

**U.S. Department of Health and Human Services
Office of the National Coordinator for Health Information Technology**



**Newborn Screening
Draft Detailed Use Case**

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1.0 Preface

In April and June of 2008, the American Health Information Community (AHIC) approved a recommendation to develop the Newborn Screening Use Case. AHIC specifically requested that the Newborn Screening Use Case focus on: the ability to order and communicate the results from screenings in all six clinical domains – metabolic, hearing, endocrine, hemoglobin, pulmonary/genetic, and congenital infections - and other domains such as galactosemia; the ability to communicate initial screening results, confirmatory testing orders, and results and information specific to referral and management of the patient; the ability to report newborn screening information to public health; and the ability to share de-identified newborn screening information with the clinical research community without requiring additional data collection or data entry. Newborn screening reporting also includes individual case reporting to public health, appropriate registries, and local health service providers.

This use case document is being developed by the Office of the National Coordinator for Health Information Technology (ONC) to represent the AHIC priorities and provide context for the national agenda activities, beginning with the selection of harmonized standards by the Healthcare Information Technology Standards Panel (HITSP). Components that need to be considered during the standards identification and harmonization activities include standardized vocabularies, data elements, datasets, and technical standards that support the information needs and processes of the ordering clinician and receiving laboratory. During the development of the document, there will be an opportunity for review and feedback by interested stakeholders within both the private and public sectors. To facilitate this process, this use case will be developed in two stages. There will be one round of public feedback which will occur prior to the publication of the Detailed Newborn Screening Use Case.

- The **Draft Detailed Use Case** documents all of the events and actions within the use case at a detailed level and facilitates initial discussion with stakeholders; and
- The **Detailed Use Case** documents all of the events and actions within the use case at a detailed level and reflects the feedback received from stakeholders.

This Draft Detailed Use Case is divided into the following sections:

- Section 2.0, Introduction and Scope, describes the priority needs identified by one or more AHIC workgroups and includes initial decisions made regarding the scope of the use case;
- Section 3.0, Use Case Stakeholders, describes individuals and organizations that participate in activities related to the use case and its components;
- Section 4.0, Issues and Obstacles, describes issues or obstacles which may need to be resolved in order to achieve the capabilities described in the use case;



- Section 5.0, Use Case Perspectives, describes how the use case combines similar roles (or actors) to describe their common needs and activities. The roles are intended to describe functional roles rather than organizations or physical entities;
- Section 6.0, Use Case Scenarios, describes how various perspectives interact and exchange information within the context of a workflow. Use case scenarios provide a context for understanding information needs and are not meant to be prescriptive;
- Sections 7.0 and 8.0, provide a greater level of detail for each scenario and include information flows. Specific events and actions for each perspective and scenario are presented and discussed. These are also not intended to be prescriptive;
- Section 9.0, Information Exchange, describes the role of information exchange in the use case at a high level;
- Section 10.0, Dataset Considerations, identifies specific information opportunities relevant to this use case that may support future standardization and harmonization activities; and
- Appendix A, the Glossary, provides draft contextual descriptions of key concepts and terms contained in the detailed use case.



2.0 Introduction and Scope

In June of 2008, the AHIC approved a recommendation to develop a use case addressing newborn screening. The Newborn Screening Use Case is focused on the electronic exchange of information between ordering clinicians, pediatric clinicians, clinical specialists, consumers, public health, testing laboratories, and audiology service providers to support:

- The communication of newborn screening information, orders, and results; and
- The communication of abnormal or out of range results, confirmatory results, and any additional relevant information.

This use case also addresses the potential need for the consumer to receive educational material regarding the screening and/or a suspected or confirmed condition, understand the implications of consent to screening, and provide additional information and/or specimens. The clinician may receive direction on screening requirements, request patient- or test-specific information, order initial tests, receive results, request second specimens, order confirmatory tests, request referrals/interventions, and report public health cases. Public Health may determine and communicate screening requirements, receive and/or process screening orders, receive initial/confirmatory/second specimen results, and track and report long-term outcomes. Audiology Services may receive orders, perform tests, and report results including interpretation and referral recommendations.

In specific terms:

- Clinicians could benefit from electronic communication supporting: the determination of which newborn screening tests are required, the ordering of newborn screening tests, the receipt of newborn screening results, the ordering of genetic/genomic tests (addressed in the 2008 Personalized Healthcare Use Case), the reporting of public health case reports (addressed in the 2008 Public Health Case Reporting Use Case), the requesting of referrals and/or interventions, and the exchange of information to support patient care (addressed in the 2008 Consultations and Transfers of Care Use Case);
- Consumers could benefit from better care, earlier appropriate interventions and increased public safety. The communication of standard comprehensive educational information to consumers could result in them providing more relevant information and greater cooperation and receiving better understanding and improved care;
- Public Health could benefit from electronic communication supporting: the exchange of screening requirements, the receipt of orders, the communication of orders to contracted or third party laboratories, the receipt of laboratory or audiology results, the receipt of public health case reports (addressed in the 2008 Public Health Case Reporting Use Case), and the receipt of information for the purposes of determining long-term outcomes;



- Audiology services could benefit from electronic communication supporting: the receipt and processing of orders, the sending of results, the receipt of additional relevant information regarding the patient and family history.

This use case is focused on the exchange of newborn screening information between providers of care in various settings. This use case is also focused on the communication of relevant information to the consumer, including patients, parents, current guardian(s), surrogates, foster parents, and other caretakers. In particular, the scope of this use case includes an articulation of needs to enable the exchange of information among providers, between providers and public health, and between providers and patients in relation to newborn screening.

The use case identifies information for sharing in a standardized manner during the newborn screening process. This information may include: analytes, conditions, hearing tests/results, dates, and other pertinent data. Specifics regarding datasets, data elements, and nomenclature considerations are addressed in the Datasets Section of this document, as well as the Resource Guide for the Newborn Screening Draft Detailed Use Case (resource guide) which can be found at: <http://www.hhs.gov/healthit/usecases>.

This use case assumes the developing presence of electronic systems such as Electronic Health Records (EHRs), Laboratory Information Systems (LISs), Public Health Systems/Intermediaries, Audiology Systems, Personally Controlled Health Records, and other local or Web-based solutions that support clinicians, consumers, public health, and other healthcare providers while recognizing the issues and obstacles associated with these assumptions.



3.0 Use Case Stakeholders

The Stakeholders section provides a listing of all roles, organizations, groups, and entities involved in the processes described in the use case. Rather than providing a definition for each term, a contextual description is provided. This is intended to allow the reader to understand the terms as they are used within the document.

Figure 3-1. Newborn Screening Stakeholders Table

Stakeholder	Contextual Description
Audiology Service Providers	Professionals engaged in practice to promote healthy hearing, communication, and competency through the prevention, identification, assessment, and rehabilitation of hearing, auditory function, balance, and other related systems. Also serve as a reference for health care professionals, education professionals, consumers, members of the general public, and policy makers.
Clinicians	Healthcare personnel with patient care responsibilities, including physicians, advanced practice nurses, physician assistants, nurses, psychologists, pharmacists, and other licensed and credentialed personnel involved in treating patients.
Consumers	Members of the public that include patients as well as caregivers, patient advocates, surrogates, family members, and other parties who may be acting for, or in support of, a patient receiving or potentially receiving healthcare services.
Electronic Health Record (EHR)/Personal Health Record (PHR) System Suppliers	Organizations that provide specific EHR and/or PHR solutions to clinicians and patients such as software applications and software services. These suppliers may include developers, providers, resellers, operators, and others who may provide these or similar capabilities.
Geographic Health Information Exchange/Regional Health Information Organizations	A multi-stakeholder entity, which may be a free-standing organization (e.g., hospital, healthcare system, partnership organization) that supports health information exchange and enables the movement of health-related data within state, local, territorial, tribal, or jurisdictional participant groups. Activities supporting health information exchanges may also be provided by entities that are separate from geographic health information exchanges/Regional Health Information Organizations including integrated delivery networks, health record banks, and others.



Stakeholder	Contextual Description
Government and Regulatory Agencies	Federal, state, local, territorial, or tribal departments within the United States government responsible for the oversight and administration of a specific function. Government agencies may include: Department of Health and Human Services (DHHS), Food & Drug Administration (FDA), Centers for Disease Control and Prevention (CDC), Centers for Medicare & Medicaid Services (CMS), Health Resources and Services Administration (HRSA), Department of Defense (DoD), Department of Agriculture's Women, Infant, and Children Program (WIC), Administration for Children and Families (AFC), and Indian Health Services (IHS). Examples of regulations include the Clinical Laboratory Improvement Amendments (CLIA) and the Family Educational Rights and Privacy Act (FERPA).
Healthcare Entities	Organizations that are engaged in or support the delivery of healthcare. These organizations could include hospitals, ambulatory clinics, long-term care facilities, community-based healthcare organizations, employers/occupational health programs, school health programs, dental clinics, psychology clinics, care delivery organizations, pharmacies, home health agencies, hospice care providers, and other healthcare facilities.
Healthcare Payors	Insurers, including health plans, Medicaid, self-insured employer plans, and third party administrators, providing healthcare benefits to enrolled members and reimbursing provider organizations.
Laboratory Associations	Advocacy/professional organizations or societies such as the College of American Pathologists (CAP), Association of Public Health Laboratories (APHL), Public Health Informatics Institute (PHII), or the National Committee for Clinical Laboratory Standards (NCCLS), which are concerned with the appropriate use of laboratory technology and interpretation of laboratory information in clinical medicine.
Patient	Members of the public who receive healthcare services. Synonyms may include baby, infant, newborn, client, resident, customer, consumer, and healthcare consumer.
Public Health Agencies	Federal, state, local, territorial, and tribal government organizations and clinical care personnel who exist to help protect and improve the health of their respective constituents.
Research Entities	Organizations that are engaged in or support healthcare research including entities performing research, clinical trials, or other research activities (e.g., National Institutes of Health, academic centers).



Stakeholder	Contextual Description
Registries	Organized systems for the collection, storage, retrieval, analysis, and dissemination of information to support health needs. This also includes government agencies and professional associations which define, develop, and support registries. These may include newborn screening registries, patient registries, and disease registries.
Specialty Healthcare Entities	Organizations that are engaged in or support the delivery of healthcare in specific areas. These organizations could include geneticists, audiologists, pediatric specialists and others focused on specific newborn conditions.
Testing Laboratories	A testing laboratory (often abbreviated lab) is a setting where specimens are sent for testing and analysis and are resulted. Results are communicated back to the requestor. The types of testing laboratories may include clinical/medical and may be either private and/or public entities.



4.0 Issues and Obstacles

Realizing the full benefits of health information technology (HIT) is dependent on overcoming a number of issues and obstacles in today's environment. Examples of specific issues and obstacles that are applicable to the Newborn Screening use case appear below in problem and consequence form:

Information interoperability and exchange:

- There is currently a lack of financial, network, technical, and policy infrastructures to enable information exchange that is secure, consistent, appropriate, reliable, and accurate.
 - Consequently, healthcare facilities (e.g., hospitals, clinics, laboratories, ancillary clinical facilities) may not have the capabilities to electronically collect, process, and transmit newborn screening data in a secure and timely manner. In some instances, third party laboratories may be necessary to conduct newborn screenings for states that do not have sufficient public health testing facilities in place. This significantly limits the effectiveness of an electronic reporting process.
- There may be a need to integrate external screening and related health information into EHRs along with locally captured information.
 - Without integration of external screening information into EHRs and/or PHRs, the implementation of Personal Health Records may be difficult to achieve. Ideally, the information would be able to move interoperably between clinical EHR systems and personal health record systems and/or screening devices.
- The exchange of screening related information across various systems, sites and settings of care may be constrained by lack of agreed upon standards for screening orders and/or results.
 - Without harmonized standards and consistent nomenclature, interoperable systems are difficult to develop. There may be a need to standardize terminology for all screening-related information. Reference the Resource Guide for Newborn Screening contains details about the need for standard terminologies for all screening-related information.
 -
- There may be a need to create a feedback loop that includes distribution of feedback to care-givers of newborns for all screening results (including all normal results).
 - Without the ability to accurately and effectively close the information loop on all newborn screening orders, some results may not be reported to the necessary persons and in some instances, critical information may not make



it back to the clinician or patient. This is an area where effective interoperability may make a significant difference in the successful application of newborn screening initiatives.

Confidentiality, privacy, security, and data access:

- The implementation of genetic/genomic information may create additional risk of misuse of family history, disease risk, and predisposition information. In addition, whenever personal health information is stored, transmitted, archived or destroyed, it must be appropriately secured. Audit trails may be made available.
 - Consumers may lose privacy protection or face unfair consequences (e.g., denial of health insurance or increased premiums) through improper disclosure of family history, disease risk, and predisposition information unless proper safeguards are put in place.
- There may be secondary uses of newborn screening information (e.g., for research or public health use) that are not directly addressed by privacy agreements.
 - Secondary use of data may violate patient privacy and confidentiality.
- In some cases, dissimilar regulations may act as an obstacle to the exchange of newborn screening information, particularly across state boundaries.
 - Patients and providers may not have access to adequate information, thereby preventing appropriate care.
- State and federal laws vary in how they address consent and/or authorization. Therefore, these laws and associated policies need to be analyzed before rules for authorization and consent can correctly protect patient privacy and confidentiality.
 - Without the appropriate guidelines for information access, clinicians and consumers may not be willing to adopt some of the new technologies available for personalized healthcare.
- There are situations of complex current guardian(s)hip which may make the sharing of necessary information difficult. These arrangements may require special agreements involving agencies responsible for the investigation and protection of at-risk children.
 - A child in foster care presents a particularly difficult challenge that may interfere with the timely and accurate reporting of and acting on newborn screening results. These challenges may be exacerbated by the fact that custody arrangements may change over time.

Family health history information interoperability and privacy:



- Information included in a family health history is not always precise. Also, current terminologies do not always incorporate metrics reflecting the level of certainty at which this information can be obtained.
 - Without the adoption of standardized structure and/or form, interoperable systems may be difficult to develop. A document entitled “Family Health History Multi-Stakeholder Dataset Requirements Summary” has been recently published in JAMIA. This document is available for review on the 2008 Personalized Healthcare Use Case website located at <http://www.hhs.gov/healthit/usecases>.
- Advances in the use of genetic and genomic information are being made at a rapid rate. This may cause situations such as new information is discovered about a specific nucleotide sequence after a patient has been diagnosed and treated. Knowledge suppliers could incorporate newborn screening requirements, analytes, and conditions and then communicate them in a timely manner.
 - Without this type of surveillance and communications system in place, patients may not benefit from future advances in genetic/genomic knowledge.



5.0 Use Case Perspectives

The Newborn Screening Draft Detailed Use Case describes the flow of clinical information between providers of care and testing facilities. In this sense, a provider may be an Ordering clinician (as in the case of ordering and resulting) or a Pediatric Clinician (as in the case of abnormal and out of range results). Testing facilities may be a testing laboratory or audiology services. This use case includes five perspectives that are intended to indicate roles and functions rather than organizations or physical locations. Each perspective describes the need for the exchange of clinical information from a particular viewpoint. Each perspective is described below:

Consumer

The consumer perspective includes members of the public who receive healthcare services, as well as caregivers, patient advocates or surrogates, family members, and other parties who may be acting for, or in support of, a patient. For the purposes of the Newborn Screening Use Case, the consumer will always be the advocating party for the infant being screened. This may be a parent, legal guardian, or responsible party in agencies responsible for the investigation and protection of at-risk children.

Ordering clinician

The ordering clinician is directly involved in the care of the infant after birth and during the screening process. This perspective includes family physicians, pediatricians, obstetricians, oncologists, internists, clinical specialists, advanced practice nurses, physician assistants, genetic counselors, medical geneticists, audiologists, psychologists and other personnel involved in Newborn Screening processes. The ordering clinician may be working in a birthing facility, such as a hospital or birthing center or may be involved in a home birth.

Pediatric clinician

This group comprises a wide array of clinical practitioners including family physicians, pediatricians, oncologists, internists, advanced practice nurses, physician assistants, genetic counselors, medical geneticists, audiologists, psychologists and other personnel involved in handling primary, interventional, specialty, or follow-up care of infants whose newborn screening results were abnormal or out of range. The pediatric clinician may also become involved at later stages of the process, including medical home and primary care. Pediatric clinicians may receive or report screening and testing results to or from those performing confirmatory testing or diagnostic evaluations. They may also direct ongoing treatment in response to the screening results.



Testing Facility

The testing laboratory perspective includes medical laboratory personnel such as the laboratory director, laboratory supervisor, laboratory technicians, or other relevant staff. These personnel perform dried blood spot, genetic, or other biochemical analyses. The testing laboratory may receive specimens from birthing facilities, medical home/primary care providers, subspecialty providers, midwives, or public health agencies. They may concurrently report results to primary care providers, subspecialty providers, and/or public health agencies.

Audiology services include hearing tests as part of the Newborn Screening program. They also provide testing results to the same providers and organizations as described above for the Testing Laboratories. The results may be in the form of a narrative consult rather than discrete quantitative data values.

Public Health

The public health perspective includes federal, state, territorial, tribal, and local public health organizations with responsibility to monitor the health status of populations or individuals. This role may also be present within healthcare delivery organizations or other entities having responsibility to monitor the health status of specific populations or individuals. Public health is responsible for abnormal range results reporting, rates for screening results, timeliness of screening, case confirmation, or diagnostic evaluation, and receipt of clinical or early intervention services. Public health should receive screening results and facilitate the multidirectional flow of information through the diagnostic evaluation in order to determine “confirmed” cases and to assure follow-up and treatment.

Information Exchange

The information exchange perspective may include free-standing or geographic health information exchanges (e.g., Regional Health Information Organizations (RHIOs)), integrated care delivery networks, provider organizations, health record banks, public health networks, and specialty networks. These entities may support specific functional capabilities which assist in facilitating health information exchange activities.



6.0 Use Case Scenarios

The Newborn Screening Use Case focuses on two scenarios: Ordering and Resulting; and Abnormal and Out of Range Results.

Scenario 1: Ordering and Resulting

This scenario covers initial screening testing, both for Newborn Dried Blood Spot (NDBS) and Early Hearing Detection and Intervention (EHDI) and ends with the reporting of results, either within normal limits, or notification of the need for confirmatory testing if results are outside of normal limits.

After the parents or current guardian(s) receive the appropriate educational material on the screening testing process (ideally during the prenatal period) and are appropriately consented, the specimen is taken, typically in the form of a blood spot, and the hearing test is performed at the birthing facility. In some instances, these may need to be done at a later time by the pediatric clinician and/or the audiologist. The blood spot is sent to a testing laboratory which may sub-divide the specimen for testing at other facilities. Results of the hearing test and the laboratory tests are reported to the birthing facility, appropriate public health facilities and in some instances, the consumer(s). Result confirmation may be necessary in certain situations. If the results are normal, the Newborn Screening process is complete.

Scenario 2: Abnormal and Out of Range Results

This scenario covers the diagnostic work up for an out of range (or abnormal) screening test either from the NDBS or the EHDI.

When the NDBS result is out of range or abnormal, additional testing may be required. If known and available, the infant's primary care provider is notified, a family history is obtained and in certain situations, a sub-specialist or sub-specialist team may be notified. Consultations may take place between any or all of these parties and the results of any additional or confirmatory testing are reported back to the health department. If the results are confirmed, the child is referred to a specialist where proper counseling and education can be initiated.

When the EHDI result is abnormal, similar processes are initiated. A family history is obtained and confirmatory auditory testing is done. An audiology evaluation may involve referral for molecular genetic testing. Again, consultations may take place between a primary care provider, various specialists and/or the public health department. After confirmation of hearing loss, the audiologist or primary care provider refers the patient to the appropriate specialists and results are reported to the public health department.

This scenario also covers clinical management of children with conditions identified by newborn screening, public health reporting, program monitoring, and health services.



Clinical management is consistent with research derived from outcome data from prior newborn screening.

During the initial follow-up period, children may require a variety of medical interventions, which might include emergency management, nutritional therapy, audiology follow-up and/or management, or other forms of treatment. Information must follow patients as they move between providers of care. New information may need to be added to the patient's EHR in a longitudinal manner. This is particularly important considering the type of information gathered by Newborn Screening since much of it has an impact during childhood development.

Long term follow-up and outcomes should be reported to the health departments at appropriate intervals. Other registries and research organizations may also benefit from receiving this information. Research organizations would receive only de-identified data. This information is important to determine the effectiveness of Newborn Screening processes as well as determining which patients did not receive this screening.



7.0 Scenario 1: Ordering and Resulting

Figure 7-1. Ordering and Resulting

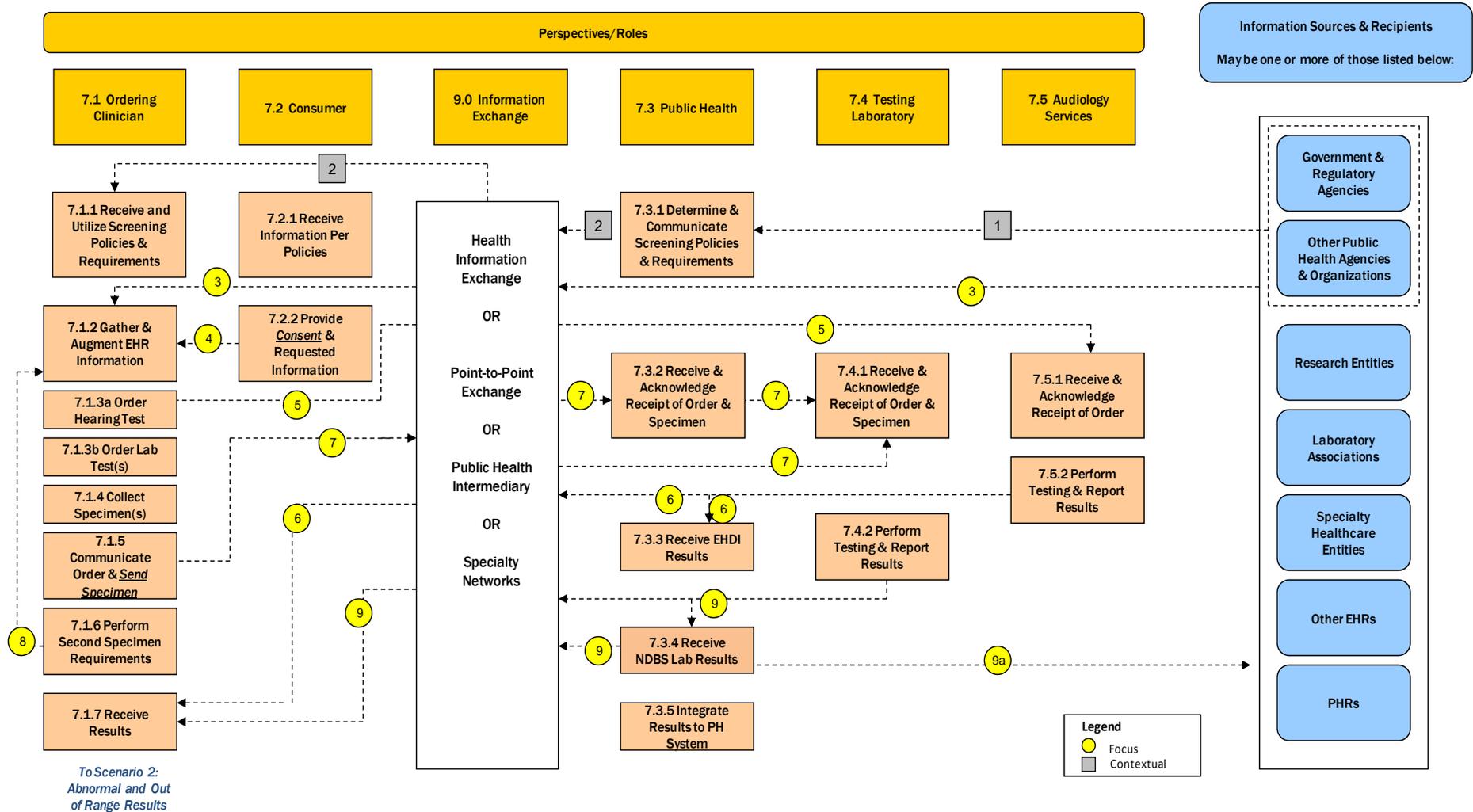




Figure 7-2. Ordering and Resulting Scenario Flows

Scenario 1 – Information Flows

- 1 Information retrieved from various sources by Public Health to help determine newborn screening policies and requirements.
- 2 Information disseminated by Public Health to help birthing facilities and clinicians determine and implement newborn screening policies and requirements
- 3 Patient medical and demographic information
- 4 Additional information regarding the birth and the clinician who will provide care after hospital discharge
- 5 Order and patient information sent to auditory services for EHDI
- 6 Results of hearing test sent back to ordering clinician and/or Public Health, often prior to communicating the Blood Spot order requisition.
- 7a Order and patient information that may accompany the specimen may be sent to Public Health who, in turn, sends it to the testing laboratory. This order often includes the results of the EHDl
- 7b Order and patient information that may accompany the specimen may be sent directly to the testing laboratory. This order often includes the results of the EHDl
- 8 A second specimen may be ordered in certain states and locales. The order and specimen go through the same pathway as the initial specimen.
- 9 Testing results are sent directly from the testing laboratory to the ordering clinician/birthing facility or to Public Health and then transmitted to the ordering clinician.
- 9a Results may be sent to research organizations in de-identified form

Legend

- Focus: Information exchange that is a primary focus of this use case
- Contextual: Information exchange that is not the primary focus of the use case, but is provided for contextual understanding.



Figure 7-3 Screening Test Ordering and Resulting, Ordering Clinician Perspective

Code	Description	Comments
7.1.1	Event: Receive and Utilize Screening Policies & Requirements	Figure 7-1, contextual flow 2
7.1.1.1	Action: Receive and use information regarding NBS to manage program.	The birthing facility receives and maintains policies and procedures for Newborn Screening. These will differ from state to state and are mandated at the state level. This information helps guide the clinical staff in administration of the Newborn Screening program.
7.1.1.2	Action: Utilize information for patient education.	These materials can be used by the clinical staff to educate the patients about what tests will be done and the reason for the screening.
7.1.1.3	Action: Obtain consent from parents/current guardian(s).	Either consent or refusal are typically obtained at this stage and are documented. Some states mandate the consent that is obtained before proceeding.
7.1.2	Event: Gather and Augment EHR Information	Figure 7-1, Focus Flow 4 and Focus Flow 8
7.1.2.1	Action: Obtain key pieces of information. (should there be an additional step related to adding this to the EHR?)	In order for the Newborn Screening process to proceed properly, the clinician(s) at the birthing facility may need to gather certain discrete pieces of information. These data include the pediatric clinician who will provide healthcare for the infant after discharge and certain data elements related to the birth such as gestational age, birth weight and other pertinent data.
7.1.3a	Event: Order Hearing test(s)	Figure 7-1, Focus Flow 5
7.1.3a.1	Action: A clinician at the birthing facility orders EHCI testing.	An order is written for the EHCI. The initial hearing screen is typically performed in the hospital or birthing facility. Results of the EHCI are usually available within 24 hours after birth.



Code	Description	Comments
7.1.3b	Event: Order NDBS screening test(s)	Figure 7-1
7.1.3b.1	Action: A clinician at the birthing facility orders NDBS testing.	The specific tests are mandated (usually at the state level). The resource guide to this use case includes details on NDBS including a matrix showing the intersection of conditions and testing.
7.1.4	Event: Collect Specimen(s)	Figure 7-1
7.1.4.1	Action: A clinician obtains blood and prepares the dried blood spot.	The blood spots are placed on a serial numbered piece of filter paper designed for this specific purpose. An entry is made into a log at the hospital or birthing facility associating the infant and the NDBS specimen in order to facilitate follow-up.
7.1.5	Event: Communicate Order and Send Specimen	Figure 7-1, Focus Flow 7
7.1.5.1	Action: Specimen and order information are sent to Public Health.	The NDBS is sent to the Public Health department along with the test order requisition and all required information. The testing orders include all the metabolic and other lab tests to be performed on the NDBS plus the order for the hearing screen. Information on risk factors may be included as part of the screening information to Public Health. The results of the EHDl are often available and sent along with the test orders and blood spot. This is routine in at least 17 states.
7.1.6	Event: Perform Second Screening Specimen Requirements	Figure 7-1, Focus Flow 8



Code	Description	Comments
7.1.6.1a	Action: Second specimen collected.	In some states and locales a second specimen is required regardless. A second specimen may also be drawn due to circumstances. In some instances, the initial NDBS was collected too early (less than 24 hours after birth), or in other instances, a NDBS was never collected. This specimen must have a unique identifier and a second identifier to ensure that all data generated from laboratory testing are associated with this specimen and not the original specimen.
7.1.6.2	Action: Second hearing screen is ordered.	In some instances, a second hearing test may be necessary. This may be mandated by the state, or may become necessary if the test was either inaccurate, inconclusive, or was never performed. If the infant has already been discharged from the hospital at this point, the testing may be ordered by the Pediatric Clinician and may be done at an auditory services facility outside the original hospital or birthing facility.
7.1.6.1b	Action: Second specimen follows similar pathway to initial specimen.	Any second specimen, if collected, must have a second set of orders associated with it and must then pass through the same pathway as the initial specimen.
7.1.7	Event: Receive Results	Figure 7-1, Focus Flow 6 and Focus Flow 9
7.1.7.1	Action: Clinician or facility receives test results.	The results of the NDBS and EHDI testing are sent to the birthing facility and/or the ordering clinician of record. These results may come directly from the testing laboratory or from the Public Health department. If the results are normal, the Newborn Screening process is complete. If any tests are abnormal or out of range, the process moves to scenario 2.



Figure 7-4 Screening Test Ordering and Resulting - Consumer Perspective

Code	Description	Comments
7.2.1	Event: Receive Information Per Policies	Figure 7-1
7.2.1.1	Action: Consumer receives information regarding policies.	The consumer receives information about the existing policies regarding Newborn Screening. This information may be in the form of a direct consultation with a clinician or may be in the form of printed materials. These materials often originate from the Public Health department. Both the specific policies and the specific tests mandated may vary from state to state. Educational materials may be received by the parents or current guardian(s) during the prenatal period.
7.2.2	Event: Provide Consent and Requested Information	Figure 7-1, Focus Flow 4
7.2.2.1	Action: Consumer provides consent for Screening tests.	In some states, the consumer must consent to Newborn Screening if testing is to be done. Documentation of consent or refusal must be obtained by the hospital or birthing facility.
7.2.2.2	Action: Consumer provides other requested information.	The consumer may need to augment the medical record at the hospital or birthing facility by identifying the pediatric clinician. Also, specific information collected at the birth may need to be identified to augment the screening orders and requisition (e.g., birth weight or complications at birth).



Figure 7-5 Screening Test Ordering and Resulting – Public Health Perspective

Code	Description	Comments
7.3.1	Event: Determine and Communicate Screening Policies & Requirements	Figure 7-1, Contextual Flow 1 and Contextual Flow 2
7.3.1.1	Action: Public Health determines newborn screening policies and requirements.	Public health departments are responsible for the determination and dissemination of Newborn screening policies and requirements. These requirements are mandated at the state level and must be communicated to the birthing facilities and the clinicians providing care in those facilities. This information may be in the form of documents or brochures, or may in certain cases come from direct communication between Public Health and the birthing facilities. In addition, Public Health departments may need to follow-up with infants in the care of agencies responsible for the investigation and protection of at-risk children. These agencies may or may not be under the direct control or guidance of the public health department.
7.3.2	Event: Receive & Acknowledge Receipt of Order and Specimen	Figure 7-1, Focus Flow 7



7.3.2.1	<p>Action: Public health receives and acknowledges the testing order(s).</p>	<p>The order and specimen for metabolic screening testing from the NDBS may be sent through the Public Health department before it is sent on to the testing laboratory. The laboratory may be part of the public health department or a private laboratory that is contracted to perform the testing. In some instances, the order and specimen may be sent directly to the laboratory either in parallel to the health department or as a proxy for the public health department. In some circumstances, the testing laboratory may even be in a different state than the Public Health department. However, the local health department is still responsible for data collection and accurate resulting of the mandated tests in the state where the birth took place.</p>
7.3.3	<p>Event: Receive EHDl Results</p>	<p>Figure 7-1, Focus Flow 6</p>
7.3.3.1	<p>Action: Results of the EHDl are received by Public Health.</p>	<p>Results of the hearing screen may be sent directly to public health from the auditory services facilities at the birthing center.</p>
7.3.3.1a	<p>Alternative Action: EHDl results are received with the NDBS specimen and testing order.</p>	<p>In at least 17 states the results of the hearing screen test are sent to the Public Health department or the Public Health department laboratory along with the testing orders for the NDBS.</p>
7.3.3.1b	<p>Alternative Action: EHDl results are noted on the birth certificate.</p>	<p>In a few states the results of the hearing screen are noted on the certificate of birth which is forwarded to the Public Health Department.</p>
7.3.3.1c	<p>Alternative Action: EHDl results are received at the time of the NDBS results.</p>	<p>In some situations, the results of the hearing screen may not reach Public Health until the results of the laboratory tests from the NDBS have been received.</p>
7.3.4	<p>Event: Receive NDBS Lab Results</p>	<p>Figure 7-1, Focus Flow 9 and Focus Flow 9a</p>



7.3.4.1	Action: Public Health department receives the results of the NDBS testing.	When results are completed by the testing laboratory, a report is received by the Public Health department. The report typically is in the form of positive or negative results, rather than specific values. There may be an advantage for these reports to have the underlying specific data available to the user.
7.3.4.1a	Alternative Action: Public health department receives results from birthing center.	The ordering clinician may have sent the NDBS orders and specimen directly to the testing laboratory. In this situation, the testing laboratory may send the results back to the ordering clinician. The ordering clinician may send the results to Public Health to be recorded.
7.3.5	Event: Integrate Results into Public Health System	Figure 7-1
7.3.5.1	Action: Results are integrated into the public health system.	All results of newborn screening tests including EHD1 and NDBS may be integrated into the data systems within the Public Health department for long term demographic, epidemiologic, or tracking purposes. The results would retain patient identification information at this stage.
7.3.5.2	Action: Results are sent to research entities or other public health agencies or organizations.	The results of the newborn screening tests, both EHD1 and NDBS may be sent to various research entities or other public health agencies (such as the CDC). The data is de-identified at this stage to protect the privacy and confidentiality of the consumer.



Figure 7-6 Screening Test Ordering and Resulting – Testing Laboratory Perspective

Code	Description	Comments
7.4.1	Event: Receive and Acknowledge Receipt of Order and Specimen	Figure 7-1, Focus Flow 7
7.4.1.1	Action: Testing laboratory receives order and NDBS specimen from birthing facility.	A state public health laboratory or a state public health contracted laboratory receives the NDBS and the testing orders with all appropriate information from the birthing facilities.
7.4.1.1a	Alternative Action: Testing laboratory receives order and NDBS specimen from clinician outside the hospital setting.	In the circumstance of a home birth or a birth in a setting other than a state-regulated hospital or other birthing facility, the specimen and order may be sent from the primary care clinician.
7.4.1.2	Action: Specimen is accessioned.	The testing laboratory associates the NDBS with laboratory accession number(s). The testing laboratory ensures that the specimen and information is adequate to perform the tests. If the specimen is deemed adequate, the specimen is divided into multiple aliquots for different types of screening. In some instances, the specimen is sent to a subcontracted testing laboratory for specific tests that are not performed in the primary testing laboratory.
7.4.1.2a	Alternative Action: Specimen is deemed inadequate for testing.	If the specimen is deemed inadequate for any reason, the laboratory informs the ordering clinician, the birthing facility and the parents or current guardian(s) where possible and requests a second screening specimen.
7.4.2	Event: Perform Testing & Report Results	Figure 7-1, Focus Flow 9



7.4.2.1	Action: The laboratory performs the tests.	The testing laboratory performs the complete battery of tests as defined and mandated by each state. These tests and the information needed are described in section 10 of the use case and the resource guide.
7.4.2.2	Action: Report results.	The laboratory reports are sent to the birthing facility, the pediatric clinician (if identified at the time of the specimen collection) and to public health, if the laboratory is not within the public health system. Test results are reported as positive and negative, but there may be a need to develop these reports to have underlying actual lab results available for clinicians. The details of these reports are outlined in section 10 of the use case and in the resource guide.

Figure 7-7 Screening Test Ordering and Resulting – Audiology Services Perspective

Code	Description	Comments
7.5.1	Event: Receive and Acknowledge Receipt of Order	Figure 7-1, Focus Flow 5
7.5.1.1	Action: Audiology receives order for EHDI.	Audiology services within the birthing center are informed of the birth and are given the clinician-generated state-mandated order for hearing screening. (Couldn't the audiology services be somewhere other than in a birthing center? A hospital? A separate clinic?)
7.5.2	Event: Perform Testing & Results	Figure 7-1, Focus Flow 6
7.5.2.1	Action: Audiology performs testing in the birthing center.	Hearing screening may be done at the bedside or in an audiology center within the hospital or birthing center.



<p>7.5.2.2</p>	<p>Action: EHDI results are reported.</p>	<p>The results of the hearing screen are reported to the ordering clinician and may also be reported directly to the Public Health department. In some states, the results of the hearing screen are reported to Public Health along with the order for NDBS screening. In a few states, the results of the EHDI are noted on the birth certificate and, in this manner, the results are communicated to the Bureau of Vital Statistics and Public Health. If the results of the EHDI are within the normal range, the newborn screening process for EHDI is complete. If the results are abnormal or out of range, then the process moves to Scenario 2.</p>
<p>7.5.2.2a</p>	<p>Alternative Action: Second test is required.</p>	<p>If the results of the EHDI are abnormal, inadequate, or ambiguous - or if the test was not performed for some reason, then a second hearing screen is requested and ordered by the hospital, birthing center, or primary care clinician. (see Event 7.1.6).</p>



8.0 Scenario 2: Abnormal and Out of Range Results

Figure 8-1 Abnormal and Out of Range Results

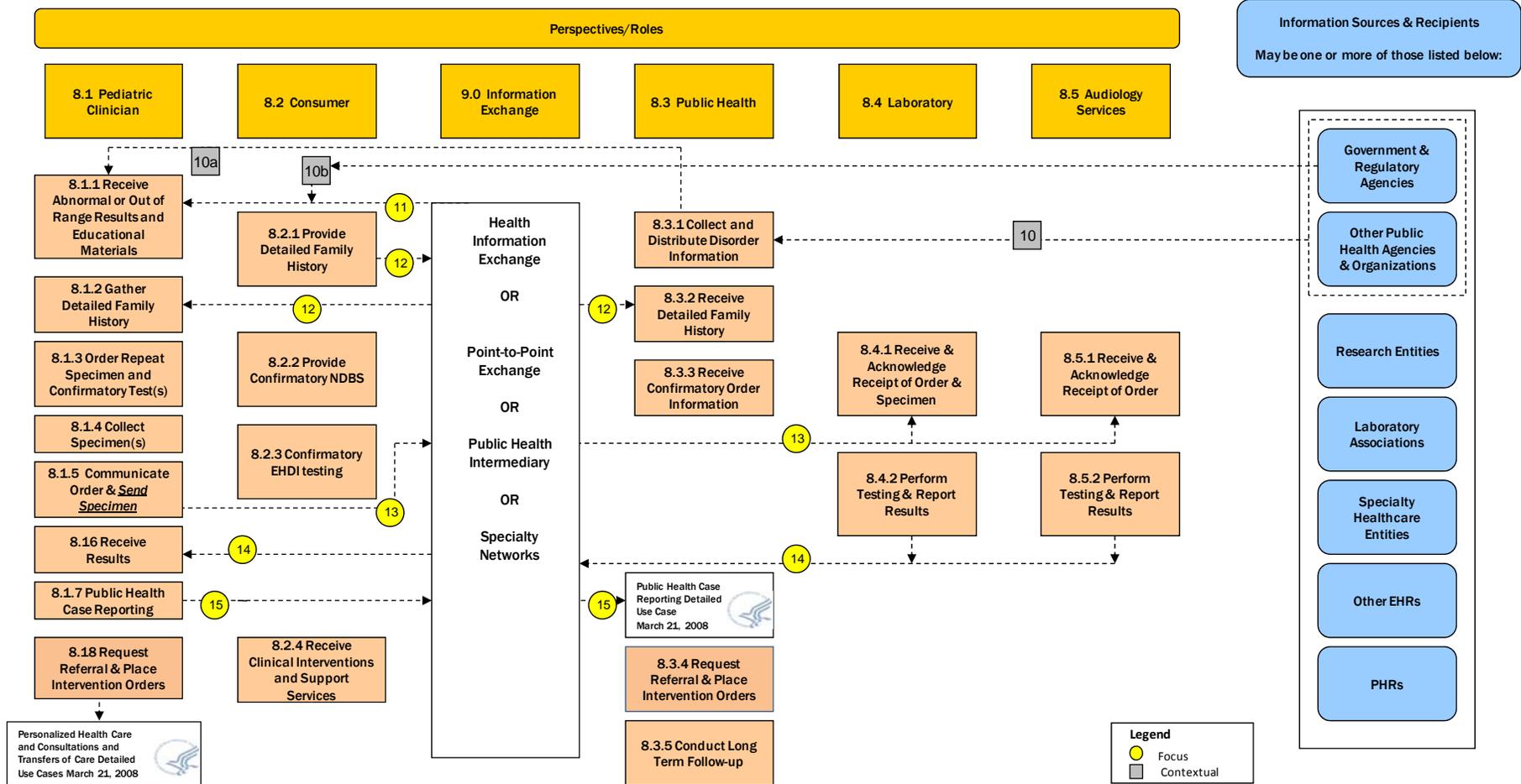




Figure 8-2 Abnormal and Out of Range Results

Scenario 2 – Information Flows

- 10 Information retrieved from various sources by Public Health to help educate and inform the clinicians and consumers about various abnormal or out of range results after screening tests
- 10a Information about specific disorders disseminated by Public Health to help clinicians and consumers understand the meaning of abnormal or out of range results
- 10b Information about specific disorders accessed directly by consumers from the web or web portals
- 11 Abnormal or out of range results diagnosed at the time of the screening test(s) as a continuation from scenario 1
- 12 Detailed family history if necessitated by the specific screening results; this information may be transmitted to the pediatric clinician and/or the public health department
- 13 Orders and specimens required as confirmatory results after the initial screening test(s)
- 14 The testing results are transmitted directly back to the pediatric clinician from the testing laboratory and/or auditory services
- 15 The pediatric clinician reports the case(s) back to Public Health where it is integrated into the Public Health data

Legend

- Focus: Information exchange that is a primary focus of this use case
- Contextual: Information exchange that is not the primary focus of the use case, but is provided for contextual understanding.

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Figure 8-3 Abnormal and Out of Range Results – Pediatric Clinician Perspective

Code	Description	Comments
8.1.1	Event: Receive Abnormal or Out of Range Results and Educational Materials	Figure 8-1, Contextual Flow 10a and Focus Flow 11
8.1.1.1	Action: Clinician receives out of range result.	If the results of the NDBS or the EHDI are abnormal or out of range, a different pathway of activity and information exchange is set into motion. In most instances, the primary actor at this time is the pediatric clinician who is providing care for the infant after discharge from the hospital or birthing center. A specialist team may be informed of the results from the testing laboratory or from auditory services
8.1.1.2	Action: Clinician receives information from Public Health.	The Public Health department may make available information about the disorder implicated by the abnormal or out of range result. These materials are generally educational and may be in the form of brochures, ACT sheets or other similar printed materials. Alternatively, the local Public Health department may have a web portal or other electronic means of delivering this information to clinicians.
8.1.1.3	Action: Clinician obtains information from an online source.	The pediatric clinician may obtain information from any number of public sources independent of the local public health department. These sources may include other public health entities, research entities or other organizations that may disseminate information about the wide array of disorders tested for by the Newborn Screening process.
8.1.2	Event: Gather Detailed Family History Information	Figure 8-1, Focus Flow 12



Code	Description	Comments
8.1.2.1	Action: Gather detailed family history from the consumer by consultative interview.	In response to the abnormal or out of range results from either the NDBS or EHDl testing, the clinician(s) may need to gather a more detailed family history. Currently this history is taken in a direct consultation with the parents or current guardian(s) of the infant. The detailed family history may be helpful to understand the significance of the abnormal or out of range results as well as help to guide further diagnosis and treatment of any possible disorders these results may represent. The detailed family history may also be communicated to the public health department.
8.1.2.2	Alternative Action: Gather detailed family history using electronic tools.	The clinician may gather a more detailed family history by accessing information from the consumer’s EHR or PHR if that is available. As detailed in the 2008 Personalized Healthcare Use Case, a specific tool (“Family History Multi-Stakeholder Workgroup Datasets Requirements Summary”) has been developed for this purpose and could be used in this context. This document can be found on the 2008 Personalized Healthcare Use Case website located at http://www.hhs.gov/healthit/usecases .
8.1.3	Event: Order Repeat Specimen and Confirmatory Tests	Figure 8-1
8.1.3.1	Action: A repeat specimen or repeat EHDl test is ordered.	In response to abnormal or out of range results on either the NDBS or EHDl screening tests, specific test(s) may be ordered to confirm or extend the initial screening results.
8.1.3.2	Action: The pediatric clinician consults with a specialist.	In some circumstances, the pediatric clinician may consult with a specialist or a specialty group to determine the appropriate confirmatory test.
8.1.4	Event: Collect specimen(s)	Figure 8-1



Code	Description	Comments
8.1.4.1	Action: Clinician obtains specimen for confirmatory test(s).	One or more specimens may be obtained for confirmatory testing after an initial abnormal or out of range screening result. The number and type of collection tubes will depend on the disorder(s) being tested and how many testing laboratories may be involved.
8.1.5	Event: Communicate Order and Send Specimen	Figure 8-1, Focus Flow 13
8.1.5.1	Action: The order is written and communicated along with any specimens collected.	If a confirmatory test(s) is/are ordered, the pediatric clinician sends the order, any appropriate information and any specimens directly to the testing laboratory. There may be a need for repeat or confirmatory EHDl testing. This testing may take place within a hospital or at an ambulatory facility independent of the original hospital or birthing center. The specimen(s) may be sent to any testing laboratory capable of performing the necessary test. This laboratory may be a private facility, part of the Public Health system or a laboratory contracted by the public health system.
8.1.6	Event: Receive Results	Figure 8-1, Focus Flow 14
8.1.6.1	Action: The clinician receives confirmatory results from the testing laboratory or auditory services.	The pediatric clinician receives results of the confirmatory testing directly from the testing laboratory or auditory services facility.
8.1.6.2	Action: The Pediatric Clinician receives additional information.	If the results from the confirmatory testing confirm an abnormal or out of range result, the clinician may obtain additional information about the disorder.
8.1.6.3	Action: The clinician refers the consumer to a specialist.	When the pediatric clinician receives confirmatory results following an abnormal or out of range screening result the clinician may refer the child to a specialist and share the results of the initial screening and confirmatory test with the specialist. In the case of certain very rare diseases, the pediatric clinician and the consumer(s) may require additional consultation.



Code	Description	Comments
8.1.6.4	Action: Confirmatory genetic testing was done.	If genetic testing is part of the confirmatory testing, for example, Cystic Fibrosis, specimens are sent to a genetic testing laboratory.
8.1.6.5	Action: Further testing is done to identify genetic sub-types.	For certain genetic disorders, such as Phenylketonuria (PKU), specimens may be sent to a genetic testing laboratory to identify genetic sub-types in order to help direct interventions and treatments.
8.1.7	Event: Public Health Case Reporting	Figure 8-1, Focus Flow 15
8.1.7.1	Action: Pediatric clinician files public health case report.	When the confirmatory testing is completed, and the pediatric clinician has the results of the metabolic or genetic test(s), the confirmatory hearing test or both, the clinician files a case report with the public health department. The details of public health case reporting have been described in the 2008 Public Health Case Reporting Use Case which can be found at http://www.hhs.gov/healthit/usecases .
8.1.8	Event: Request Referral & Place Intervention Orders	Figure 8-1
8.1.8.1	Action: Consumer is referred for education and counseling for a genetic disorder.	If a carrier state has been identified, appropriate education and counseling may be provided. The results are shared with the parents or current guardian(s) and a plan may need to be developed to inform the child when the child is old enough. If congenital hearing loss is confirmed, various molecular DNA genetic tests may be needed to help identify the etiology of the hearing loss. This may require careful coordination between the public health program, the pediatric clinician and in some cases the specialist or sub-specialist. Many of these interactions have been detailed in the 2008 Consultations and Transfers of Care and/ or the 2008 Personalized Healthcare Use Cases which can be found at the following location: http://www.hhs.gov/healthit/usecases .



Code	Description	Comments
8.1.8.2	Action: Consumer is referred for education and counseling for an abnormal confirmatory hearing test.	After diagnostic evaluation, the audiologist determines appropriate follow-up and referrals and forwards results to the public health department. Many states have specific forms and/or procedures for this information exchange. The audiologist may make referrals to a pediatric Ear, Nose and Throat specialist, early intervention services, or back to Audiology for hearing aid/assistive listening device evaluation. Other referrals may take place as well. Again, the general details of these kinds of transfers of care have been detailed in the 2008 Consultations and Transfers of Care and/or the 2008 Personalized Healthcare Use Cases, which can be found at the following location: http://www.hhs.gov/healthit/usecases .
8.1.8.3	Action: Emergency management is required.	If emergency management is required for metabolic conditions, initial therapy is administered in the hospital or emergency department. This may take place immediately following the birth or at some time in the short term following birth.
8.1.8.4	Action: Nutritional intervention is required.	If nutritional intervention is required, parents or current current guardian(s) receive instructions. This may involve several hours or days of in-patient or outpatient visits to appropriately educate the families about any specialized formula preparation or other nutritional instructions. For infants this may require monthly follow-up visits for metabolite, growth and nutritional monitoring.



Code	Description	Comments
8.1.8.5	<p>Action: Children with various disorders require periodic follow-up visits.</p>	<p>Children with disorders discovered from Newborn Screening programs may need periodic follow up visits with their pediatric clinician or the appropriate specialist or sub-specialist. For example, children and adults with inborn errors of metabolism continue to require specialized treatment throughout their lives; children with hearing loss will need to be followed by audiologists and/or otolaryngologists to determine appropriate management (such as hearing aids or cochlear implants); children with endocrine disorders such as hypothyroidism or congenital adrenal hyperplasia will need continuing follow-up by endocrinologists to manage medications. As tests for other disorders are added to the battery of newborn screening tests, or new modalities are implemented, specialized treatment centers may be needed for specialized treatments such as pharmacogenetic management of various disorders.</p>
8.1.8.6	<p>Action: Families are referred for social services.</p>	<p>If appropriate, the family may be referred to specific agencies or organizations for social, early intervention, or special education services. This is especially important for disorders that can be managed with medical, nutritional or lifestyle changes if the family receives related education.</p>



Figure 8-4 Abnormal and Out of Range Results –Consumer Perspective

Code	Description	Comments
8.2.1	Event: Provide Detailed Family History and any appropriate consent	Figure 8-1, Focus Flow 12
8.2.1.1	Action: Parents or current current guardian(s) provide detailed family history and consent for confirmatory testing.	If a newborn screening result is abnormal or out of range, the parents or current guardian(s) of the infant may be asked to give a detailed family history to help identify a specific disorder. This family history, as described in 8.1.2.1 and 8.1.2.2 above, may be obtained through a standard consultation and interview by a clinician; or it may be obtained electronically either from the provider’s or the patient’s EHR or PHR, or the patient may use the “Family History Multi-Stakeholder Workgroup Datasets Requirements Summary” which is described in section 8.1.2.2
8.2.2	Event: Provide Confirmatory NDBS	Figure 8-1
8.2.2.1	Action: Patient provides a blood specimen for confirmation of the blood spot screening test result.	If the newborn screening result from the blood spot is abnormal or out of range, a blood specimen(s) may be required to confirm the result. A standard blood specimen is collected from the infant and used for this confirmatory testing.
8.2.3	Event: Confirmatory EHDI Testing	Figure 8-1
8.2.3.1	Action: Infant undergoes confirmatory hearing test.	If the EHDI is abnormal, out of range, ambiguous or was not done, a second confirmatory hearing test may be performed on the infant. This testing may be performed in the original hospital or birthing center, or may be in another setting. The location for the confirmatory hearing test is independent of the initial EHDI screening.



Code	Description	Comments
8.2.4	Event: Receive Clinical Interventions and Support Services	Figure 8-1
8.2.4.1	Action: Consumer receives referrals for interventions, treatments and other support services.	If the confirmatory testing (hearing or blood) confirms the initial result, the infant and his/her family may be referred for a variety of interventions, medical treatments and/or support services. These referrals are specific to the disorder or hearing loss diagnosed in the above steps. Several circumstances are described above in steps 8.1.8.1 through 8.1.8.6.

Figure 8-5 Abnormal and Out of Range Results – Public Health Perspective

Code	Description	Comments
8.3.1	Event: Collect and Distribute Disorder Information	Figure 8-1, Contextual Flow 10 and Contextual Flow 10a
8.3.1.1	Action: Public Health collects data regarding disorders.	Public Health departments may be responsible for collecting data and disseminating information that pertains to all the disorders tested for during the initial Newborn Screening process. This information may be in the form of published cut sheets, brochures, or other paper derived products, or may be published electronically.
8.3.1.2	Action: Condition information is made available to clinicians.	This information may be made available to clinicians in order to help them educate and inform the patients who are receiving the Newborn Screening services. The clinicians may in turn deliver specific information publications to the family before, during, or after the Newborn Screening process.
8.3.2	Event: Receive Detailed Family History	Figure 8-1, Flow 12



Code	Description	Comments
8.3.2.1	Action: Public Health Receives the detailed family history.	As described in 8.1.2 above, in certain situations after an abnormal or out of range result on the newborn screening tests, a detailed family history is obtained for the family of the infant. This detailed family history may be sent to the public health department either electronically or on paper from the pediatric clinician.
8.3.3	Event: Receive Confirmatory Order Information	Figure 8-1
8.3.3.1	Action: Public Health receives the case report.	When there is an abnormal or out of range result on any of the Newborn Screening tests, the pediatric clinician must send a case report to Public Health. This general process is described in detail in the “Public Health Case Reporting Use Case” which can be found at the following location: http://www.hhs.gov/healthit/usecases .
8.3.4	Event: Request Referral & Place Intervention Orders	Figure 8-1
8.3.4.1	Action:	In certain circumstances, the public health department will request a referral for the infant to consult with a specialist based on the results of a confirmatory test for either hearing loss or a specific metabolic or genetic disorder based on laboratory testing. Public health is also often involved in writing orders for early interventional treatment and/or medical treatment for disorders diagnosed initially from newborn screening.
8.3.5	Event: Conduct Long Term Follow-Up	Figure 8-1



Code	Description	Comments
8.3.5.1	Action: Public Health conducts long term follow up of the screening results.	Following the receipt of the public health case report, as described in the 2008 Public Health Case Reporting Use Case, the public health department conducts long term follow-up on the infants and families that had abnormal or out of range results on their screening reports. Long term outcomes of the screening process may be reported back to the health department.
8.3.5.2	Action: Track outcomes through registries	Various public health related registries can be used to track outcomes and dates of last contact with patients receiving dietary or other management for conditions first identified through Newborn Screening.
8.3.5.3	Action: Hearing detection follow-up.	Long term follow-up of hearing detection may require linkages and reporting from schools or other educational systems. This reporting may be subject to informed consent from the parents or current guardian(s) and compliance with the Family Educational and Privacy Act (FERPA).
8.3.5.4	Action: Information is used to track key parameters of the Newborn Screening process.	Once information has been integrated into the Public Health system, it may be used for tracking several key parameters related to Newborn Screening: 1) tracking and estimating screening rates for both NDBS and EHDI, 2) tracking and confirmation of completion of all recommended confirmatory or diagnostic testing for both NDBS and EHDI, 3) tracking and confirmation of clinical or early intervention service delivery, 4. track the number of infants with conditions identified with Newborn Screening, 5) measure rates of false positives, 6) data may be used to evaluate selection of cut-off points for normal or within/out of range results.
8.3.5.5	Action: Results of Newborn Screening programs reported back to healthcare providers and consumers.	The results of newborn screening programs may be reported back to hospitals, birthing facilities, medical homes, clinicians, early intervention providers and the family.



Code	Description	Comments
8.3.5.5a	Alternative Action: Public Health focuses on unconventional situations.	Public Health may focus and track reporting for unconventional situations of birth or follow up such as: screening of infants not born in hospital facilities, children in foster care, infants born within a state or jurisdiction and receiving follow-up services in another state or jurisdiction.



Figure 8-6 Abnormal and Out of Range Results –Testing Laboratory Perspective

Code	Description	Comments
8.4.1	Event: Receive & Acknowledge Receipt of Order and Specimen	Figure 8-1, Focus Flow 13
8.4.1.1	Action: Testing laboratory receives order and blood specimen from pediatric clinician.	A testing laboratory receives the specimen and the testing orders with all appropriate information from the pediatric clinician. This testing laboratory may be part of the public health department, contracted by the Public Health department or a private laboratory. The laboratory may be a genetic testing laboratory as detailed and described in section 8.1.6.3 and 8.1.6.4.
8.4.1.2	Action: Specimen is accessioned.	The testing laboratory associates the specimen with laboratory accession number(s). The laboratory ensures that the specimen and information is adequate to perform the testing. In some instances, the specimen or a part of the specimen is sent to a subcontracted laboratory for specific tests that are not performed in the primary testing laboratory.
8.4.2	Event: Perform Testing & Report Results	Figure 8-1, Focus Flow 14
8.4.2.1	Action: The laboratory performs the tests.	The testing laboratory prepares for and performs the ordered tests.
8.4.2.2	Action: Results are reported.	The laboratory results are reported to the ordering clinician. The results of confirmatory tests may include a significant interpretive section depending on the complexity of the test. Some of the details of this information exchange are described in the 2008 Personalized Healthcare Use Case which can be found at the following location: http://www.hhs.gov/healthit/usecases .



Figure 8-7 Abnormal and Out of Range Results – Auditory Services Perspective

Code	Description	Comments
8.5.1	Event: Receive & Acknowledge Receipt of Order	Figure 8-1, Focus Flow 13
8.5.1.1	Action: Audiology services receive the order for a confirmatory test.	An audiology evaluation is the primary form of confirmatory hearing tests. If a detailed family history is available, it may be sent with the hearing test order. The Audiology services may be provided in a hospital or ambulatory setting.
8.5.2	Event: Perform Testing & Report Results	Figure 8-1, Focus Flow 14
8.5.2.1	Action: Audiology service performs testing.	A confirmatory hearing test is performed by an audiologist or audiology service facility.
8.5.2.2	Action: Results are reported to ordering clinician.	If a confirmatory hearing test confirms the initial abnormal or out of range result from the EHDI screening process a series of steps are initiated by the audiologist. Determining the etiology of the congenital hearing loss often involves molecular DNA testing and may require careful coordination between the public health program, the Pediatric Clinician, and any number of specialists.
8.5.2.3	Action: Audiologist makes appropriate referrals.	As previously described in 8.1.8.2, after confirmation of an abnormal or out of range hearing test, an audiologist may make referrals to a variety of specialists or sub-specialists depending on the disorder involved. Some of these interactions are generally described in the 2008 “Consultations and Transfer of Care” Use Case which can be found at the following location: http://www.hhs.gov/healthit/usecases .

9.0 Information Exchange

This section highlights selected information exchange capabilities that enable the scenarios described in this use case. These functional capabilities may be provided fully or partially by a variety of organizations including free-standing or geographic health information exchanges (e.g., RHIOs), integrated care delivery networks, provider organizations, health record banks, public health networks, specialty networks, and others supporting these capabilities.

Figure 9-1 Newborn Screening Information Exchange Capabilities

<i>Code</i>	<i>Capability</i>	<i>Comments</i>
9.1	Data delivery – including secure data delivery, data receipt, and confirmation of delivery to EHRs, personally-controlled health records, other systems, and networks	Capability to securely deliver data to the intended recipient and confirm delivery, including the ability to route data based on message content, if required. For example, routing may be applicable to identify the destination testing laboratory which is to receive the newborn screening testing orders.
9.2	Data retrieval – including data lookup, retrieval, and data location registries	Capability to locate and retrieve requested data subject to consumer access decisions and local policies. For example, retrieving the consumer's family health history information involves determining the availability of the requested information as well as delivery to the requestor.
9.3	Subject-data matching	Capability to match available data to the appropriate person during retrieval or routing. For example, when a clinician makes a request for newborn screenings for a specific infant, the systems, processes, and policies facilitating information exchange are utilized to confirm that the data available for retrieval match the person of interest to the clinician.

<i>Code</i>	<i>Capability</i>	<i>Comments</i>
9.4	Data provisioning – including support for secondary uses – data provisioning and distribution of data transmission parameters	Capability to distribute pre-determined data reporting requirements, logical algorithms, vocabularies, guidelines, or similar information to target systems so these systems can implement the associated capabilities. For purposes of this use case, target systems may include EHRs, LISs, and systems involved in information exchange. In some cases, the data transmission parameters include information reporting requirements (e.g., filtering criteria, data to report, vocabularies to use, reporting formats, and destinations). For example, reporting requirements for notifiable diseases could be distributed electronically to systems capable of receiving and implementing them to evaluate data being processed through routine care activities.

While not described in this section, other capabilities could support information exchange including: data integrity and non-repudiation checking; subject and user identity arbitration with like identities during information exchanges; access logging and error handling for data access and exchange; consumer review of disclosure and access logs; and routing consumer requests to correct data.

Health Information Exchange (HIE): For the purpose of this use case, this includes the functional capability to exchange health information between networks in order to support comprehensive health information on individuals. These functional capabilities may be provided fully or partially by a variety of organizations including free-standing or geographic health information exchanges (e.g., RHIOs), integrated care delivery networks, provider organizations, health record banks, public health networks, specialty networks, and others supporting these capabilities.

Specialty Network: Specialty networks may provide all or a portion of the capabilities needed to accomplish the activities involved in the exchange of health information. Specialty networks may focus on the exchange of specific types of health information, may focus on specific patient populations, may focus on the capabilities needed to support specific types of healthcare activities, or may perform a combination of information exchange activities and other services.

Point-to-Point Exchange: For the purposes of this use case, point-to-point exchange includes direct interactions between two systems which do not involve intermediary information exchange functions to route and deliver the data. Representative architectures could include point-to-point messaging, service-oriented-architectures, or information exchange among participants using a common application platform.

10.0 Newborn Screening Dataset Considerations

This section provides a listing of information types that may be relevant for the scenarios previously discussed. The information types shown are not intended to be a comprehensive listing. At this time, there is discussion regarding what might comprise a summary dataset and/or standards for the transfer of appropriate and necessary information to facilitate Newborn Screening between EHRs, PHRs, etc. To date, there is no established common dataset associated with Newborn Screening.

Datasets are still being developed and is expected to be the result of a complementary parallel process involving the various efforts in the industry. The following non-exhaustive information categories and limited examples are for the purposes of addressing the scenarios in this use case.

For Newborn Screening, the following broad categories may be considered:

- Dates and Events
- Birth History
- Newborn Information Required for Screening Order
- Analytes
- Conditions
- Hearing Screenings

Dates and Events for HITSP Consideration may Include:

- Date of Newborn Screening
- Date of Diagnosis
- Diagnosis
- Date of Referral
- Type of Referral
- Date of Enrollment in Treatment Plan/Services
- Type of Treatment Plan/Services

One of the newborn screening recommendations approved by AHIC was to report both the clinical conditions identified and the quantitative analytes measured on newborn screening reports to support both patient focused care and population health activities.

The Personalized Healthcare Workgroup has developed a newborn screening resource guide of cross-mappings. This document is being made available for public feedback as a companion document to facilitate the development of electronic laboratory reports for newborn screening.

The document entitled “Resource Guide for Newborn Screening Draft Detailed Use Case” is available for review and public feedback at the Newborn Screening Use Case website located at <http://www.hhs.gov/healthit/usecases>.

Below you will find a high-level listing which represents the draft hierarchy of analytes and conditions which are expressed in the resource guide.

- Tandem Mass Spectrometry
 - ACMG Primary Targets
 - Amino Acids
 - Fatty Acid Oxidation
 - Organic Acid
 - ACMG Secondary Conditions
 - Amino Acids
 - Fatty Acid Oxidation
 - Organic Acid
 - ACMG Other Conditions
 - Amino Acids
 - Fatty Acid Oxidation
 - Organic Acid
- Non-Tandem Mass Spectrometry Conditions
 - Endocrine
 - Thyroid
 - Adrenal
 - Hemoglobin
 - Hemoglobinopathies

- Hemoglobinopathy Traits
 - Galactose
 - Cystic Fibrosis
 - Infectious Disease
- Early Hearing Detection and Intervention
 - Hearing Loss, Bilateral
 - Hearing Loss, Right
 - Hearing Loss, Left
 - Hearing Loss, Unspecified

11.0 Appendix A: Glossary

These items are included to clarify the intent of this use case. They should not be interpreted as approved terms or definitions but considered as contextual descriptions. There are parallel activities underway to develop specific terminology based on consensus throughout the industry.

AHIC: American Health Information Community; a federal advisory body chartered in 2005, serving to make recommendations to the Secretary of the U.S. Department of Health and Human Services in regards to the development and adoption of health information technology.

Audiology Service Providers: Professionals engaged in practice to promote healthy hearing, communication, and competency through the prevention, identification, assessment, and rehabilitation of hearing, auditory function, balance, and other related systems. Also serve as a reference for health care, education, and other professionals, and for consumers, members of the general public, and policy makers.

Care Coordination: Functions that help ensure that the patient's needs and preferences for health services and information sharing across people, functions, and sites are met over time.

Care Coordinators: Individuals who support clinicians in the management of health and disease conditions. These can include case managers and others.

CCHIT: The Certification Commission for Healthcare Information Technology; is a recognized certification body (RCB) for electronic health records and their networks, and an independent, voluntary, private-sector initiative. CCHIT's mission is to accelerate the adoption of health information technology by creating an efficient, credible, and sustainable certification program.

Clinical Support Staff: Individuals who support the workflow of clinicians.

Clinicians: Healthcare providers with patient care responsibilities, including physicians, advanced practice nurses, physician assistants, nurses, psychologists, pharmacists, and other licensed and credentialed personnel involved in treating patients.

Consumers: Members of the public that include patients as well as caregivers, patient advocates, surrogates, family members, and other parties who may be acting for, or in support of, a patient receiving or potentially receiving healthcare services.

Department of Health and Human Services (HHS): The United States federal agency responsible for protecting the health of the nation and providing essential human services with the assistance of its operating divisions that include: Administration for Children and Families (ACF), Administration on Aging (AOA), Agency for Healthcare Research and Quality

(AHRQ), Agency for Toxic Substances and Disease Registry (ATSDR), Centers for Disease Control and Prevention (CDC), Centers for Medicare & Medicaid Services (CMS), Food and Drug Administration (FDA), Health Resources and Services Administration (HRSA), Indian Health Services (IHS), National Institutes of Health (NIH), Program Support Center (PSC), and Substance Abuse and Mental Health Services Administration (SAMHSA).

Electronic Health Record (EHR): An electronic, cumulative record of information on an individual across more than one healthcare setting that is collected, managed, and consulted by professionals involved in the individual's health and care. This EHR description encompasses similar information maintained on patients within a single care setting (a.k.a., Electronic Medical Record (EMR)).

Electronic Health Record (EHR)/Personal Health Record (PHR) System Suppliers: Organizations which provide specific EHR and/or PHR solutions to clinicians and patients such as software applications and software services. These suppliers may include developers, providers, resellers, operators, and others who may provide these or similar capabilities.

FDA: Food and Drug Administration; a federal agency within the Department of Health and Human Services responsible for the safety regulation of foods, dietary supplements, vaccines, drugs, medical devices, veterinary products, biological medical products, blood products, and cosmetics.

Geographic Health Information Exchange/Regional Health Information

Organizations: A multi-stakeholder entity, which may be a free-standing organization (e.g., hospital, healthcare system, partnership organization) that supports health information exchange and enables the movement of health-related data within state, local, territorial, tribal, or jurisdictional participant groups. Activities supporting health information exchanges may also be provided by entities that are separate from geographic health information exchanges/Regional Health Information Organizations including integrated delivery networks, health record banks, and others.

Government and Regulatory Agencies: Federal, state, local, territorial, or tribal departments within the United States government responsible for the oversight and administration of a specific function; government agencies may include: Department of Health and Human Services (DHHS), Food & Drug Administration (FDA), Centers for Disease Control and Prevention (CDC), Centers for Medicare & Medicaid Services (CMS), Department of Defense (DoD), Department of Veterans Affairs (VA), Indian Health Services (IHS), and Department of Homeland Security (DHS). An example of a regulatory agency is Clinical Laboratory Improvement Amendments (CLIA).

Health Information Exchange (HIE): The electronic movement of health-related data and information among organizations according to specific standards, protocols, and other agreed criteria. These functional capabilities may be provided fully or partially by a variety

of organizations including free-standing or geographic health information exchanges (e.g., Regional Health Information Organizations (RHIOs)), integrated care delivery networks, provider organizations, health record banks, public health networks, specialty networks, and others supporting these capabilities. This term may also be used to describe the specific organizations that provide these capabilities such as RHIOs and Health Information Exchange Organizations.

Healthcare Entities: Organizations that are engaged in or support the delivery of healthcare. These organizations could include hospitals, ambulatory clinics, long-term care facilities, community-based healthcare organizations, employers/occupational health programs, school health programs, dental clinics, psychology clinics, care delivery organizations, pharmacies, home health agencies, hospice care providers, and other healthcare facilities.

Healthcare Payors: Insurers, including health plans, self-insured employer plans, and third party administrators, providing healthcare benefits to enrolled members and reimbursing provider organizations.

HITSP: The American National Standards Institute (ANSI) Healthcare Information Technology Standards Panel; a body created in 2005 in an effort to promote interoperability and harmonization of healthcare information technology through standards that would serve as a cooperative partnership between the public and private sectors.

Laboratory Associations: Advocacy/professional organizations or societies such as the College of American Pathologists (CAP) or the National Committee for Clinical Laboratory Standards (NCCLS) which are concerned with the appropriate use of laboratory technology and interpretation of laboratory information in clinical medicine.

Medical Home: The Medical Home is a concept whereby a patient's primary care physician office would operate as the care coordinator for all the patient's medical conditions and needs. This would include coordinating test results and feedback from the patient's multiple providers and ensuring that the patient's care addressed all co-morbid conditions.

ONC: Office of the National Coordinator for Health Information Technology; serves as the Secretary's principal advisor on the development, application, and use of health information technology in an effort to improve the quality, safety, and efficiency of the nation's health through the development of an interoperable harmonized health information infrastructure.

Patients: Members of the public who receive healthcare services. Synonyms used by various healthcare fields include baby, infant, newborn, client, resident, customer, consumer and healthcare consumer.

Personal Health Information (PHI): PHI is confidential, personal, "identifiable" health information about individuals that is created or received by a health plan, provider, or healthcare clearinghouse and is transmitted or maintained in any form. "Identifiable" means

that a person reading this information could reasonably use it to identify an individual. PHI includes written documents, electronic files, and verbal information. Information from an informal conversation can be considered PHI. Examples of PHI include completed healthcare claim forms, detailed claim reports, explanations of benefits (EOB), and notes documenting discussions with plan participants.

Personal Health Record (PHR): An electronic, cumulative record of health-related information on an individual, drawn from multiple sources, that is created, collected, and managed by the individual or an agent acting for the individual. The content of and rights of access to the PHR are controlled by the individual or agent. The PHR is also known as the electronic Personal Health Record (ePHR).

Providers: The healthcare clinicians within healthcare delivery organizations with direct patient interaction in the delivery of care, including physicians, nurses, psychologists, and other clinicians. This can also refer to healthcare delivery organizations.

Public Health Agencies/Organizations (local/state/territorial/federal): Federal, state, local, territorial, and tribal government organizations and clinical care personnel that exist to help protect and improve the health of their respective constituents.

Registries: Organized systems for the collection, storage, retrieval, analysis, and dissemination of information to support health needs. This also includes government agencies and professional associations which define, develop, and support registries. These may include emergency contact information/next of kin registries, patient registries, and disease registries.

Specialty Healthcare Entities: Organizations that are engaged in or support the delivery of healthcare. These organizations could include geneticists, audiologists, pediatric specialists, and others providing health services specific to newborn conditions.

Testing Laboratories: A testing laboratory (often abbreviated lab) is a setting where specimens are sent for testing and analysis, are resulted, and then results are communicated back to the requestor. The types of testing laboratories may include clinical/medical, environmental, and veterinarian, and may be both private and/or public.