screening system and proposed the following:

- All state screening programs should reflect current technology.
- All states should test for the same disorders.
- Parents should be informed about screening procedures and have the right to refuse screening, as well as the right to keep the results private and confidential.
- Parents should be informed about the benefits and risks associated with newborn screening.

If you're concerned about whether your infant was screened for certain conditions, ask your child's health-care doctor for information about which tests were done and whether further tests are recommended.

Iowa Newborn Screening From the Past to the Present
By Esther Blanchard and Pat Timmins

We believe we were selected to write this article because we are the “oldies” on the staff. We’re not convinced this is a compliment. Esther has worked in the newborn screening lab since 1980 and Pat since 1990. I tell people Esther was just a baby when she started, and to this day she still looks 29!

PKU analysis was being done in the University Hygienic Lab located in Iowa City, Iowa, beginning in 1966. In 1979, it was moved to the Wallace building in Des Moines. The newborn screening staff consisted of a supervisor, Jan Susanin and one other employee, along with a couple of cross-trained employees (including Esther). Jan Susanin continued as supervisor of the program until her retirement in November, 1999.

In 1990, the newborn screening hemoglobin section received recognition for discovering two unknown hemoglobins, Hb-Davenport and Hb-Iowa. In the last couple of years, we identified a baby having one of these rare hemoglobin disorders.

By 1992, the Hygienic Lab in Des Moines was starting to outgrow its britches, so the newborn screening section was moved to 2nd Avenue. It was a rental property, but gutted and designed to our liking – even with pink and blue walls!
Newborn Screening from the Past to the Present  
*(continued from Page 2)*

Barely had we settled into our new lab when we were hit with the 100-year flood of 1993. We found our lab nearly surrounded by water, yet no running city water to be found. All neighboring businesses were under water. That evening, the staff and their families arrived at the lab to move all items off the floor and to “higher ground.” One of our staff members volunteered to spend the next couple of nights in the lab and call if the water threatened to enter the front door. It came within inches of our back door but never succeeded in entering through the front door. Luckily we did not have to implement our evacuation plan. The main lab in Iowa City was able to provide us with carboys of de-ionized water, and we retrieved other water from the “bladders” established throughout the City by the Army Corps of Engineers and being manned by military personnel. For personal hygiene, we had two choices: drive to a nearby kybo or flush the toilet with a bucket of water once everyone had used it. You can imagine what the preferred option was!! Twice we were evacuated due to toxic barrels floating in the back of our building.

Even with these hardships, the staff continued to test and provide results on a daily basis.

After 13 years in the same place, it was now time to put all staff under one roof. In May, 2005, we moved into a new building in Ankeny. The rental property had to be returned to its prior condition, which meant removing all lab cabinets, walls, etc. The contractor who purchased the interior cabinets allowed the newborn staff to each have one set of benches, if so desired. Pat placed a set in her lake home garage, so a piece of the lab continues to be a part of her life. After we left, it was remodeled into a restaurant where a few of us have eaten; once eating in the prior hemoglobin area and once in the log-in area.

Again as we became comfortable in our new lab, another significant turn of events occurred in the fall of 2005 – **HURRICANE KATRINA**. Overnight we saw our workload triple as we began testing Louisiana babies. Staff members from other departments of the lab (Iowa City and Ankeny) volunteered for a “quick cross-train” course in newborn screening and helped us until we were able to hire temporary staff. During that time, the hemoglobin section (where Pat was working) was running 12 to 16 gels each day. Newborn screening staff was working overtime to meet our goals. Once the Louisiana staff was reassembled, they too came and helped us. It was a challenging time for all; but with the great support of others, we got through it and made some new friends.

About the time we thought we had seen the last of change for awhile, a courier service and night shift was implemented. The night shift would begin receiving and running the initial tests on the specimens and the day shift would check results, repeat specimens with abnormal test results on a duplicate run, and report abnormal test results to consultants.

The courier service began as a transport for newborn screening specimens, but later was expanded to include delivery of other Iowa specimens (i.e., mumps and H1N1) that were being tested in the Iowa City lab. We now receive the newborn specimens on the same day or next day after collection from Iowa, North Dakota and South Dakota facilities, thus allowing almost immediate results and treatment for our precious newborns.

So, as you can see through the timeline, what started out as a PKU test is now a panel of tests. What started as a program with two staff members has now grown to a staff of 17, under Dr. Stan Berberich, program manager and Marcia Valbracht, supervisor.

Despite the transitions and challenges we have faced over the years, Esther and Pat agree that overall it has been a rewarding and fun job. We know we’ve made a positive difference in many lives.
NEWBORN SCREENING ... Past to Present
by Pam DeBoer, RN

In 1965, the Iowa state legislature enacted a law that recommended testing infants for phenylketonuria (PKU). The Iowa University Hygienic Lab (UHL) began providing PKU testing for Iowa in 1966.

Below is the year that each specific disorder was added:

1981 – Galactosemia, MSUD and congenital hypothyroidism
1988 – Hemoglobinopathy screening
1991 – Congenital adrenal hyperplasia
2001 – Medium chain acyl-CoA dehydrogenase deficiency
2002 – Biotinidase deficiency
2003 – Expanded panel disorders
2006 – Cystic fibrosis and the implementation of the courier service

Iowa’s UHL also has contracted to processes newborn screens for the states of South Dakota and North Dakota. Here are the years they started:

1992 – North Dakota
2007 – South Dakota

Over the past seven years the Newborn Screening Program has grown and made lots of positive changes. Here are the ways our program has grown:

**University Hygienic Laboratory:**

- In May 2005, the UHL moved from its Des Moines, Iowa, location into a new building in Ankeny, Iowa.
- With the addition of the courier service, UHL added additional staffing to cover first and third shifts seven days a week.

**Weekend/Holiday on-call:** The Short term follow-up case manager nurses have covered weekends and holidays since the implementation of the 24 hour courier service.

**Short Term Follow-Up (STFU):** The addition of two nurses to the Iowa program has helped to cover the follow-up needed for North Dakota and South Dakota. We have a total of four case managers and a Nursing Supervisor. When Iowa took over all of the newborn screening follow-up for North Dakota (January 2009), we added a regional newborn screening coordinator for Iowa and North Dakota who resides in North Dakota.

**Protocols/SOP:** These are in place for the UHL and STFU protocols/SOP for which we follow. These are updated yearly and are ongoing with updates as changes occur in the Newborn Screening Programs.

**Medical Director of Newborn Screening:** Dr. Val Sheffield is the interim medical director of the Newborn Metabolic Screening Program. Dr. Sara Copeland was the medical director of newborn screening from July 2004 to October 2009. She had made the program one of the best in the nation. We are now in the process of searching for a new medical director.

**General Contact Phone Numbers:**

- University Hygienic Laboratory, 515-725-1630
- Short Term Follow-Up, 866-890-5965 or 319-384-5097
The sunlight is waning as I groggily ease myself out of bed. Catching a glance at my clock, I realize it is 4:30 in the afternoon. By this time, the majority of the world is wrapping up their workday and heading home. My day is just beginning, much as it has for the past year and a half. To sleep while the sun is beaming through any crack in the wall it can find is, shall we say, an acquired taste, but not impossible.

Iowa Newborn Metabolic Screening Program implemented a night shift in February 2006; a few months after Iowa began to take on Louisiana specimens due to Hurricane Katrina. At the time I was still in college, so I am no expert on "ye olden days." I began as a temp in the lab in October 2007, at the tail end of the testing for Louisiana. As I came into the lab each morning, I recall seeing paperwork on the counters, machines still processing specimens and tiny bits of evidence that led one to believe someone had been there working. The people of the night shift intrigued me. The only proof of their existence was the notes left by them and their initials written on various pieces of paper. I eventually met these "ghosts" in all-lab staff meetings.

I became one of the "ghosts" in June 2008. Armed with caffeine and the fear every new kid in school must feel, I slowly adjusted to life in the PM. It's kind of a trip to think I come into work one day and I'm actually leaving the next. It also leads to my never actually knowing what day it is.

Through much trial and error, my predecessors established a fairly routine schedule for processing each night. Our work begins as soon as we walk into the door: preparing the multi-punches and testing plates, logging in specimens, and reading over communication books. The processing of tests is largely based upon what time our couriers arrive. We are fortunate to have a group of drivers that are dedicated to their jobs and see to it that our specimens arrive in a consistently timely manner every night, even through the worst of Midwest weather patterns.

In October 2008, we implemented a new scheduling system to ensure staff members had adequate time to spend with their families and friends. Each staff member works nine-hour shifts for five nights followed by four nights off. This pattern stays in place over weekends and holidays in order to maintain optimal staffing numbers. Working through a time when such scheduling wasn't an option and staffing was a little sparse, it has been extremely beneficial. Our 5-4 rotations allow us a day to get caught up on sleep, two days to function with the "real" world and a day to acclimate ourselves into working hours again. It's more or less equivalent to a normal weekend, albeit much of our time off falls in the middle of the week.

This isn't to say that our shift doesn't have its own unique set of challenges to overcome. Our operating hours are not conducive for consulting outside technical support. Our supervisors are never more than a phone call away (even at 3 am and I do speak from experience on that), but there are times when an instrument goes down or we encounter an error that simply cannot be dealt with that night. Fortunately, this doesn't happen often, and the day shift is always quick to act when they arrive in the morning. Needless to say, we've all become pretty adept in thinking on our feet in order to prevent any delay in processing. As the winter months approach, it's also safe to say that inclement weather will play a hand in the workflow of our shift. Working a 10-hour shift or a little more isn't unheard of around here — if working in newborn screening teaches you anything, it's that this is not a 9 to 5 job. Well, at least not in the traditional sense.

However, with doing the bulk of the testing at night, we are all fortunate to be at the heart of a very dynamic aspect of public health. The rumblings of what's to come in tandem mass spectrometry (i.e., lysosomal storage disorders) are exciting and yet present a challenge to our already large testing panel. In April of this past year, we began our pilot testing for SUAC — an analyte typically elevated in Tyrosinemia I. The addition of one marker added an entire hour to our processing of plates in TMS. Such projects call for reevaluation of our processes on the night shift and how we can be more efficient. With companies and research teams producing new ways of testing for markers all the time, we are sort of in a state of flux just trying to keep up. In all honesty, I feel as though things change every time I return from time off. I don't consider it an inconvenience — it's the nature of the work and a sign to me that we're not allowing ourselves to be "okay" with the status quo.

While our night shift has become more refined with time, there is still a lot that can be done in order to improve our program. Education is an ongoing necessity, as we can make hospitals more aware of the value of our courier service and decrease time between the collection of the specimen and the arrival at our laboratory. Overall promotion of newborn screening is another key to our success: it shouldn't be a secret as to what we're doing and parents should be at least somewhat familiar with our mission and goals. I don't need recognition for what I do personally, but I do think the public would be pleased to know of the dedication of our staff to making the lives of their newborns that much better.

I can honestly say that I enjoy performing my job responsibilities. There are days when I am fully aware of the limitations that come with working on an opposite schedule of the rest of the world. I believe you can make any job as good or as bad as you want it to be, and I find what I do to be extremely rewarding, even if to some I'm only known by my initials on a piece of paper.
Creating Capacity in a Rural State

A clinic that affects patients' lives, access to care and public policy

Alan Kenien's (BMF'90) goal was to “become a resource in the diagnosis and management of inborn errors of metabolism.”

Kenien had always had an interest in children with metabolic disorders, fitting them into his schedule as needed, but the fellowship – and, later, the help of a proposal writer at his Fargo practice – allowed him to create a multi-disciplinary clinic serving children in North Dakota and neighboring states.

“The multidisciplinary approach is important to a rural area,” Kenien said. “Many of the patients travel long distances for treatment, and having a multidisciplinary clinic where they can see several providers in one day makes it much easier for them.”

Initial state funding for the Fargo clinic has created access to medical care for patients from three states and diverse backgrounds. Kenien said, “I work closely with other specialists and to keep the clinic viable to all who need it. I am not a specialist in all inborn metabolic disorders,” but now many can be treated close to home. Before the clinic was established, many families had to drive to the Mayo Clinic or to the Twin Cities for care.

Kenien’s work also has affected North Dakota state policy regarding reimbursement for a special formula for infants with PKU (a metabolic disorder that can cause mental retardation if not treated early). He led an effort to make legislators aware of the costs of not treating the affected infants. “We spread the word that if these children didn’t get their formula, they were going to be huge tax burden to the State of North Dakota. Although the formula is very expensive, it prevents the need to institutionalize these children as they get older and allows them to mature into healthy and tax-paying adults.” (As a result of Kenien’s campaign, the legislature rescinded its rejection of payment for the formula.

The 1990 Bush Medical Fellowship was a turning point for Kenien. Without it, he observed, “I would not have had the confidence or training or the experience” to create the clinic.

A physician specialist familiar with his work noted that education – of both physicians and patients – is a key strategy Kenien employs: “He provides education about the metabolic disorders and what they mean, and how they should be managed and what treatment is.” The specialist also spoke of Kenien’s impact on physicians in North Dakota by serving as the state’s consultant for the newborn screening program. “He took over more responsibility for metabolic disease in the state. There isn’t anyone specifically trained for that here. It’s providing improved care and management for children with metabolic disorders. It means that kids will be diagnosed earlier and appropriate treatment will be provided earlier.”
Center for Congenital and Inherited Disorders (CCID)

Iowa legislation mandates that the Iowa Department of Public Health (IDPH) develop and administer the state's policy with respect to the conduct of scientific investigations and research concerning the causes, prevention, treatment and cure of birth defects. The Center for Congenital and Inherited Disorders (CCID) was established within the department in 1976 to initiate, conduct and supervise genetic investigations and research in order to provide for the protection and promotion of the health of Iowans. Since its creation, the Center for Congenital and Inherited Disorders, in partnership with families, health and human service providers and communities, advances the health and well-being of children with genetic conditions and special health needs. The Center also works with the University of Iowa and health-care providers throughout the state to develop programs that have provided Iowa with state-of-the-art genetics health care. The programs of the Center for Congenital and Inherited Disorders address all steps of the life cycle: prenatal, neonatal, pediatric and adult.

The Congenital and Inherited Disorder Advisory Committee is composed of subject matter experts and parent and consumer representatives. The committee provides advice to the Iowa Department of Public Health on matters of genomic programming.

The Hemophilia Advisory Committee provides program guidance and recommendations to the IDPH regarding hemophilia and other bleeding or clotting disorders programming.

As the state genetics coordinator, Kimberly Noble Piper coordinates the state’s public health genomic programs.

These programs include:

- The Iowa Neonatal Metabolic Screening Program (INMSP) – Dr. Val Sheffield, interim medical director; Dr. Stanton Berberich, INMSP lab director
- The Maternal Prenatal Screening Program (IMPSP) – Dr. Roger Williamson, medical director; Dr. Stanton Berberich, IMPSP lab director
- The Iowa Registry for Congenital and Inherited Disorders (IRCID) – Dr. Paul Romitti, director
- The Regional Genetics Consultation program – Cathy Evers, program manager
- The Neuromuscular and Related Disorders program – Dr. Katherine Mathews, medical director

The CCID also administers discretionary grants that include:

- The Iowa Stillbirth Surveillance Project
- The Iowa Newborn Screening Surveillance Project
- The Iowa Family Participation Project

In addition, the CCID promotes family health history initiative and stillbirth awareness and prevention activities.
Quality Specimens

All procedures in the newborn metabolic screening process reflect a commitment to produce accurate, clinically useful results. No procedure is more important than the first – specimen collection. Every specimen arriving at the neonatal metabolic screening laboratory is evaluated before testing. If an unacceptable specimen is submitted, a request for a new specimen will be made, which causes delays that could have serious medical consequences for an afflicted child.

Last year, specimens for 289 babies were rejected because of poor quality. The second specimen for 34 of these babies were never collected and sent in for testing. Filter paper with correctly collected blood spots contains a calculated amount of blood. The laboratory uses the blood spots to perform quantitative tests. Poor quality specimens will affect the validity of the test.

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<tr>
<th>SPECIMEN PROBLEM</th>
<th>POSSIBLE CAUSE</th>
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| Quantity Insufficient | • Removing the filter paper before blood has completely filled the circle.  
| | • Using a blood drop that is too small. |
| Appears Clotted Or Layered | • Touching same circle to blood more than one time.  
| | • Filling the circle on both sides of the filter paper.  
| | • Allowing the blood in a capillary tube to coagulate before applying to filter paper. |
| Appears Scratched | • Drawing blood on the paper with a capillary tube.  
| | • Touching the heel to the filter paper. |
| Appears Contaminated | • Filter paper has come into contact with hand, or substances such as alcohol, water, lotion, etc. |
| Shows Serum Separation | • Alcohol was not wiped from the puncture site before making the skin puncture.  
| | • Blood in the capillary tube coagulated before applying it to filter paper.  
| | • Squeezing around the puncture site. |
| Supersaturated | • Applying excess blood to filter paper.  
| | • Applying blood to both sides of filter paper.  
| | • Filling the capillary with too much blood before applying. |

All of the information on the specimen collection form must be entered and is critical to the interpretation and reporting of the results. The laboratory depends on the information submitted on the specimen collection form. It is very important that the handwriting on the specimen collection form is legible, complete and accurate. Currently, if our laboratory receives specimen forms with inaccurate or missing information, a fax is sent to the submitting facility requesting additional information. The submitting facility contact person is asked to correct and complete the information and fax it back to the newborn screening laboratory. This often delays the time it takes to get the screening results.

Educational materials are available to assist birthing/submitting facilities with the specimen collection process. The educational materials include a video about specimen collection. Upon request, the Iowa Neonatal Metabolic Screening Program staff can provide an on-site education and training session. Contact the central lab at 515-725-1630.
Lab Quality Improvement

The following checklist is for your newborn metabolic screening program. It is an opportunity to review the collection and submission of newborn metabolic screening specimens in your laboratory.

How are your newborn screening (NBS) forms monitored?
- Are they stored in a clean dry place in a vertical position?
- Is the supply monitored to ensure that the availability of forms is within the expiration date?

Who completes the Newborn Metabolic Screening Forms?
- Does the nursery or lab collect data to ensure ALL fields are completed, legible and accurate?

Is your facility doing adequate documentation?
- Is there a log in the nursery or lab documenting each newborn’s date and time of birth and blood collection?
- Does your facility use the log to track the specimens until the results are received?
- Does your facility keep the carbon copy of the NBS form, and is it viewed for completeness and legibility?
- Is there someone at your facility to track unsatisfactory specimens?
- Does your facility have a system set up to guarantee that ALL newborns are screened prior to discharge?

Is parent education conducted?
- Is newborn screening education started during the perinatal period?
- Does the nursery or lab give parents the free NBS pamphlet provided by the program?

Are staff who perform heel sticks at your facility properly trained?
- Are they properly trained in the collection procedure on filter paper?
- Are they able to describe a satisfactory specimen?
- Are they able to describe an unsatisfactory specimen?
- Can they list the diseases for which Iowa screens?
- Do you track unsatisfactory specimens back to the individual who collected it and retrain as needed?

Are specimens handled and mailed properly?
- Are specimens dried for at least three hours away from heat and sunlight on a horizontal, level, non-absorbent surface, such as a drying rack, prior to mailing?
- Are all specimens mailed within 24 hours of collection?
- Are steps taken to avoid subjecting the specimens to heat and humidity prior to mailing?
- Has your facility assigned someone to review each newborn screen prior to mailing to make sure the form is complete, legible and satisfactory?

Why does baby’s weight at the time of specimen collection matter?
- The test interpretation for CAH is weight dependent.
- The weight of a baby at the time of specimen collection is required to calculate the screening result.
How Can I Help You?

It has been said that education is the key to opening the world. Education is also the key to opening and understanding the world of newborn screening. It starts with educating the newborn screening follow up and laboratory personnel with processes, operating procedures, staffing and budgets. Protocols become developed and redefined over time. Education then needs to take place in the community with providers, birthing facilities, parents, consumers, legislators and so on.

How can newborn screening educate you? That depends on your needs. Our program can tailor education to meet your needs and resources. We are available by phone, fax, internet or in person to answer your questions.

Here is an interesting education point. Did you know the Iowa Neonatal Metabolic Screening Program (INMSP) operates short term and long term follow-up from Iowa City, Iowa, at the University of Iowa Hospitals and Clinics? The initial and repeat blood spot cards are sent and processed at the University Hygienic Laboratory in Ankeny, Iowa. While follow-up and the laboratory are parts of the same program, they exist in two different cities approximately 106 miles apart.

The next time you have a question about newborn screening, let us know. Here’s how:

Iowa Neonatal Metabolic Screening Program (INMSP)
100 Hawkins Drive - S267 CDD, Iowa City, IA 52242
Phone: 319-384-5097, 866-890-5965 toll free
Fax: 319-384-5116
E-mail: Contact the nursing supervisor at kimberley-turner@uiowa.edu OR Contact the laboratory supervisor at marcia-valbracht@uiowa.edu

STICKY NOTES: TO USE OR NOT TO USE, THAT IS THE QUESTION

Multifunctional uses:
- Make quick reminders
- Attach to files
- Keep track of tasks
- Mark a place in a book

While “sticky notes” or as some are called “post it notes” have wonderful uses for all of us, we request that sticky notes not be used on the newborn screening blood spot card. It is so easy for that note to detach from the card and be lost, or to attach itself to another card. Any information not written directly on the specimen card must include the baby’s name and birth date on the same piece of paper to prevent mistakes and errors.

We thank you and appreciate your assistance with this.

Collection Tip of the Quarter

Even though the form says to apply the blood from the reserve side, it really does NOT matter which side you use. However, the filter paper portion of the form should NEVER come into contact with anything except the baby’s blood.

1. Do not let hands or gloves touch the circle part where blood is applied.
2. Do not set the filter paper on the countertop or bench while applying the blood. When the liquid blood touches the countertop, anything on the countertop may contaminate the blood.
3. Do not use anything other than the baby’s blood to fill up circles on a newborn’s filter card. An affected baby may be missed due to a false normal results from someone else’s blood.

"Most folks are about as happy as they make up their minds to be.”
Abraham Lincoln