

TOOLKIT

VERSION 4



Genomics and Newborn Screening is inclusive of the Early Hearing Detection and Intervention Program and Indiana Birth Defects and Problems Registry.



This toolkit serves as a reference guide for those involved with Indiana's state-mandated Genomics and Newborn Screening and its best practices, procedures, and follow-up. Introduction and references to the Indiana Birth Defects and Problems Registry are also included within its content.

Please email questions to the GNBS Program at ISDHNBS@isdh.IN.gov.

Contents

ACRONYMS	1
Mission Statement	2
Stakeholder Roles and Responsibilities	2
Genomics and Newborn Screening Program	3
What is Genomics?	3
Genomics vs. Genetics: What is the difference?	3
Continuity of Operations Plan	4
Newborn Screening Law (16-41-17)	5
What is it?	5
How much does it cost?	5
Religious Waiver for the Newborn Screening (IC 16-41-17-2)	6
Newborn Screening Stakeholders	7
Birth Defects and Problems Registry Law (16-38-4)	8
What is it?	8
How does the IBDPR receive reports?	8
Why is the IBDPR important?	9
IBDPR Medical Chart Abstraction Process and Contacts	9
IBDPR Stakeholders	10
Community Birth Partners	11
ADMINISTRATIVE CODE (410 IAC 3-3-6 Sec. 6)	12
The Three Screens	12
Hearing Screening	13
Pulse Oximetry Screening	13
Heel Stick Screening	14
NBS Log	15
Newborn Screening Lab	16
NOW Courier Information	17
NBS CARD 101	18
NBS CARD: Policy for Addition or Change of Demographic Data	19
RESULTS AND FOLLOW-UP	20
INSTEP	20
Requesting Results	21
Presumptive Positive or Abnormal Heel Stick Results	22

GNBS Community Partners	23
Newborn Screening Best Practices	24
Hearing Screen	24
Pulse Oximetry Screen	26
Heel Stick Screen.....	29
New Conditions.....	29
Heel Stick Results Protocol.....	29
NBS Card Quality Assurance	31
Last Quality Check.....	32
Dried Blood Spot (DBS)	33
Dialogue to Families.....	34
Suggested Statements from Staff to Family	35
Monthly Summary Report (MSR).....	36
MSR 101	37
Exception Entry	38
Explanations of Exception Page	39
Exception Entry Definitions.....	40
Exception Entry for Pulse Oximetry	41
Summary Data Entry	42
Holdovers	43
Tips for avoiding MSR Error	44
Quality Improvement Initiative.....	45
NewSTEPS360.....	45
Birthing Facility Report Cards.....	45
Tools and Resources for GNBS Stakeholders	46

ACRONYMS

AAP – American Academy of Pediatrics

CCHD – Critical Congenital Heart Disease

CHIP – Child Health Information Profile

COOP – Continuity of Operations Plan

DBS – Dried Blood Spot

EHDI – Early Hearing Detection and Intervention

FAQ – Frequently Asked Questions

GBYS – Guide by Your Side

GNBS – Genomics and Newborn Screening

IBDPR – Indiana Birth Defects and Problems Registry

INSTEP – Indiana Newborn Screening Tracking and Education Program

IPQIC – Indiana Perinatal Quality Improvement Collaborative

ISDH – Indiana State Department of Health

MCH – Maternal and Child Health

MCP – Maintaining a Centralized Program

MSR – Monthly Summary Report

NBS – Newborn Screening

PCP – Primary Care Provider

PGG – Perinatal Genetics and Genomics Advisory Board

PID – Patient Identification

QI – Quality Indicator

RR – Religious Refusal

RUSP – Recommended Universal Screening Panel

SACHDNC – Secretary's Advisory Committee on Heritable Disorders in Newborns and Children

UNHS – Universal Newborn Hearing Screen



Mission Statement



*To promote, protect, and improve the health
of identified Hoosier babies with genetic conditions
and birth defects.*



Stakeholder Roles and Responsibilities

Within the GNBS program, we have worked toward identifying and building relationships across the state. If you are identified as being a GNB stakeholder, then you are an essential piece to ensuring the best health outcomes for Hoosier newborns and their families. You are a building block to successful NBS practices for all stakeholders across the state and nationally.

Genomics and Newborn Screening Program

Genomic medicine in neonatology, through newborn screening, allows for the rapid identification of various genetic structural deviations that result in disease that would otherwise go undetected until symptom onset. In today's advancing medical science, detection of disease through genome sequencing is becoming more prevalent, not only allowing for the identification of disease, but also providing more effective treatments and in some cases, a cure.

What is Genomics?

Genomics is the medical practice of using an organism's DNA to understand its physiological processes. This practice applies to all living organisms, allowing for a comprehensive knowledge of how organisms interact with and impact each other. In medical science, genomic medicine is considered a subset of precision medicine using a person's genetic coding to identify diseases impacting healthy human physiology. Genomics is applicable to every medical field; however, there are a few well-known and widely utilized genomic practices in today's medical fields, including oncology, pathology, infectious disease and pediatrics.

Identifying disease using genomic practices can be done a number of ways, including whole genome sequencing, whole exome sequencing and targeted gene sequencing. Whole genome sequencing is a scan of the full set of DNA's base pairs (adenine, thymine, cytosine and guanine or ATCG) for structural deviations, whereas whole exome sequencing is a scan of only protein-coding RNA. Exome sequencing is a less broad view than genome sequencing, enabling faster identification of diseases with known biomarkers. However, a targeted gene sequencing approach is most efficient and affordable for diseases with well-established locations for genetic mutations or deletions.

Genomics vs. Genetics: **What is the difference?**

Genomics focuses on physiological processes dictated by genetic coding. Genomics is a broader view of how genes interact and then how to use that interaction to identify and treat disease. Genetics is more narrowly focused on the coding process and DNA/RNA structure building. The differences are slight, but in short, genomics is a field of study and medical practice that uses genetics to understand physiology, health and disease.



Continuity of Operations Plan

A Continuity of Operations Plan (COOP) as defined in the National Continuity Policy Implementation Plan and the National Security Presidential Directive 51/Homeland Security Presidential Directive 20 is an effort to ensure that primary mission-essential functions continue to be performed during a wide range of emergencies, including localized acts of nature, accidents and technological or attack-related emergencies. The critical business functions your COOP plans for are those that must be:

- Operational no later than 12 hours after activation of COOP
- Capable of maintaining sustained operations for up to 30 days

In the case of any emergencies, it is critical that you refer to your facility's own COOP.

In the timeframe that a COOP is put into action, or other barriers to care occur, the Indiana GNBS program and designated NBS lab staff can be contacted at the provided methods of contact here:

<https://www.in.gov/isdh/27866.htm>

When these COOPs are in review, it is important to have an NBS stakeholder involved to ensure the NBS process can continue to occur in a timely manner.

Newborn Screening Law (16-41-17)

<http://www.in.gov/legislative/iac/T04100/A00030.PDF>

What is it?

Newborn screening (NBS) is a set of **three tests** that aims to detect health conditions in babies that would otherwise go undetected until the baby became symptomatic. This is important as a newborn may look healthy but can have a serious health condition. When left undetected and untreated, these conditions result in severe developmental delays and fatalities. When caught early, the treatments for these conditions can lead to a stronger, healthier development and a longer life.

As of 2018, Indiana's NBS law requires every baby to be tested for 49 different conditions including endocrine disorders, cystic fibrosis, inborn errors of metabolism, hemoglobinopathies, SCID, SMA, critical congenital heart defects and hearing loss. To quickly detect these conditions for early intervention, the following three tests must be completed before the baby leaves the hospital:

1. **Universal Newborn Hearing Screen (UNHS)** detects hearing loss. This can be completed as soon as six hours after birth.
2. **Pulse oximetry** measures pulse rate and oxygen levels to determine heart and lung function. This must be performed between 24 and 48 hours after birth.
3. **Heel stick**, a blood test, is used to screen for genetic conditions that would otherwise go undetected. This must be performed between 24 and 48 hours after birth.

How much does it cost?

The cost of an initial newborn screen is \$100 as of 2019*. These funds go to the lab equipment necessary to screen for and ensure that community resources and medical care are available for the children who test positive for these conditions. Some of the expenses that result from these conditions include special medical foods, prescription medications, laboratory testing, frequent physician visits, specialized therapies and surgeries. So even when a baby's tests are negative, the fee goes to help a family whose baby tested positive and now needs expensive medical treatments.

**Indiana Genomics and Newborn Screening Panel and fee are subject to change each year. Please refer to www.NBS.IN.gov or the Indiana General Assembly for the most recent news.*



Religious Waiver for the Newborn Screening (IC 16-41-17-2)

<https://www.in.gov/isdh/27967.htm>

Indiana has mandated that all newborns undergo newborn screening: hearing screen, pulse oximetry and heel stick.

Per law (IC 16-41-17-2), a family is able to refuse one, two or all three portions of the newborn screening due to religious beliefs. To refuse any portion of newborn screening, the parent/legal guardian must fill out the Religious Waiver form. For a religious exemption, please have the parent/legal guardian clearly fill in, indicate which screen(s) they are refusing to be administered to the infant and sign the form (along with a witness).

 RELIGIOUS WAIVER FOR THE NEWBORN SCREENING PROGRAM State Form 54102 (R2 9-17) INDIANA STATE DEPARTMENT OF HEALTH		ATTACH LABEL HERE OR SEND DEMOGRAPHICS WITH WAIVER.													
<p><i>I have been informed about the Newborn Screening Program for the State of Indiana and have received and read information about the screening tests required by law, but I choose to object to the following being done on my child for reasons pertaining to my religious beliefs:</i></p> <p><input type="checkbox"/> Hearing Screening (for hearing loss) <input type="checkbox"/> Heelstick Screening (for metabolic and endocrine conditions, hemoglobinopathies, and cystic fibrosis) <input type="checkbox"/> Pulse Oximetry (for critical congenital heart disease/defects)</p> <p>Name of Child: _____ Date of Birth: ____/____/_____ Mother's Name: _____ Date of Birth: ____/____/_____ Best Contact Number: _____ Birth Facility/Midwife(ry): _____</p> <p>Signature of Parent _____ Date (month, day, year) _____ Signature of Witness _____ Date (month, day, year) _____</p> <p><i>Please use this form to report Newborn Hearing Screening and Pulse Oximetry when there is a Religious Refusal for the Heelstick Screening (in place of the NBS card).</i></p> <p>Hearing Screening (for hearing loss) Pulse Oximetry (for critical congenital heart disease/defects)</p> <table border="1"><tr><td><input type="checkbox"/> Initial <input type="checkbox"/> Rescreen Left</td><td><input type="checkbox"/> Initial <input type="checkbox"/> Rescreen Right</td></tr><tr><td>Date of Screen ____/____/____</td><td>Date of Screen ____/____/____</td></tr><tr><td>Results <input type="checkbox"/> Pass <input type="checkbox"/> Refer</td><td>Results <input type="checkbox"/> Pass <input type="checkbox"/> Refer</td></tr><tr><td colspan="2">Risk Factors <input type="checkbox"/> Yes <input type="checkbox"/> No</td></tr></table> <table border="1"><tr><td>1st Reading Date of Screen ____/____/____ Time _____ Right Hand ____ Foot ____ <input type="checkbox"/> Pass <input type="checkbox"/> Did NOT Pass</td></tr><tr><td>2nd Reading Date of Screen ____/____/____ Time _____ Right Hand ____ Foot ____ <input type="checkbox"/> Pass <input type="checkbox"/> Did NOT Pass</td></tr><tr><td>3rd Reading Date of Screen ____/____/____ Time _____ Right Hand ____ Foot ____ <input type="checkbox"/> Pass <input type="checkbox"/> Did NOT Pass</td></tr><tr><td colspan="2">ECHO CARDIOGRAM Date of Screen ____/____/____ Time _____ Interpretation: _____</td></tr></table>			<input type="checkbox"/> Initial <input type="checkbox"/> Rescreen Left	<input type="checkbox"/> Initial <input type="checkbox"/> Rescreen Right	Date of Screen ____/____/____	Date of Screen ____/____/____	Results <input type="checkbox"/> Pass <input type="checkbox"/> Refer	Results <input type="checkbox"/> Pass <input type="checkbox"/> Refer	Risk Factors <input type="checkbox"/> Yes <input type="checkbox"/> No		1 st Reading Date of Screen ____/____/____ Time _____ Right Hand ____ Foot ____ <input type="checkbox"/> Pass <input type="checkbox"/> Did NOT Pass	2 nd Reading Date of Screen ____/____/____ Time _____ Right Hand ____ Foot ____ <input type="checkbox"/> Pass <input type="checkbox"/> Did NOT Pass	3 rd Reading Date of Screen ____/____/____ Time _____ Right Hand ____ Foot ____ <input type="checkbox"/> Pass <input type="checkbox"/> Did NOT Pass	ECHO CARDIOGRAM Date of Screen ____/____/____ Time _____ Interpretation: _____	
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Always report religious refusals by submitting the waiver(s) to ISDH by faxing the forms directly to the GNBS Program or by mailing a copy of the waiver(s) with your facility's NBS cards. Note that this does not bypass the requirement of submitting religious exemptions as an exception in your MSR. If one screen is refused, it does NOT change the value in reporting the other two portions of NBS to ISDH.

Note: There are no guidelines for specified religious affiliation. Regardless of the family's beliefs, the ISDH GNBS program needs a signed copy of the Religious Waiver. Ensure the family indicates which screen (hearing, pulse ox and/or heel stick) is being waived for religious purposes. Upon receipt of the signed and indicated waiver, the family can be assured that no further contact regarding GNBS and follow-up will be made.

Newborn Screening Stakeholders

Newborn screening is a public health initiative aimed at identifying conditions that can affect a child's long-term health or survival. Early detection, diagnosis and intervention can prevent death or disability and enable children to reach their full potential.

Those who have a hand in ensuring that all babies born in Indiana receive a valid and timely screen and any follow-up necessary are important to ISDH. As of 2019, Indiana has 87 birthing hospitals, and many birthing centers and midwiferies throughout the state. GNBS follow-up continues into the pediatricians' offices, health departments, specialty groups and others. It is important to recognize the work and maintenance that goes into ensuring all of Indiana's newborns are well taken care of in regards to GNBS. Efforts are spread throughout the community, and each of us has a crucial role to play in the early detection and intervention of these life-threatening and life-altering diseases.

Our screeners, testers, reporters, follow-up and other teams include the following:

- Birthing facility managers
- Registered nurses
- Unit secretaries
- Laboratory technicians
- Medical records staff
- Audiologists
- Nurse practitioners
- Hospital physicians
- Midwives
- Pediatricians
- Medical assistants
- Specialists
- ISDH community partners
- ISDH vendors
- ISDH staff
- Indiana Hospital Association
- AAP, Indiana Chapter
- IPQIC PGG Advisory Board



Birth Defects and Problems Registry Law (16-38-4)

www.BirthDefects.IN.gov

What is it?

The Indiana Birth Defects and Problems Registry (IBDPR) is a population-based surveillance system that seeks to promote fetal, infant and child health. The purpose behind the IBDPR is to prevent childhood development disabilities, enhance the quality of life of affected children and their families and reduce infant mortality. After conditions are diagnosed at birth, follow-up with referral to care can be provided to the children.

The IBDPR law allows the Indiana State Department of Health (ISDH) to gather relevant information for epidemiological and environmental studies as well as informing and providing resources to families impacted by congenital anomalies.

How does the IBDPR receive reports?

The list of reportable conditions is updated annually based on recommendations from the National Birth Defects Prevention Network, the Indiana Perinatal Quality Improvement Collaborative and ISDH. Birthing hospitals are required by law to report all cases of congenital anomalies to ISDH monthly. If the diagnosis does not occur during the initial admission after birth, reports of confirmed cases are to be made to the IBDPR within 60 days of diagnosis. Additionally, a subset of the reportable conditions requires a medical chart abstraction for confirmation. These targeted conditions are of public health importance and are reported annually. Targeted conditions can change based on the initiatives of the MCH programs and emerging public health threats. The IBDPR program receives reports through hospital discharge information, physician reporting and newborn screening.

1. **Hospital reports** are received through a specified file format uploaded into the IBDPR database. The IBDPR program hopes to implement new electronic data standards and submissions to reduce the burden on the reporting hospital staff.
2. **Physicians** are required to report to the IBDPR directly through the Physician Reporting Portal. This includes pediatricians, psychiatrists, psychologists, dentists, midwives, registered or licensed practical nurses, optometrists, podiatrists, chiropractors, physical therapists, local health departments, health maintenance organizations and audiologists.



3. **Newborn screening reports**, such as failed pulse oximetry screens, are reported to the IBDPR for follow-up and confirmation of critical congenital heart defects. All other newborn screening conditions (e.g., metabolic conditions, hemoglobinopathies, endocrine disorders, hearing loss, etc.) are reportable conditions for surveillance purposes.

Information on how to register for the Physician Reporting Portal, reportable condition list and condition descriptions can be found at www.BirthDefects.IN.gov.

Why is the IBDPR important?

Birth defects are COMMON, COSTLY and CRITICAL.

- **1 in every 33** babies are born with a birth defect.
- Birth defects cost more than **\$2.6 billion** in hospital expenses each year.
- **602 Hoosier infants died** before their first birthday in 2017, and birth defects was the second leading cause of infant deaths. More infant mortality data can be found here: <https://secure.in.gov/isdh/26292.htm>

Current Public Health Initiatives Associated with Birth Defects

- ✓ Neonatal abstinence syndrome (NAS) in relation to the opioid epidemic that Indiana is currently facing.
- ✓ Critical congenital heart disease (CCHD) includes 12 heart conditions, seven of which the pulse oximetry screening may be able to detect.

IBDPR Medical Chart Abstraction Process and Contacts

Medical chart abstractions are required for targeted conditions for confirmation and accurate annual reporting. Medical chart abstracting is the process of collecting and reviewing medical records for information to confirm diagnosis. Medical chart abstractions take place either at the hospital or remotely. A list of charts requiring abstraction can be provided to the hospital prior to review in order to prepare charts onsite or remotely in a queue for medical chart abstractors' review. With advancements in technology, many hospitals and health networks have allowed the abstractors remote access to their EMR. Remote access reduces the burden on both the hospital staff and ISDH. Hospital staff do not have to schedule time or provide space to the



abstractors, and abstractors do not have to drive to all the different hospitals in the state. Remote access, therefore, increases productivity and reduces costs for hospitals and ISDH.

IBDPR Stakeholders

Birth defect surveillance is the first step in preventing birth defects by identifying and collecting information. The information collected through birth defects surveillance systems is used to understand the following:

- Prevalence rates
- Causes and risk factors for birth defects
- Education to the community
- Implementation of prevention strategies
- Referrals to service

Birthing facilities are the first contact for a baby born with a birth defect. As of 2019, Indiana has 87 birthing hospitals and many birthing centers and midwiferies throughout the state. Follow-up continues into the pediatrician's offices, health departments, specialty groups and others. It is important to recognize the work and maintenance that goes into ensuring all of Indiana newborns are well taken care of in regards to GNBS. Efforts are spread throughout the community, and each of us has a crucial role to play in the early detection and intervention of birth defects.

Key stakeholders in the birth defect surveillance include the following:

- Birthing facility managers
- Registered nurses
- Unit secretaries
- Laboratory technicians
- Medical records staff
- Audiologists
- Nurse practitioners
- Hospital physicians
- Pediatricians
- Medical assistants
- Specialists
- Indiana Hospital Association
- AAP, IN Chapter
- IPQIC PGG Advisory Board
- ISDH community partners
- ISDH staff

For more information on the IBDPR program, medical chart reviews or remote access to your facility's EMR, please contact the IBDPR program manager at IBDPR@isdh.IN.gov or visit online at www.BirthDefects.IN.gov.



Community Birth Partners

The GNBS program collaborates with community partners to reach our program mission of ensuring all babies are offered genetic services based off of these results. These community-based healthcare providers are midwives, doulas and other birth attendants providing support before, during and after delivery outside of the hospital setting.

“Birthing center” means any nonhospital facility in which live births routinely take place.

“Health care provider” means the medical professional providing care after birth.

“Midwife” means an individual licensed under IC 25-23-1-13.1.

“Physician” means an individual licensed under IC 25-22.5-5.

“Parent” means a natural (birth parent, step-parent, adoptive parent) legal guardian or other legal custodian of an individual.

“The responsible physician, midwife, birthing center, or hospital shall collect.”

IAC (16-41-17)

Under Indiana Article 3-3-1, when a live birth occurs in a facility other than a licensed hospital or birthing center, it shall be the responsibility of the physician or midwife in attendance at the birth to ensure the family is referred and confirm arrangements for newborn screening.

Newborn Screening with a Midwife or Doula

In the absence of an attending physician or a -midwife, the registrar of births shall refer the newborn or infant immediately to the parent's physician or to the local health department for NBS.

When a live birth occurs in a facility other than a licensed hospital or birthing center, it shall be the responsibility of the physician or midwife in attendance at the birth to ensure the newborn or infant is referred to an appropriate facility, such as a physician office, hospital, birthing center, or local health department, and to make the arrangements to complete and submit all three parts of NBS.

Required Reporting

Hospitals, birthing centers, midwives, and physicians providing home birth services shall provide monthly reports to the department indicating the total number of live births and the number of newborns or infants for whom newborn screenings were submitted.

Visit our webpage link [here](#) for everything healthcare providers are required to report monthly for all live births to INSTEP and diagnosed birth defects to the IBDPR.

For more information on required reporting, see the INSTEP MSR and IBDPR sections of this toolkit or contact us.



ADMINISTRATIVE CODE (410 IAC 3-3-6 Sec. 6)

The Three Screens

http://iac.iga.in.gov/iac/iac_title?iact=410

The Indiana Administrative Code (IAC), or administrative law, is the body of law that governs the activities of administrative agencies of government. In other words, the 410 IAC 3-3-6 Section 6 reflects the NBS Law (IC 16-41-17-2) and gives detail on how the state and the program stakeholders are to perform and adhere to those laws stated. In this section those codes are reviewed and discussed as they relate to achieving program objectives and improving program outcomes.

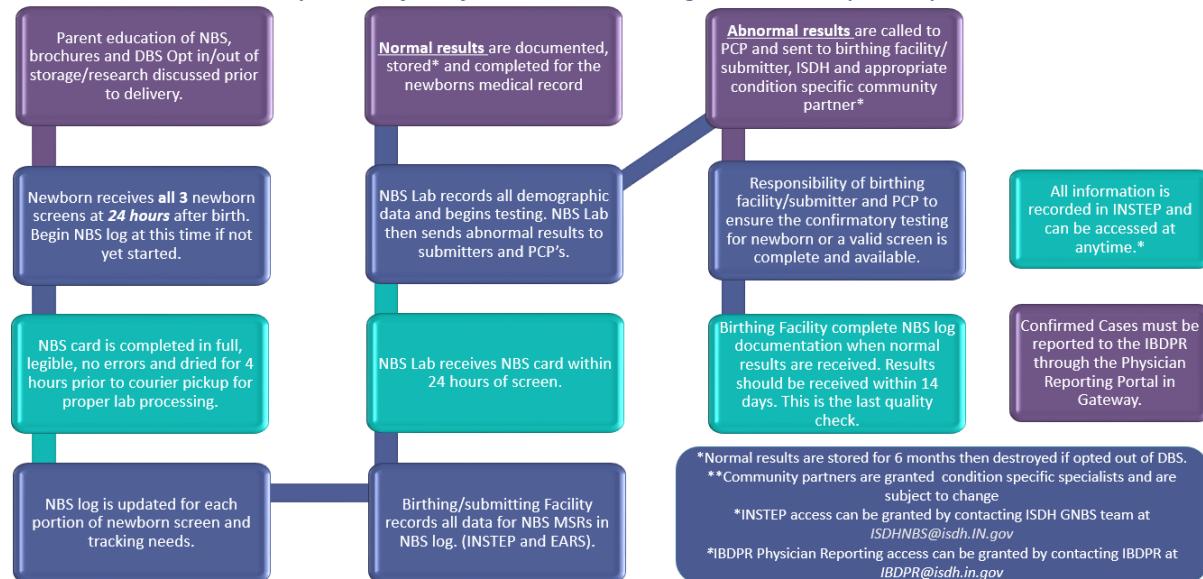
Additional guidance is provided by the GNBS program, which assigns a GNBS Education Specialist to perform annual site visits to all Indiana birthing facilities and is improving outreach to the community birth settings.

For information on scheduling your facility's annual meeting, please email ISDHNBS@isdh.IN.gov.



Genomics and Newborn Screening Process Map

This map is a recommendation for all birth facilities and community birth partners to follow for best outcomes and aligns with NBS Law (16-41-17)



Hearing Screening

A maximum of two inpatient hearing screens can be performed as early as six hours after birth and preferably before discharge. The Early Hearing Detection and Intervention (EHDI) program aims to screen by one month, have a confirmatory evaluation by three months to identify hearing loss, and act on early intervention by six months.

When the hearing screen is not done prior to the DBS being sent to the lab, the pink carbon copy titled “Hearing Screen” should be submitted to the lab with the next set of NBS cards to be picked up by the courier.

*More information on the hearing screen can be found on p. 20 of this toolkit.
For more information and best practices, please refer to the EHDI Policy Manual
by visiting www.Hearing.IN.gov or calling the EHDI program at 317-232-0972.*

Pulse Oximetry Screening

The pulse oximetry test needs to be complete within the first 24 and 48 hours of life to avoid false positives and take advantage of early interventions prior to the ductus arteriosus closing. Pre-term newborns or infants shall be given a pulse oximetry screening, including repeat screenings, at or near the time the heel stick is administered as provided for in section 3(f) of this rule and in conformance with 3-3-3(f), which requires repeat heel stick screenings to be taken 14 days and 30 days after birth or the day of discharge, whichever comes first.

During the test, the baby should be awake, calm, and warm with one probe placed on the baby’s right hand and a second probe placed on one of the baby’s feet. The results along with the date and time are to be documented on the NBS card and your facility’s NBS log. In the case of a religious exemption for the heel stick, the pulse oximetry results’ date and time need to be documented on the religious waiver for ISDH to know a valid test has been administered and documented.

When the pulse oximetry is not completed prior to the DBS being sent to the lab, the pink carbon copy titled “Hearing Screen” should be submitted to the lab with the next set of NBS cards to be picked up by the courier with the pulse oximetry results legibly written in. If there was an echocardiogram in place of the pulse oximetry, document the echocardiogram results in your facility’s NBS log for follow-up purposes as well as on the Monthly Summary Report (MSR) as an exception.

More information on the pulse oximetry screening can be found on p. 21 of this toolkit.



Heel Stick Screening

The heel stick screening is state mandated to take place 24 hours after birth and not later than 48 hours after birth. In the event of discharge or a transfer before 24 hours, and or a transfusion before 24 hours, it is required that a heel stick occur prior to those events if it will not intervene with life-saving measures. This protocol is also relevant for NICU and other special scenarios.

Additional screening can occur for those in the NICU and other special scenarios. The current rule recommends a 14- and 30-day collection timeline and monthly thereafter until discharge or three months of age, whichever comes first.

For NICU and other special cases, it is best practice to still achieve the heel stick by 24 hours old rather than delay detection, intervention, and follow-up care.

An appropriate heel stick procedure should start by warming the proper site with a soft moistened cloth and cleansing with an alcohol towelette. A valid specimen requires all circles to be filled with blood soaked through to the other side of the filter paper while avoiding excessive layering of the blood. Take caution not to touch or smear the specimen, and allow it to dry for four hours prior to submission to the courier service.

Once the DBS is processed at the NBS lab, all GNBS results will then be faxed and or mailed from the NBS lab to the submitting facility to ensure that NBS logs can be updated and resolved, rescreens can be scheduled per the birthing facility, and follow-up with the listed PCP can be initiated.

All information is to be completed legibly on the NBS card then mailed to the NBS lab and documented in your facility's NBS log according to Indiana Administrative Code.

More information on the heel stick screening can be found on page 23 of this toolkit.



NBS Log

Each hospital or birthing center and midwife or physician are to maintain an NBS log documenting the following information for all infants born, transferred in, or screened at the facility:

- Name of newborn or infant
- Attending physician or midwife (*follow-up PCP is recommended*)
- Medical record number
- Form number of sample sent
- Date sample collected
- Date sample sent
- Date results received
- What the results were (hearing screen, pulse oximetry, heel stick)
- Name of person notified of positive results and date and time of notification

The NBS log should be reviewed daily to ensure that they are in compliance and that results are recorded within 14 days – if results are not received within 14 days, the laboratory must be contacted by telephone. If a baby has been discharged prior to receiving mandated tests, the responsible health care provider and the NBS follow-up care coordinator must be contacted immediately by telephone and written notification to inform them that a specimen must be taken within three days. The GNBS follow-up care coordinator must be contacted within three days if the responsible healthcare provider cannot be contacted. The follow-up care coordinator will then contact the local health officer who will ensure that the specimen is taken. If the healthcare provider is notified by the laboratory that a specimen is inadequate, the provider must obtain a repeat specimen within 48 hours.

If unable to repeat the specimen within 48 hours, the GNBS team must be contacted **immediately by telephone at 888-815-0006** to assist in follow-up on all babies who have been reported as not having received a complete and valid screening. This includes those discharged before being screened or *discharged without NBS*.

Reporting these cases by phone does not take the place of reporting within the MSRs.

Newborn Screening Lab

The GNBS Program contracts Indiana University Newborn Screening Laboratory as a vendor of ISDH. Beginning in 1991, this NBS lab was selected as the centralized location for needs regarding the distribution, collection, and processing of the NBS cards for the entire state. The NBS lab serves all Indiana birthing facilities, midwiferies, and other NBS card submitters across the state.

The following is a quick reference to the lab's process once receiving an NBS card:

NOTE: Submitters of an NBS card with the heel stick portion completed will get a faxed or mailed report from the NBS lab once the screening process has been completed for normal results. If abnormal results or issues with the specimen arise, then the NBS lab will be in contact with the submitting facility immediately.

If you experience any issues in obtaining results from the NBS lab, please contact the lab immediately to avoid lag in any follow-up or documentation in your facility's GNBS log.

If your birth facility (hospital, birthing center, midwife/midwifery) needs more brochures or NBS cards, please call the lab and request these materials. New GNBS card submitters will need to set up an account with the lab and ISDH GNBS for submitting and reporting.

ALSO
KNOWN
AS

- Heel stick card
- Guthrie card
- Blood spot collection
- Heel prick card
- DBS card
- PKU test/card

NBS Lab
Phone: 800-245-9137
Phone: 317-278-3245
Fax: 317-321-2495

NOW Courier Information

NOW Courier is the statewide courier service used for all newborn screening specimens, which ensures timely delivery to the NBS lab, located in Indianapolis, from all Indiana birthing facilities. NOW's mission is to provide the most efficient, cost-effective, and value-added courier services through quality service, people, and proven technology while developing long-term partnerships with customers. NOW serves all Indiana birthing facilities six days a week to ensure all DBS cards are delivered in a timely manner to the NBS lab. NOW offers a tracking system to track every DBS card that leaves a birthing facility and can be found at any point of the route. This ensures no cards are lost. It is best to become aware of your birthing facility's NOW courier service route and times to ensure your daily pick-up is made with no delay.

You can view more information about NOW Courier service at <http://nowcourier.com>.

If your facility does not have high birth volumes that require a set schedule for courier services, you can indicate the need for pick-up by emailing NewbornScreening@NowCourier.com. This service for low-volume facilities is only available Monday-Friday.

The following lists the regional numbers to call in case your facility needs information about a scheduled route or if an error has occurred.

NOW Courier Contacts	
Email: NewbornScreening@NowCourier.com	
Web: http://nowcourier.com	
Regional Contacts	
Indianapolis Phone: 317.638.7071	Evansville Phone: 812.474.0400
Louisville Phone: 502.895.4777	Cincinnati Phone: 513.563.6066
Fort Wayne Phone: 260.747.2393	South Bend Phone: 574.233.1838
Merrillville Phone: 219.736.6066	

The designated Newborn Screening laboratory is subject to change at the end of every odd fiscal year as this is when the contract is up for renewal.

NBS CARD 101

Utilizing language that is well known across all stakeholders is critical in maintaining a centralized program for all involved. With NBS beginning over five decades ago, many terms have been created to refer to NBS. Please see the sidebar for additional names for the *newborn screening card*.

For instance, referring to the heel stick screen results as “an abnormal PKU result” can derail efforts by families and physicians alike when an abnormal result occurs. The result could be an abnormal metabolic, but it also could be an abnormal endocrine, cystic fibrosis, spinal muscular atrophy (SMA), hemoglobinopathy, or even a severe combined immunodeficiency result (SCID). These conditions vary in follow-up and confirmatory testing.

This communication is also unique and important for those involved with the hearing screen alone. The NBS card is also known as “the heel stick card,” and some stakeholders are not aware of other term, which can lead to miscommunication when it is referred to as “the PKU test.”

The most current and approved version that is to be utilized does still contain the **protein feed time**.

Yellow = Hospital Copy

Pink = Hearing Screen Copy

Follow-up PCP is the identified provider who will be seeing the baby for well checks in the coming days to weeks.

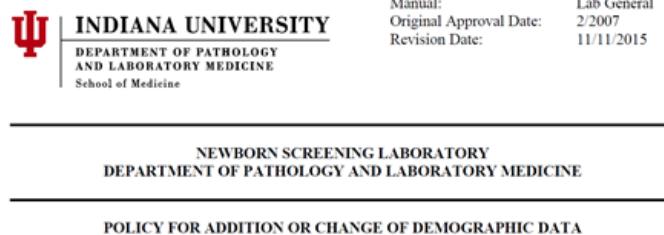
Mother of baby is the natural mother, biological parent, adoptive parent or guardian, or agency involved with the newborn.

NBS CARD: Policy for Addition or Change of Demographic Data

If you discover there is an error in the required patient information listed on your NBS log or results received from the NBS lab, you must notify the NBS lab as soon as possible by submitting the Change of Information worksheet to the NBS lab. In many cases, the NBS lab will contact you by phone if they presume an error has been made on the NBS card.

Required information includes:

- Patient name
- Medical record number
- Birth weight
- Birth date and time
- First feed date and time
- Screen collection date and time
- Patient's follow-up physician



If any of this information is missing, a phone call is made to the submitter to retrieve this data.

A screen cannot be properly evaluated and results printed without these critical pieces of information being available and accurate.

Examples of addition or change of demographic information:

- Name change
- Incorrect date of birth
- Incorrect birth weight
- Incorrect time of birth
- Type of feeding
- Transfusion status
- Correlation between transferring facilities
- Physician change
- Missing information when submitted

It is advisable that you make necessary notes of this error and make the updates within any other areas of documentation such as the NBS log, MSR, or the facility's electronic medical records system.

Policy for Addition or Change of Demographic Data can be found [here](#).

RESULTS AND FOLLOW-UP

INSTEP

<https://gateway.isdh.in.gov/Gateway/SignIn.aspx>

Primary care providers have online access to newborn screening results, available through the Indiana Newborn Screening Tracking and Education Program (INSTEP). The ISDH GNBS program highly recommends that a minimum of two people per office group have access to INSTEP to obtain GNBS results.

MSRs are submitted by hospital staff within INSTEP. ISDH strongly encourages assigning a minimum of two people to each mandatory reporting task. A two person minimum per each screening report task increases support with the additional work load, reduces delay when there is turnover or time off, and encourages collaboration resulting in fewer reporting errors.

To obtain INSTEP registration instructions, please send an email to the GNBS team at their shared email (ISDHGNBS@isdh.IN.gov) and include the following information:

- 1) Your full name as it appears on any licenses
- 2) The name of the physician's office, hospital, or other medical facility with which you are affiliated
- 3) The primary care physicians within the medical facility with which you are working
- 4) The main telephone number of that medical facility
- 5) Your role at that facility (i.e., physician, nurse, medical assistant, audiologist, etc.)
- 6) The reason you are requesting to have access to INSTEP

Parents or healthcare professionals can also contact the ISDH Genomics and Newborn Screening Program by calling 888-815-0006.



Requesting Results

If you do not have INSTEP access and you would like [to request NBS results](#), please see the previous instructions on how to obtain INSTEP access to search for the NBS results online.

Facilities can also fax the lab with your office's cover page to the Genomics and Newborn Screening Program at ISDH at 317-234-2995 and include the following child information:

- ✓ Name
- ✓ Date of birth
- ✓ Mother's name
- ✓ Birth facility

NOTE: If it is known that the baby has had an abnormal screen, please fax your request to the Newborn Screening Laboratory at 317-321-2495 or call them at 317-278-3245.

Submitters of a NBS card with the heel stick portion completed will get a faxed or mailed report from the NBS lab once the screening process has been completed for normal results. If abnormal results or issues with the specimen arise, then the NBS lab will be in contact with the submitting facility immediately.

If you experience any issues in obtaining results from the lab, please contact the lab immediately to avoid lag in any follow-up or documentation in your facility's GNBS log.

Please email questions about GNBS results to the GNBS program team at ISDHNBSP@isdh.IN.gov.



Presumptive Positive or Abnormal Heel Stick Results

<https://www.in.gov/isdh/27967.htm>

Presumptive positive or abnormal results will also be documented in your GNBS log, and this follow-up process includes phone notification from the NBS lab and contacting the PCP on file. Please refer to ISDH NBS lab's recommended guidelines.

At the time of a presumed positive heel stick condition, the NBS lab notifies both the birth and submitting facility, in addition to a condition-specific specialist or community partners, to provide and assist the primary care provider with disease intervention and in follow-up services.

At this time, the birth and/or submitting facility may be asked to obtain additional heel stick screens, assist in follow-up, or collaborate with community partners for next steps.

While false positives can occur on either an initial or a rescreen, it is imperative to take immediate action when heel stick results are presumptive positive/abnormal to ensure best outcomes for the infant.

The next page of this toolkit explain **who** the GNBS program's community partners are and **what** the contracted expectations are for NBS and our families.

For the most recent list of the Indiana Newborn Screening panel, please visit www.NBS.IN.gov.



GNBS Community Partners

The GNBS program collaborates with community partners to reach our program mission of ensuring all babies are offered genetic services based on these results. These community-based healthcare providers are condition-specific specialists that are contracted through a competitive grants process occurring biennially.

Community partners are a selected group of specialists who provide follow-up for abnormal and presumptive positive newborn screening heel stick conditions. This follow-up includes, but is not limited to, confirmatory testing, genetic counseling, social services, nutrition services, and general public and academic education.

INBORN ERRORS OF METABOLISM*	<ul style="list-style-type: none">Community Health Clinic (CHC)Indiana University (IU)
ENDOCRINE DISORDERS	<ul style="list-style-type: none">Indiana University (IU)
HEMOGLOBINOPATHIES (HGB)	<ul style="list-style-type: none">Indiana Hemophilia and Thrombosis Center (IHTC)
SEVERE COMBINED IMMUNODEFICIENCY(SCID) AND SPINAL MUSCULAR ATROPHY (SMA)*	<ul style="list-style-type: none">Community Health Clinic (CHC)Indiana University (IU)
CYSTIC FIBROSIS (CF)	<ul style="list-style-type: none">Indiana University (IU)
LYSOSOMAL STORAGE DISORDERS (LSD)	<ul style="list-style-type: none">PENDING

**These follow-up services are served by two vendors (community partners) based on geographic location. CHC is focusing on the following 18 Indiana counties: Adams, Allen, Daviess, DeKalb, Dubois, Elkhart, Fulton, Jay, Kosciusko, LaGrange, Marshall, Noble, Orange, Parke, Steuben, Washington, Wayne, and Whitley, to serve the Plain population and others. The second, IU, will focus on statewide coverage and is located centrally in Marion County.*

For further questions regarding GNBS results and the community partners, please email the GNBS Program team at ISDHNBS@isdh.IN.gov.

For the most recent list of the Indiana Newborn Screening panel, please visit www.NBS.IN.gov.



Newborn Screening Best Practices

Hearing Screen

EHDI Manual found at www.Hearing.IN.gov

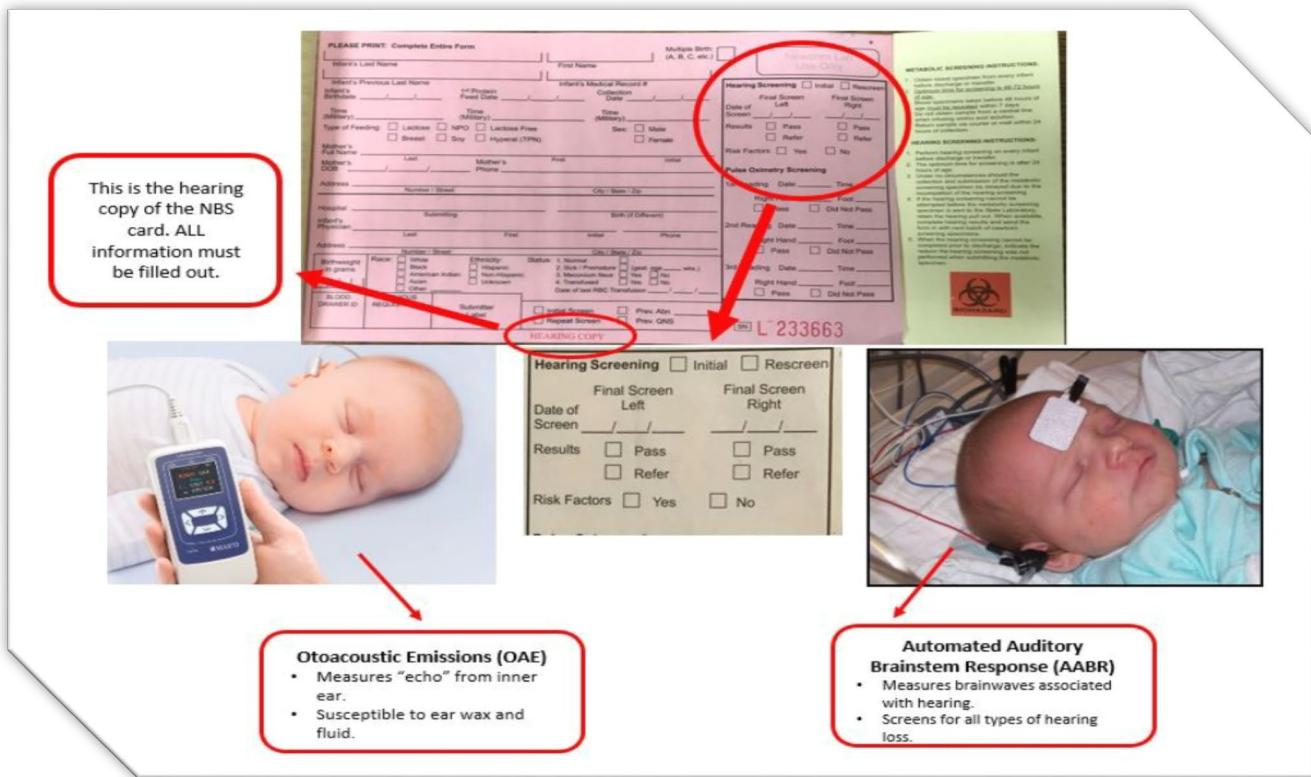
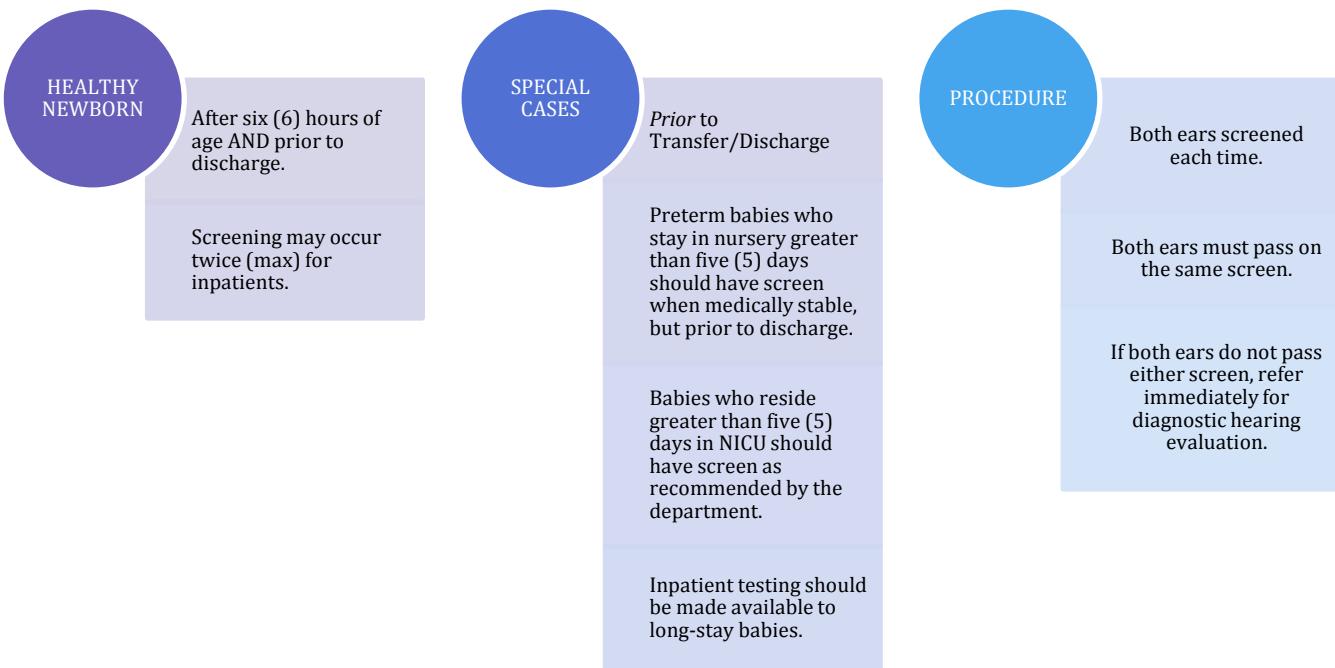
Each year in the United States, approximately three of every 1,000 babies are born with permanent hearing loss; in Indiana, this accounts for 250 babies annually. Additionally, Indiana has a unilateral (one ear) hearing loss rate of nearly 40 percent. While not all hearing loss can be reversed or prevented, newborn screening provides the opportunity for early detection and intervention for improved growth, development, and social-emotional health. By including hearing screening in Indiana's newborn screen, most of these children can be identified before three months of age and enrolled in appropriate intervention services for them to achieve the best outcomes. It is important to find hearing loss early so that early intervention services and language development can begin as soon as possible.

Mandated by Indiana Statute IC 16-41-17-2, the Universal Newborn Hearing Screening (UNHS) can be performed as early as six hours after birth, and it is best practice that not more than two inpatient hearing screens are done. Similar to the heel stick and pulse oximetry screens, the UNHS needs to be completed prior to discharge.

The Early Hearing Detection and Intervention (EHDI) program aims to screen by one month, have confirmatory evaluation by three months to identify hearing loss, and act on early intervention by six months. All babies can and should have their hearing screened before they leave the birthing facility. When an infant has not passed two newborn hearing screens prior to discharge, hospitals and birthing facilities are responsible for providing a referral to a diagnostic audiologist and/or to the primary care physician. It is not recommended to rescreen the infant who did not pass the initial two screenings. The birthing facility is also required to report the hearing screening results to Indiana's EHDI program.

Following a failed newborn hearing screen, the diagnostic evaluation should be completed prior to three months of age to identify a hearing loss within the recommended time frame to reduce parental anxiety, decrease the need for sedation, reduce the lost-to-follow-up rate, and ensure appropriate early intervention services in a timely manner.

A hearing screen technique reference is provided on the following page.



Submit results to ISDH GNBS via pink slip (hearing copy) when not completed with the heel stick, or use a religious refusal form when heel stick refused or when pink sleep is unavailable.

Pulse Oximetry Screen

<https://www.in.gov/isdh/27967.htm>

Beginning Jan. 1, 2012, Indiana became the second state to have pulse oximetry screening added to the state newborn screen. Since then, every baby born in Indiana has been required to have a valid pulse oximetry screening, unless the baby's parents object to newborn screening based on their religious beliefs. *See page 5 of this toolkit for more information on the religious refusal.*

The pulse oximetry is used as part of newborn screening to determine how healthy a baby's cardiovascular and pulmonary functions are by detecting oxygen levels in the blood. Babies who have low oxygen levels may have CCHD. "Pulse oximetry screening in newborns is critical since CCHDs are not always detected in utero; this screening can alert providers of possible CCHDs before major signs and symptoms occur after birth."

WELL-BABY PULSE OXIMETRY: Screen all newborns between 24 and 48 hours after birth and prior to discharge.

PASS: Greater than or equal to 95% in either the right hand AND foot with no variance greater than 3%.

RESCREEN: Less than or equal to 94% in either hand or foot and/or a variance greater than 3%; repeat the screen in one hour up to three times.

If failed rescreens, immediate clinical assessment and referral to cardiology.

IMMEDIATE REFERRAL: Less than or equal to 90% in either right hand or foot.

Pulse oximetry screening must take place between the first 24 to 48 hours after birth. This test can occur a maximum of three times, with a minimum of one hour between screens, before further assessment is needed. There are scenarios when this screen is bypassed and an echocardiogram is done in place.

In these scenarios, please document both pulse oximetry and echocardiogram results when one occurs following a valid failed pulse oximetry screening and always indicate if and when the echocardiogram occurred and a brief description in the notes of the exception to support follow-up needs. Documentation is done on the NBS card, within INSTEP on their CHIP in the general notes section, religious waiver, and/or on your MSR submission.



To pass the pulse oximetry screen, a baby must have results of 95% or higher in the right hand or a foot and no difference equal to or greater than 3% between the right hand and foot.

A failed pulse oximetry screen consists of results in either the hand or a foot lower than 95% and/or a difference greater than 3% between the right hand and foot.

SPECIAL CASES PULSE OXIMETRY: Screen at the time the heel stick is administered for early detection and intervention between 24 and 48 hours of life.

PASS: Greater than or equal to 95% in the right hand and foot with no variance greater than 3%, and continue to screen on subsequent heel sticks if preterm and or low birth weight.*

RESCREEN: Less than or equal to 94% in either hand or foot and/or a variance greater than 3%; repeat the screen in one hour up to three times.

If failed rescreens, immediate clinical assessment and referral to cardiology.

IMMEDIATE REFERRAL: Less than or equal to 90% in either right hand or foot.

IMPORTANT: If newborn is on supplemental oxygen, screen after weaned to room air for 24 hours and prior to discharge.

If not being weaned, an echocardiogram (ECHO) should be completed.

A prenatal diagnosis (no need to screen if already diagnosed) and a post-natal diagnosis are to be documented in the pulse oximetry MSR for the newborn with supporting documentation such as interpretation of ECHO and date of diagnosis for immediate follow-up purposes and medical abstraction review for the IBDPR.**

*See reference 3(f) in 410 IAC 3-3-3.5 Pulse oximetry measurement for critical congenital heart disease.

**Report confirmed diagnosis to the Indiana Birth Defects and Problems Registry (IBDPR) within 30 days.

It is important for parents to know that pulse oximetry cannot identify every child with CCHD and that most babies who pass the pulse oximetry screen will not have CCHD. Therefore, it is important for parents to know the signs of CCHD (including cyanosis of the skin/fingernails/lips, fast breathing and poor feeding or poor weight gain). Encourage a family to contact their baby's doctor if they notice any of these signs.

A pulse oximetry screen technique reference is provided on the following page.

A protocol flow chart is available for pulse oximetry screens as reference to best practices.



PULSE OXIMETRY PROCEDURE

Pulse oximeter is placed on baby's right hand and one of baby's feet.

Baby should be awake, calm, and warm.

PLEASE PRINT: Complete Entire Form

Infant's Last Name _____ First Name _____ Multiple Birth: (A, B, C, etc.)

Infant's Previous Last Name _____ Infant's Medical Record # _____

Infant's Birthdate _____ Feed Date _____ Collection Date _____

Time (Military) _____ Time (Military) _____ Time (Military) _____

Type of Feeding: Lactose NPO Lactose Free
 Breast Soy Hyperal (TPN)

Sex: Male Female

Mother's Full Name _____ Last _____ Mother's First _____ Initial _____

Mother's DOB _____ Phone _____

Address _____ Number / Street _____ City / State / Zip _____

Hospital _____ Submitter _____ Birth of (different) _____

Infant's Physician _____ Last _____ First _____ Initial _____ Phone _____

Address _____ Number / Street _____ City / State / Zip _____

Birthweight _____ in grams _____ Race: White Black Hispanic American Indian Non-Hispanic Unknown Ethnicity: Hispanic Non-Hispanic Unknown Status: Normal Sick / Precursors Gest. Age _____ wks.
 Sick Non-Hispanic Unknown Transfused Yes No
 Non-Hispanic Unknown Transfused Yes No
 Unknown Non-Hispanic Unknown Transfused Yes No

Date of test NBS Transfusion _____

BLOOD DRAWER ID _____ PREVIOUS REGISTRATION # _____ Submitter _____ Labeled _____

Initial Screen: Previous Abn:
Repeat Screen: Previous QM:

Newborn Lab Use Only

Hearing Screening: Initial Rescreen

Date of Screen: _____

Final Screen Left _____ Right _____

Results: Pass Fail Refer

Both Sides: Yes No

Pulse Oximetry Screening

1st Reading Date _____ Time _____

Right Hand _____ Foot _____

Pass Did Not Pass

2nd Reading Date _____ Time _____

Right Hand _____ Foot _____

Pass Did Not Pass

3rd Reading Date _____ Time _____

Right Hand _____ Foot _____

Pass Did Not Pass

NBS COPY L233663

METABOLIC SCREENING INSTRUCTIONS:

- Observe infant spontaneously breast feeding within 24 hours of birth.
- Infant must be accessible in 48-72 hours of birth.
- Normal conditions before earliest 48 hours of birth.
- Normal blood glucose levels for a newborn are 70-100 mg/dL. If blood glucose levels are below 70 mg/dL, infant should be fed a breast milk meal containing protein and calcium.
- Infant should be fed a breast milk meal or most within 24 hours of collection.

HEARING SCREENING INSTRUCTIONS:

- Perform hearing screening on newly born infants discharged or transferred. The earliest time for screening is after 24 hours of age.
- Under no circumstances, administer hearing screening equipment too delayed due to the following:
 - Failure to respond to the hearing screening equipment before the testable screening age.
 - Failure to respond to the hearing screening equipment after the testable screening age.
 - Failure to respond to the hearing screening equipment in the presence of infection.
 - Failure to respond to the hearing screening equipment in the presence of a medical condition.
 - Failure to respond to the hearing screening equipment in the presence of a metabolic condition.
- Infant should be fed a breast milk meal containing protein and calcium prior to discharge. Administer the hearing screening equipment after the breast milk meal.





Pulse Oximetry Screening

1st Reading Date _____ Time _____

Right Hand _____ Foot _____

Pass Did Not Pass

2nd Reading Date _____ Time _____

Right Hand _____ Foot _____

Pass Did Not Pass

3rd Reading Date _____ Time _____

Right Hand _____ Foot _____

Pass Did Not Pass



Submit results to ISDH GNBS via pink slip (hearing copy) when not completed with the heel stick, or use Religious Refusal form when heel stick is religiously refused.

Heel Stick Screen

<https://www.in.gov/isdh/27967.htm>

New Conditions

It is the mission of the ISDH GNBS program to use the RUSP as guidelines for adopting conditions to the Indiana GNBS panel. As the SACHDNC recommends new conditions to be added for all NBS programs, it is the responsibility of states to perform cost benefit analyses to identify the local community's ability to support and sustain screening and follow-up services. As of 2019, the Indiana GNBS panel detects **47* different conditions with the heel stick.**

*Starting July 1, 2020, there will be an additional three conditions screened for through the heel stick: Krabbe disease, Pompe disease, and mucopolysaccharidosis type 1 (MPS-1 or Hurler Syndrome).

Without this heel stick screen, these conditions would remain undetected until symptom onset, at which point severe developmental delays or death will have already occurred. If a child has one or more conditions, it can be confirmed up to as early as one week of life, allowing for quick detection, early intervention, and life-saving treatment.

Once a condition has been detected, early intervention and life-saving treatment need to take place. The birthing/submitting facility should receive the heel stick results via fax from the GNBS lab within seven days of the birth, prompting the receiver to complete the "Results Received" column of the NBS log. The receiver must review the results and follow the recommendations provided by the NBS lab immediately.

Heel Stick Results Protocol

Normal valid results should be received by the birthing/submitting facility within seven days of birth. These results received should then be documented in your NBS log to consider the screen resolved. No further action needs to be taken when a heel stick result is normal and valid.

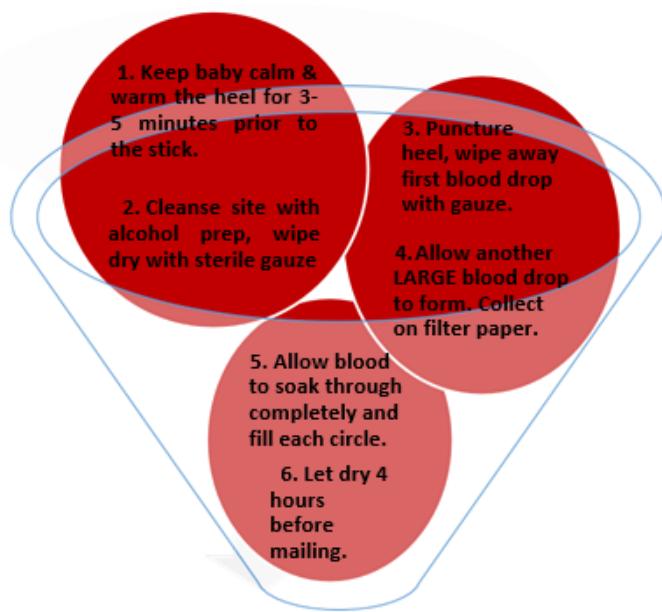
Presumptive positive or abnormal results will also be documented in your GNBS log, and this follow-up process includes phone notification from the NBS lab and contacting the PCP on file. Please refer to ISDH NBS lab's recommended guidelines.

Invalid results require a rescreen or repeat specimen within five business days of the initial screen. Please review the table below listing quality indicators for invalid results.



For NICU and other special cases such as preterm delivery, please refer to the ISDH GNBS recommended guidelines. This protocol aids in follow-up and can identify abnormalities even when the heel stick is not considered a valid test.

A heel stick screen technique reference is provided on the following page. A *protocol flow chart is available for heel stick screens as reference to best practices*.



Quality Heelstick Procedure

Healthy Newborn: 24 hours after birth

- Prior to discharge/transfer
- Special Cases: Transfusion and NICU
 - Pre-transfusion:
 - Collect before transfusion
 - Post-transfusion:
 - Collect prior to 6 days after birth
 - Collect final specimen 2-4 months post last transfusion
 - **<2,000 grams**
 - **24 hours after birth**
 - **6 days***
 - **14 days**
 - **30 days**

Monthly thereafter until discharge or 3 months of age, whichever occurs first

*The Indiana State Department of Health Genomics and Newborn Screening program recommends a screening on day 6 to increase rapid detection and intervention. Follow



NBS Card Quality Assurance

Continuous quality assurance needs to occur with all birth partners. Proper newborn screening practices start with the receipt of the NBS cards--how they are stored and then provided to those involved with filling in accurate information and reviewing the NBS card prior to sending to the NBS lab or completing the last quality check.

TIPS:

- Store the card upright and vertical, but let the blood spots dry flat, horizontally.
- Use black pen, not pencil, and write hard to reach the last carbon copy.
- If expired, do not use.
- Do not touch the filter paper.
- If card is separated, apply a patient sticker to each copy.

The highlighted areas on this image of the most up-to-date NBS card are required for analyzing the DBS. Missing information will delay early detection and intervention.

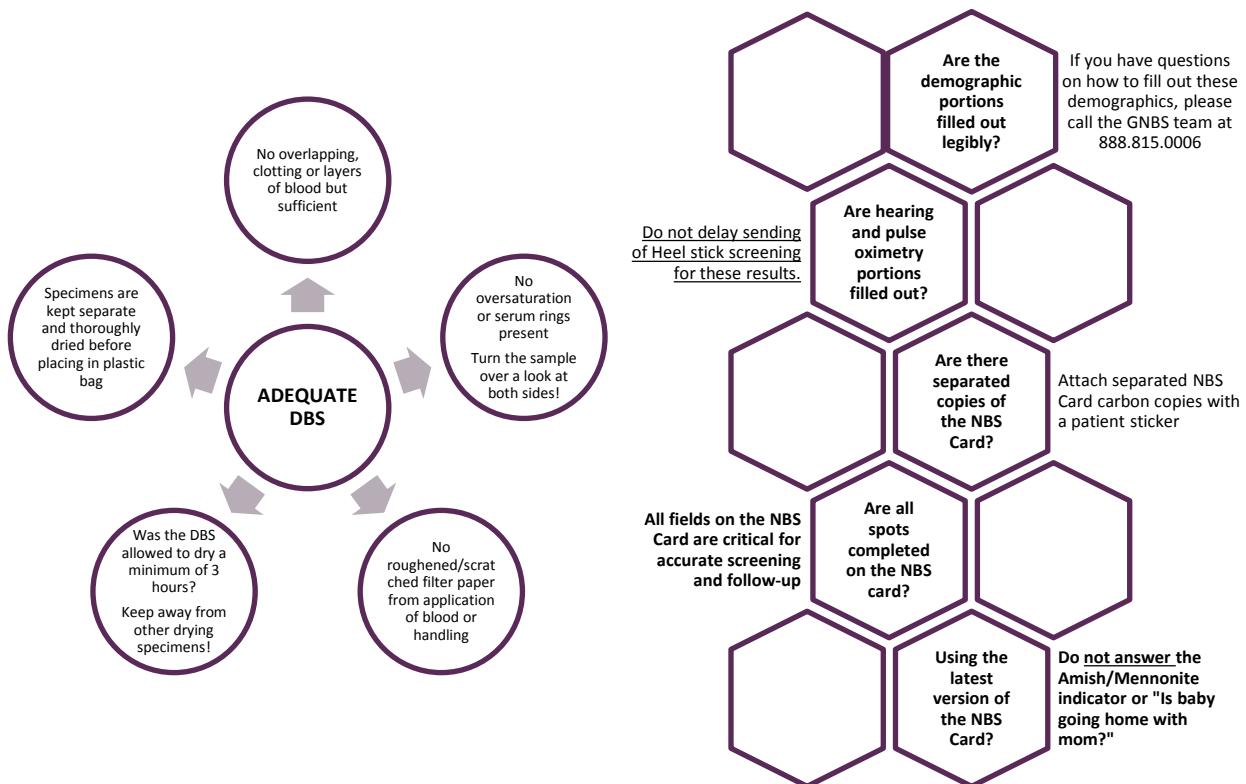
Last Quality Check

WHEN? The last quality check should occur before the baby is discharged and before the NBS card is sent to the NBS lab.

WHO? Best practice is for whomever places the card in the envelope to send to the NBS lab. However, this can vary on who collects the sample and where it is picked up.

WHERE? The GNBS program suggests that a quality check be documented in the NBS log.

The following are questions to be asked to demonstrate a quality check:



DBS sent by courier within 24 hours of collection?

Call or email NOW Courier as needed to ensure a timely pick up

Has the NBS Log been completed?

Update NBS Log fields if missing information such as pulse oximetry and/or hearing screen results or follow-up PCP, Etc.

Dried Blood Spot (DBS)

<https://www.in.gov/isdh/27967.htm>

The heel stick screens for these conditions by testing the blood that is collected onto the DBS card. At times, there may be leftover dried blood on the DBS card. According to Indiana NBS Law, the remaining DBSs can be made available for epidemiological research to identify new conditions, treatments, and preventative measures. **All research will be conducted on de-identified specimen, per Indiana Code 16-41-17-10.**

ISDH requests written consent from parents or guardians of newborns if they would like to make their child's DBS available for research purposes.

If consent is granted, the DBS of the child will be stored and made available for epidemiological research for a period of three years and then destroyed.

If parents do not consent to making their child's DBS available for research, their child's DBS is kept for six months to ensure all GNBS testing has been completed and then is destroyed.

Within the six months, parents or legal guardians can request that their child's DBS be stored for research purposes by completing and sending in a form to the GNBS Program. This form can be found at <https://www.in.gov/isdh/27967.htm>.

Within the three years, parents or legal guardians may request that their baby's DBS be destroyed by completing and sending a form to the GNBS Program. This form can be found online at <https://www.in.gov/isdh/27967.htm>.

Cards for any child born before June 1, 2013, have not been made available for research and have been destroyed in a secure manner. Other cards will be destroyed on a schedule in accordance with the three-year retention policy.

Dialogue to Families

*This section is intended to be suggested dialogue pieces to communicate with the family **PRIOR** to GNBS. This will explain the process and procedures involved with GNBS, dried blood spot collection and storage, epidemiological research options, and aftercare.*

Families should be informed of the DBS consent (opt in/out) portion! Storage beyond the required six months can assist with further testing without collecting more blood from the baby in addition to multiple other benefits to the family for the next three years after birth.

Additional DBS information for medical providers can be found on page 30 of this toolkit.

Newborn screening is required by the state of Indiana and screens for **49 different health conditions** that your baby may have. The state screens for these different health conditions through three newborn screens:

- The heel stick, which is a blood test that detects 47 different genetic (heritable) diseases
- The pulse oximetry, which measures oxygen levels and heart rate to evaluate heart and lung function
- The hearing screen to detect hearing loss

Newborn screening aims to protect the health of your baby and has been proven to be the most effective public health program across the United States. It is important that we know who your baby's doctor will be in the case of an abnormal result.

**Take this time to ensure accurate follow-up PCP is indicated on the NBS card.*

We will need your signature on the back of this card and for you to check whether or not you would like your baby's blood to be stored until his/her third birthday. Storing your child's dried blood spots that are left over after the heel stick screening has occurred has been found to be beneficial for families for paternity, missing child, further screening, and the state program for de-identified medical research efforts. If you encounter any questions regarding the status of a child's DBS, please do not hesitate to contact the Indiana Genomics and Newborn Screening Program.

www.NBS.IN.gov



Suggested Statements from Staff to Family

Newborn screening is required by the state of Indiana, aims to protect the health of your baby, and has been proven to be the most effective public health program across the United States. As of 2019, Indiana screens for 49 different health conditions your baby may have. The state screens for these health conditions through three newborn screens: the heel stick, which is a blood test that detects 47 different genetic (inheritable) diseases; the pulse oximetry, which measures oxygen levels and heart rate to evaluate heart and lung function; and a hearing screen to detect hearing loss.

Newborn screening is important to protect the health of your baby. If these conditions are left untreated, they may result in adverse health outcomes impacting the mental and physical growth and development of your baby. This can reduce your baby's quality of life and lead to lifestyle restrictions. This can also lead to an increase in your baby's use and cost of healthcare and other resources.

Please sign the back of this card and check whether or not you would like your baby's blood to be stored until his/her third birthday. Storing your child's dried blood spots that are left over after the heel stick screening has occurred has been found to be beneficial for both families and de-identified medical research efforts. If you encounter any questions regarding the status of a child's DBS, please do not hesitate to contact the Indiana Genomics and Newborn Screening Program.

The fees collected to conduct newborn screening are used to support families by ensuring that community resources and medical care are available for the children who test positive for these health conditions. These fees might help pay for expenses that result from conditions including: special medical foods, prescription medications, laboratory testing, frequent physician visits, specialized therapies, and surgeries.

If newborn screening conflicts with your family's religious beliefs, you have the right to decline any or all three of the newborn screens. The religious waiver form must be used to indicate your decision to decline any of the newborn screens. The religious waiver is then submitted to the Indiana State Department of Health (ISDH) Genomics and Newborn Screening program.

For more information, please refer to the newborn screening pamphlets provided by your birthing facility or midwife.

More information, including a full list of the conditions tested for, can be found at www.NBS.IN.gov.



Monthly Summary Report (MSR)

Birthing facilities and midwiferies are required to submit MSRs and can do so by submitting the MSR via hardcopy or electronically. INSTEP allows birthing facilities and midwiferies to electronically submit their monthly live birth totals, record of babies exempt from newborn screening for religious beliefs, transferred to another facility and/or other identified scenarios, and the total number of newborn screens completed. *For more information on INSTEP, please refer to page 18 of this toolkit.* When this data is valid, ISDH is able to utilize the data for the following initiatives:

- Ensure Indiana babies are receiving valid screens
- Be alerted when there is missing information, an invalid heel stick, abnormal results, or no record prompting follow-up care
- Reduce lost to follow-up cases
- Complete quality indicator reports for each facility and midwifery
- Ensure that facilities and midwiferies are abiding the state mandated law
- Increase awareness in the community
- Improve the Newborn Screening Program
- Share reliable statistics
- Show the impact our screens have not only on Indiana babies, but also public health initiatives

The overall purpose of adequate MSRs is to ensure that all babies born in Indiana are receiving valid screens and rescreens and getting the follow-up care they need based on those results.

To ensure your facility is submitting accurate and valid data into the MSRs, please encourage collaboration between the hearing, pulse ox, and heel stick reporters and keep your NBS log up-to-date and available to all reporters.

MSRs are due by 5 p.m. EST on the 15th of each month.

If your facility needs more waivers, the hardcopy MSR, or other resources, please refer to our [reporting page](#).

If you have any questions about completing the MSR, please contact the NBS follow-up care coordinator at 317-233-7019.



MSR 101

Prior to submitting your MSR:

Gather information from your facility's Newborn Screening Log.

Compare your numbers with other reporters within your facility. Each facility has a hearing, pulse ox, and heel stick reporter.*

Double-check your information from your facility's NBS Log.!

**Pay close attention to the live birth number as this number should be the same for each reporter monthly. Exceptions and total number of screens can vary for many reasons.*

If you have questionable or conflicting data within your NBS log or Exception Entry page, contact the NBS follow-up care coordinator at 317-233-7019 before submitting the MSR.

Exception Entry

Prior to submitting your exceptions:

Resolve Holdovers

(Promote, Accept/Decline, Edit Transfer Detail)

Avoid Duplicate Exception Entries

Input Accurate Data with Supporting
Documentation

Explanations of Exception Page

The initial exception scenario should be priority. Additional information can always be documented in the notes/comments section to assist with follow-up. When needing to update to another exception, ensure you are not creating additional CHIPs/PIDs.

PROMOTE

ACCEPT

Only accept when you are sure that the baby is in the care of your facility. If the baby is not in the care of your facility, please see the DECLINE portion and notify the GNBS follow-up care coordinator.

DECLINE

When positive that the baby is NOT in the care of your facility, you must decline the transfer. Declining will electronically send it back to the original submitter. Leaving it as is delays follow-up.

SUPPORTIVE DATA

When inputting an exception, information will need to be known in order to not only complete the exception entry, but allow for adequate follow-up based on this coinciding information (time of birth, time of death, receiving facility, date of transfer, follow-up PCP, etc.).

Provide supportive documentation when screens are delayed and/or refused to improve data and follow-up.

EX: If an echo occurs due to signs and symptoms, select 'Echocardiogram' and INPUT the interpretation from diagnostic testing.

- Religious Refusal forms must coincide with a Religious Refusal Exception.
- Medical documentation must coincide with Prenatally/Postnatally Diagnosed with CCHD.

TIMING OF ENTRIES

Historically, MSR submitters were asked to input high-risk (discharged without NBS) into the exceptions page as quickly as possible to notify ISDH GNBS. Reverting back to the *Best Practices*, please notify via telephone immediately.

Notify ISDH GNBS of all high-risk cases by phone call.

Inputting transfers accurately is crucial for the receiving Indiana birthing facility to accept/decline them prior to submitting their MSR. This allows quicker follow-up with the correct facility and avoids unresolved holdovers in the following month.

Live births that become deceased before any initial or valid NBS portion also need to be considered priority to avoid making contact with the deceased's family.

Exception Entry Definitions

- Ensure that you have the supporting documentation for the exception
- you are reporting. Exceptions and their description are listed below:

Transfer Only

- Transferred In- when valid, must accept or decline
- Transferred Out- must provide receiving facility for them to accept transfer in
 - When a baby is transferred out of your facility and is not screened prior to transferring out, exception should be Transfer Only. In this scenario, NEVER mark baby as Finally Screened! Include when you know baby will be or has been screened. This verification is for the NBS follow-up care coordinator.

Finally Screened

- Once the exception is documented as Finally Screened for an exception, this number doesn't tend to go toward your total screened number for that month, although the screen is documented within the exception report.

NICU

- Used when in your facility's NICU, not when transferred out to another facility's NICU.
- Remember that there is a special protocol for screening NICU babies. When admitted in the NICU, the baby should remain as a NICU Exception until released, and then you will promote the status within your MSR.

Initial Screen due Next Month

- Used when there is a delay in screening or when the date and time of birth do not allow for a valid screen to take place within the same month. *Applies to all babies born within the last 6-48 hours of the month and will prompt you to promote the baby in the next MSR.*

Deceased

- Always provide the date and time of death. Sometimes there is a lag in Vital Records, so providing this documentation within the CHIP's General Notes section prevents reaching out to the family with NBS follow-up.

Religious Refusal

- When documenting an RR, you will be prompted to upload the completed and signed form. This form must also be received by ISDH. You can submit via fax or send with your facility's NBS cards to the NBS lab, which will forward them to ISDH.

Discharged without NBS*

- We never want this to occur, but when it does, alert the NBS follow-up care coordinator ASAP, and please document the scenario within the CHIP's General Notes section.

Exception Entry for Pulse Oximetry

The previous exceptions are also utilized as options for the pulse oximetry MSR exceptions. Two additional exception options are valid for pulse oximetry MSRs:

Prenatally/Postnatally Diagnosed with CCHD

- When medical documentation supports this exception option, only this can be selected. When the pulse oximetry is seen to be unnecessary for this reason and an echocardiogram occurs, select this option as well as indicate when an echocardiogram occurred.

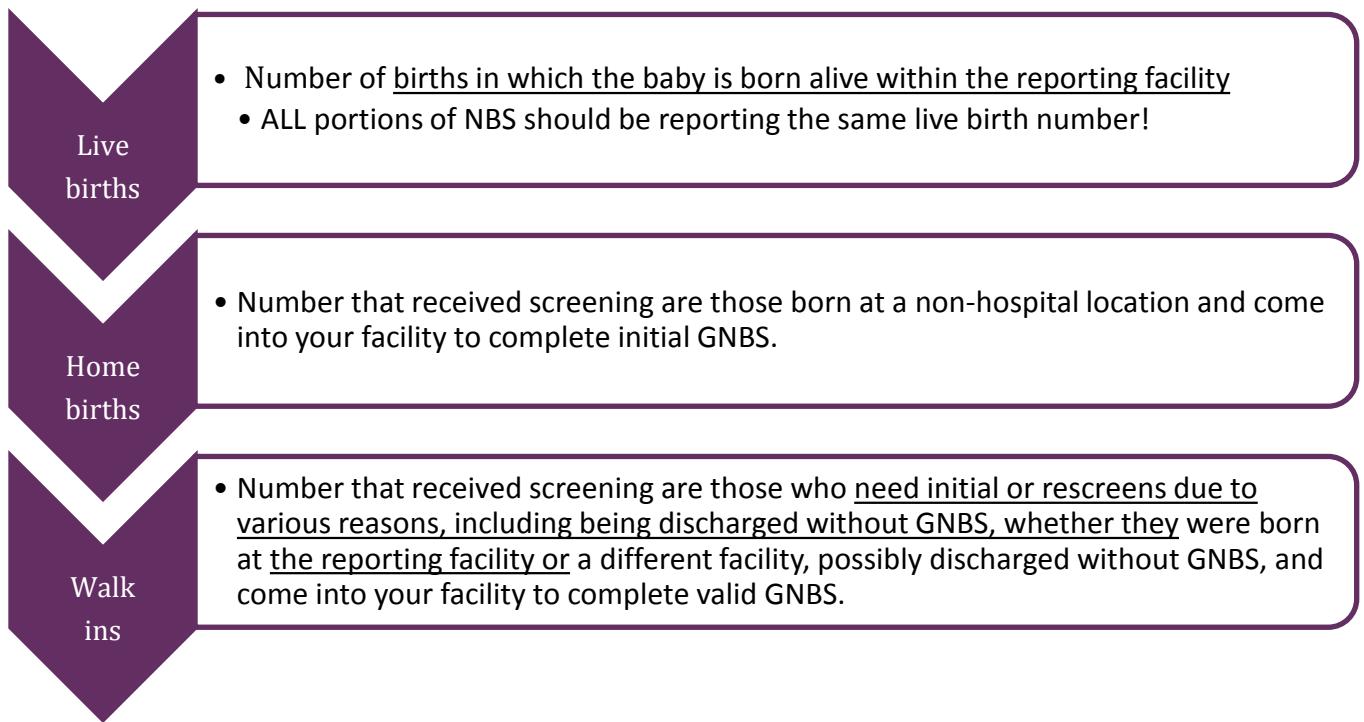
Did Not Pass Pulse Oximetry Screen -- Referred for Additional Follow-Up

- In these scenarios, always indicate if and when the echocardiogram occurred and a brief description in the notes of the exception to support follow-up needs. Please document both pulse oximetry and echocardiogram results when one occurs following a valid failed pulse oximetry screening in order to help with follow-up processes.
- Documentation is done on the NBS card, within INSTEP on their CHIP in the General Notes section, on the Religious Waiver, and or on your MSR submission.

**Please see the Discharged Home without Pulse Oximetry Screen definition on the previous page.*

Summary Data Entry

The numerical value of exceptions you entered into the Exception Entry portion will automatically populate within the table on the MSR Summary Data table. These exceptions, including *Finally Screened*, will deduct from your total number of screens once the following information is manually inputted into the tables, prompting your total number of screens to automatically populate.



Notify the follow-up care coordinator if you view any discrepancies after you have completed your MSR data entry.

Holdovers

YOU HAVE UNRESOLVED HOLDOVERS THAT YOU MUST PROMOTE/RESOLVE BEFORE SUBMITTING YOUR MSR!

When completing an MSR, the submitter could encounter unresolved holdovers that will prevent submission of the report. On the exception entry page a submitter is informed of any holdovers by showing the holdover cases highlighted in blue. These cases will also prompt the submitter to “Promote” the exception type.

Additionally, an alert box will appear on the summary data entry page notifying the MSR submitter of unresolved holdovers because sometimes the exception page may not be viewed when submitting a report, which wouldn’t allow a submitter to be prompted to promote exceptions until attempting to close the MSR.

Even when there are no new exceptions for the current MSR, these are typically from the previous month’s report. The holdovers can often occur when another facility submits its MSR at a later time, causing any incoming or outgoing transfer details to be delayed, such as a transfer in to be accepted or a transfer out being declined by the originally indicated facility.

What is a holdover?

Holdovers are cases that need further attention. Exception types that can become a holdover are the following:

- NICU
- Transfer Only
 - Transfers require the reporter to accept/decline the case at the indicated receiving facility. If the transfer out is declined by the receiving facility, it will bounce back to the submitting facility for resolution.
- Initial Screen Due Next Month

How do I resolve a holdover?

Promote the case by reviewing the health information and then appropriately accepting or declining the transfer detail, updating to Finally Screened, Deceased, or Religious Refusal.

IMPORTANT: If you have questionable or conflicting data within your NBS log or exception entry page, contact the NBS follow-up care coordinator prior to submitting the MSR.



Tips for avoiding MSR Error

Unresolved Holdovers are cases that need further attention. These are highlighted in blue on your exception entry page (even if you have no new exceptions for the month, these are typically from the previous months).

Share this responsibility with a designated backup reporter for your specific screen in addition to the other screen reporters.

- Verify contact info is up-to-date on the MCP Form.

Encourage timely and accurate recording within your facility's NBS log as births and screens occur.

Enter exceptions as they occur to prevent data errors and allow for timely follow-up.

Review your facility's NBS log prior to data submission.

Compare your data with other reporters' data when applicable (i.e., live births).

Ask questions when unsure about data entry or unusual cases by calling ISDH or another reporter within your facility.

Review PIDs and CHIPs for any conflicting data.

- Screenshot errors when noticed and send to ISDH. Alert ISDH as this can be a technical error that will need analyzed and corrected in a timely manner.

Double-check your NBS log and what you entered into your MSR exception page prior to moving forward with the overall summary data entry.

Once live birth numbers are confirmed accurate and inputted into the summary data entry page and you move to the next field, confirm auto-populated numbers are accurate for your facility.

Save and Close the MSR allowing you to submit the report.

Saving will only allow you to return to the entry at a later time.

Check your email for reminders from INSTEP to submit your MSR or that your MSR may be late.

Provide necessary attachments for religious refusal exceptions.

Select the accurate hospital name that will be accepting the transfer.

Immediately notify the ISDH if it is not an option within INSTEP.

ISDH will email and or call you with questions regarding your MSR data entry as needed.

Quality Improvement Initiative

NewSTEPs360

ISDH GNBS Program has revitalized this program with tools and suggestions provided by the NewSTEPs360 program. NewSTEPs is a program of the Association of Public Health Laboratories with an objective to reduce infant mortality and morbidity through GNBS by assisting states in being timely and accurate.

You can view more information on the NewSTEPs program at <http://www.newsteps.org/>.

The mission of this revitalization is to improve timeliness, accuracy, collaboration, education, and reporting between all IN GNBS stakeholders. With this, the GNBS program aims to aid in reducing infant mortality and morbidity through state-mandated GNBS (hearing screen, pulse oximetry, and heel stick).

Birthing Facility Report Cards

The following is an example of the Quality Indicators* that will be used by ISDH GNBS in creating your facility's *report card* and are subject to change. These report cards will be electronically distributed to a designated person and/or group within your facility and identified on the MCP Form monthly and annual reports. The reports cards will be implemented by ISDH in the near future.



*These quality indicators are subject to change.

Tools and Resources for GNBS Stakeholders

This toolkit can serve as a resource when reviewing your facility's policies, procedures, and protocols including your SOPs and COOPs. Updates will be sent electronically to a designated set of contacts to disseminate as needed and to update the hardcopy version received in 2018 during the birthing facility outreach (BFO).

To ensure you receive program updates, please sign up for the [GNBS Newsletter and list serve](#).

All resources are available 24/7 online at www.NBS.IN.gov.

