U.S. Department of Health and Human Services

Office of the National Coordinator for Health Information Technology

Newborn Screening

AHIC Detailed Use Case

December 31, 2008

Table of Contents

List of Figures

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. This Newborn Screening Use Case document has been 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 clinicians, consumers, testing facilities, and Public Health. AHIC specifically requested that the Newborn Screening Use Case focus on the ability to order and communicate the results from screenings in various clinical domains.

This 2009 Use Case has been developed by ONC with previous opportunities for review and feedback by interested stakeholders within both the private and public sectors. To facilitate this process, the use cases have been developed in two stages:

A. 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

B. 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 document is the Detailed Use Case. Feedback received on the Draft Detailed Use Case has been considered and incorporated where applicable into this document. HITSP is expected to reuse standards, where applicable, from standards previously recognized by the Secretary of Health and Human Services, to specify and constrain how standards are to be used to advance interoperability and to work with standards development organizations to see that gaps in standards are filled.

This 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 contextual descriptions of key concepts and terms contained in the detailed use case.

2.0 Introduction and Scope

The Newborn Screening (NBS) Use Case is focused on the electronic exchange of information related to newborn screening among ordering clinicians, pediatric clinicians, consumers, Public Health, testing laboratories, and audiology service providers. Newborn screening reporting and information exchanges may also include individual case reporting to Public Health, appropriate registries, and local health service providers. The details about Public Health information exchanges can be found in the 2008 Public Health Case Reporting Use Case.

The scope of this 2009 use case is focused on:

- The ability to communicate initial screening results, confirmatory testing orders, and results and information specific to referral and management of the patient; and
- The ability to report newborn screening information to Public Health.

NBS information exchanged to support the focus of this use case consists of all newborn screening orders, results, and information associated with an order or result. Result information may include normal, abnormal, out of range, and confirmatory results within the domains of hearing, hemoglobin, metabolic, and pulmonary/genetic screening tests. The newborn screening domains that are out of scope in this use case are endocrine, congenital infections, and other domains such as enzyme disorders.

This use case highlights the ability to share de-identified newborn screening information with the clinical research community without requiring additional data collection or data entry. This use case also identifies information exchanges that begin with initial newborn screening and extend to include portions of Long Term Follow-Up (LTFU). The information exchanged may include analytes, conditions, hearing tests/results, dates, and other pertinent data. The Personalized Healthcare Workgroup has developed a newborn screening reference of cross-mappings as a companion document to the use case to facilitate the development of electronic laboratory reports for newborn screening. The document entitled “Newborn Screening Coding and Terminology Guide” is available at the Newborn Screening Use Case website located at http://www.hhs.gov/healthit/usecases/nbs.html. Specifics regarding datasets, data elements, and nomenclature considerations are addressed in the Dataset Considerations section of this document, as well as in the Newborn Screening Coding and Terminology Guide.

Recognizing that states may utilize various processes, require different testing, and gather variable data when conducting NBS, these processes are generalized throughout this use case in order to achieve further standardization of information exchange. Further, this document does not prescribe policy or dictate process.

To effectively complete NBS and support communication among clinicians, consumers, Public Health, and the testing laboratory and audiology services, this use case describes some specific information exchanges.

Examples of specific information exchanges:

A. 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.

   It would be beneficial to clinicians to have 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).

B. The use case addresses the potential need for the consumer to be informed and educated about the NBS process, the implications of consenting to screening, and the potential need to provide additional information and/or specimens.

   Consumers could benefit from better care, which includes earlier appropriate interventions and receiving educational material that is audience specific and culturally appropriate regarding the screening and/or a suspected or confirmed condition. 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.

C. 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.

   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.

D. The Testing Laboratory and Audiology Services may receive orders, perform tests, and report results including interpretation and referral recommendations.

   Testing Laboratory and 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 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.

Identification, development, and harmonization of standards to support the interoperability associated with newborn screening are addressed in this document. Work with standards and professional organizations, care delivery organizations, and organizations providing information technology services and products to the healthcare industry is needed to support the interoperability needs associated with newborn screening. As mentioned in Section 1.0, the needs expressed here have not yet been fully addressed by the national health agenda's standardization efforts. Examples of gaps in industry standards are outlined in the upcoming sections of this use case document.

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	Clinicians 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 consultant 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, midwives, neonatologists, audiology service providers, physician assistants, nutritionists, nurses, genetic counselors, 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, emergency contacts, 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.

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), Maternal and Child Health Bureau (MCHB), Administration for Children and Families (AFC), the Agency for Healthcare Research and Quality (AHRQ), and Indian Health Services (IHS). Other agencies include state and county level Public Health Departments. Examples of regulations include the Clinical Laboratory Improvement Amendments (CLIA), the Health Insurance Portability and Accountability Act (HIPAA), and the Family Educational Rights and Privacy Act (FERPA).
Health Information Exchange 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 health information exchange organizations including integrated delivery networks, health record banks, and others.
Healthcare Entities	Organizations that are engaged in or support the delivery of healthcare. These organizations could include hospitals, birthing centers, 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, State Public Health Agencies, and third party administrators, providing healthcare benefits to enrolled members and reimbursing provider organizations.
Knowledge Suppliers	Entities that use data, vocabulary, technology, and/or industry standards to provide information and tools to entities delivering health care. Examples of newborn screening knowledge suppliers include the American College of Medical Genetics (ACMG), the American Academy of Pediatrics (AAP), and the American College of Obstetrics and Gynecology (ACOG).

Stakeholder	Contextual Description
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 Clinical and Laboratory Standards Institute (CLSI), which are concerned with the appropriate use of laboratory technology and interpretation of laboratory information in clinical medicine.
Laboratory Information System (LIS) Suppliers	Organizations which provide specific laboratory information system solutions. A laboratory information system is a class of software which handles receiving, processing, transmitting, and storing information generated by medical laboratory processes. These systems often must interface with instruments and other information systems such as hospital information systems. An LIS is a highly configurable application which is customized to facilitate a wide variety of laboratory workflow models.
Patients	Members of the public who receive healthcare services. Synonyms may include baby, child, 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.
Public Health Systems Suppliers	Organizations which provide specific Public Health 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.
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).
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, long-term follow up patient registries, death registries, and disease registries.
Social Service Agencies	Agencies such as social services and law enforcement that help facilitate the newborn screening process and provide information and support for patients and consumers.

Stakeholder	Contextual Description
Specialty Healthcare Entities	Organizations that are engaged in or support the delivery of healthcare in specific areas. These organizations could include geneticists, genetic counselors, audiologists, metabolic dieticians, speech pathologists, neurologists, occupational therapists, 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 screening, confirmatory 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 are discussed in this section:

State-to-state variation:

A. Newborn screening processes vary significantly from state to state. State-to-state variation not only exists between processes, but also in the testing required and data gathered.

B. The long-term follow-up (LTFU) activities that may occur after newborn screening in many cases do not follow standardized policies and procedures. There is significant variation of these processes from state-to-state.

Information interoperability and exchange:

A. There is currently a lack of financial, network, technical, and policy infrastructures to enable information exchange that is secure, consistent, appropriate, reliable, and accurate.

   i. 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.

B. Newborn screening results and related health information are not currently integrated into EHRs.

   i. If the newborn screening data does not get incorporated into the EHR, then there may be no follow up initiated. Ideally, information should be interoperable between EHR systems and/or screening devices (e.g. equipment used for audiology evaluation) so that optimal information exchange can be achieved. Specifically the information to be shared should be represented such that context and meaning can be shared. Hence, the terminologies and structure should be interoperable for the information wherever captured.

C. The exchange of screening related information among various systems, sites and settings of care may be constrained by lack of agreed upon standards for newborn screening orders and/or results.

   i. Lack of harmonized standards including consistent terminology and nomenclature used to exchange newborn screening data hampers interoperable exchanges of that information. There may be a need to standardize terminology for all newborn screening related information. Reference the companion document Newborn Screening Coding and Terminology Guide, which contains details about the need for standard terminologies for all screening-related information.

D. There is a need for distribution of feedback to care-givers that includes screening results, (including all normal and negative results).

   i. Without the ability to accurately and effectively communicate the results of all newborn screening orders, some results may not be reported to the necessary persons and in some instances, key information may not make it back to the clinician or patient.

E. At present, there is limited electronic exchange of information between vital statistics information systems, clinical EHRs, and Public Health information systems.

   i. Incorporating newborn screening data into vital health statistics information systems presents an opportunity to better ensure that fewer newborns are overlooked and consequently that all are screened.

Confidentiality, privacy, security, and data access:

A. While the Public Health population screening program has been in existence for several decades, the incorporation of genetic/genomic information into records may create additional fear 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. Consumers need the ability to receive information about who has access to their information and for what purpose.

   i. Consumers fear the loss of privacy protection and unfair consequences (e.g., denial of health insurance or increased premiums) through improper disclosure of family history, disease risk, and predisposition information unless appropriate policies are put in place.

B. There may be secondary uses of newborn screening information (e.g., for research or Public Health use) that are not directly addressed by current privacy agreements.

   i. Secondary use of data may violate patient privacy and confidentiality.

C. In some cases, dissimilar regulations within jurisdictions may act as an obstacle to the exchange of newborn screening information, particularly across state boundaries.

   i. Patients and providers may not have access to adequate information, thereby preventing appropriate care.

D. State and federal laws vary in how they address consent and/or authorization. In fact, states also vary regarding informing parents prior to newborn screening. Therefore, these laws and associated policies need to be considered before authorization and consent convention can adequately protect patient privacy and confidentiality.

   i. 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. Without the appropriate education for clinicians who

in turn educate consumers regarding newborn screening, consent and/or authorization may not as readily be given hindering newborn screening effectiveness. With the passage of the Genetic Information Non-discrimination Act (GINA) of 2007/2008, some of these issues may be mitigated.

E. There are situations of complex guardianship which may make obtaining of accurate and timely information difficult. These arrangements may require special agreements involving agencies responsible for the investigation and protection of at-risk children.

   i. An adopted child or 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:

A. Information included in a family health history is not always precise. Also, family histories that are collected by clinicians do not always reflect the level of detail and certainty at which this information can be obtained in a more detailed genetic consultation.

   i. Without the adoption of standardized structure and/or form, interoperability may be difficult to achieve. The Family Health History Multi-Stakeholder Workgroup of the American Health Information Community recently published a document entitled “Family Health History Multi-Stakeholder Dataset Requirements Summary” in the Journal of the American Medical Informatics Association. This document is available for review on the 2008 Personalized Healthcare Use Case website located at http://www.hhs.gov/healthit/usecases.

B. Advances in the use of genetic and genomic information are being made at a rapid rate and may prevent clinicians from incorporating this new information into their clinical workflow.

   i. This delay may cause situations where new information is discovered about a specific nucleotide sequence after a patient has been diagnosed and treated and not incorporated into their current treatment plan. Knowledge suppliers could incorporate newborn screening requirements, analytes, conditions, and treatments 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 2009 Newborn Screening Detailed Use Case describes the flow of clinical information between providers of care and testing facilities. In this context 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 include 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, neonatologists, midwives, oncologists, internists, clinical specialists, advanced practice nurses, physician assistants, genetic counselors, medical geneticists, audiologists, and all other clinical 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 perspective comprises a wide array of clinical practitioners including family physicians, pediatricians, neonatologists, midwives, oncologists, internists, advanced practice nurses, physician assistants, genetic counselors, medical geneticists, audiologists, and all other clinical personnel involved in handling primary, interventional, specialty, or follow-up care of infants regardless of the results of the screen. The pediatric clinician may only become involved during intervention and treatment. 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 Laboratory

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, ordering clinicians, subspecialty providers, midwives, or Public Health agencies. They may concurrently report

results to primary care providers, subspecialty providers, and/or Public Health agencies. Testing may involve concurrent testing being done by several different labs. Screening and confirmatory testing may be done in the same laboratory or different laboratory depending on the particular locale.

Audiology Services

Audiology services include hearing evaluations 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.

Public Health

The Public Health perspective includes federal, state, local, territorial, and tribal Public Health organizations with responsibility to monitor the health status of populations or individuals. This role may also be performed within healthcare delivery organizations or other entities having responsibility to monitor the health status of specific populations or individuals. Public Health may be involved in reporting results, determining screening rates, timeliness of screening, diagnostic evaluation outcomes, and receipt of clinical or early intervention services. Public Health receives screening results and facilitates the multidirectional flow of information through the diagnostic evaluation in order to determine "confirmed" cases (as discussed in the 2008 Public Health Case Reporting Use Case), and to assure short term and long term follow-up and treatment. The use case acknowledges the existence and important role of Short-Term Follow-Up (STFU) teams as an important activity of Public Health departments.

Information Exchange

The information exchange perspective may include health information exchange organizations (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 Detailed 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 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 informed and/or consented the newborn screening testing is performed.

For metabolic testing, a blood specimen is taken, typically in the form of a blood spot on specially designed filter paper. In some instances, this may need to be done at a later time by the pediatric clinician. The blood spot is sent to a testing laboratory which may sub-divide the specimen for testing at other facilities. Results of the metabolic screening are reported to the birthing facility, appropriate Public Health facilities, the ordering and pediatric clinician and in some instances, the consumer(s) who are notified by either the ordering or pediatric clinician. A second specimen may be required in some states and/or for particular circumstantial reasons.

The hearing test (EHDI) is performed at an audiology center associated with the birthing facility. Results of the hearing test are reported to the birthing facility, appropriate Public Health facilities, the ordering and pediatric clinician and in some instances, the consumer(s) who are notified by either the ordering or pediatric clinician.

Result confirmation may be necessary in certain situations. Upon the communication of normal results, the newborn screening process is complete.

Scenario 2: Abnormal and Out of Range Results

This scenario covers the processes in response to an out of range (or abnormal) screening test either from the NDBS or the EHDI.

When the NDBS result is indeterminate, of insufficient quantity, 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 may be 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 clinicians and the results of any additional or confirmatory testing are reported back to Public Health. If the results are confirmed, the infant is referred to a specialist where culturally competent or appropriate 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 genetic consultation which may in turn lead to various kinds of 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 Public Health.

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 standard of care and, when available, research derived from outcome data from prior newborn screening.

During the short term follow-up (STFU) 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. Pertinent clinical findings must follow patients as they move between clinicians as described in the 2008 Consultations and Transfers of Care Use Case. As described in the Dataset Considerations section of this document, additional information may be needed in addition to the information needs described in previous use cases. 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 (LTFU) and outcomes should be reported to Public Health at appropriate intervals. This information is important to determine the effectiveness of newborn screening. Other registries and research organizations may also benefit from receiving this information. Research organizations would receive only de-identified data.

7.0 Scenario 1: Ordering and Resulting

Figure 7-1. Ordering and Resulting

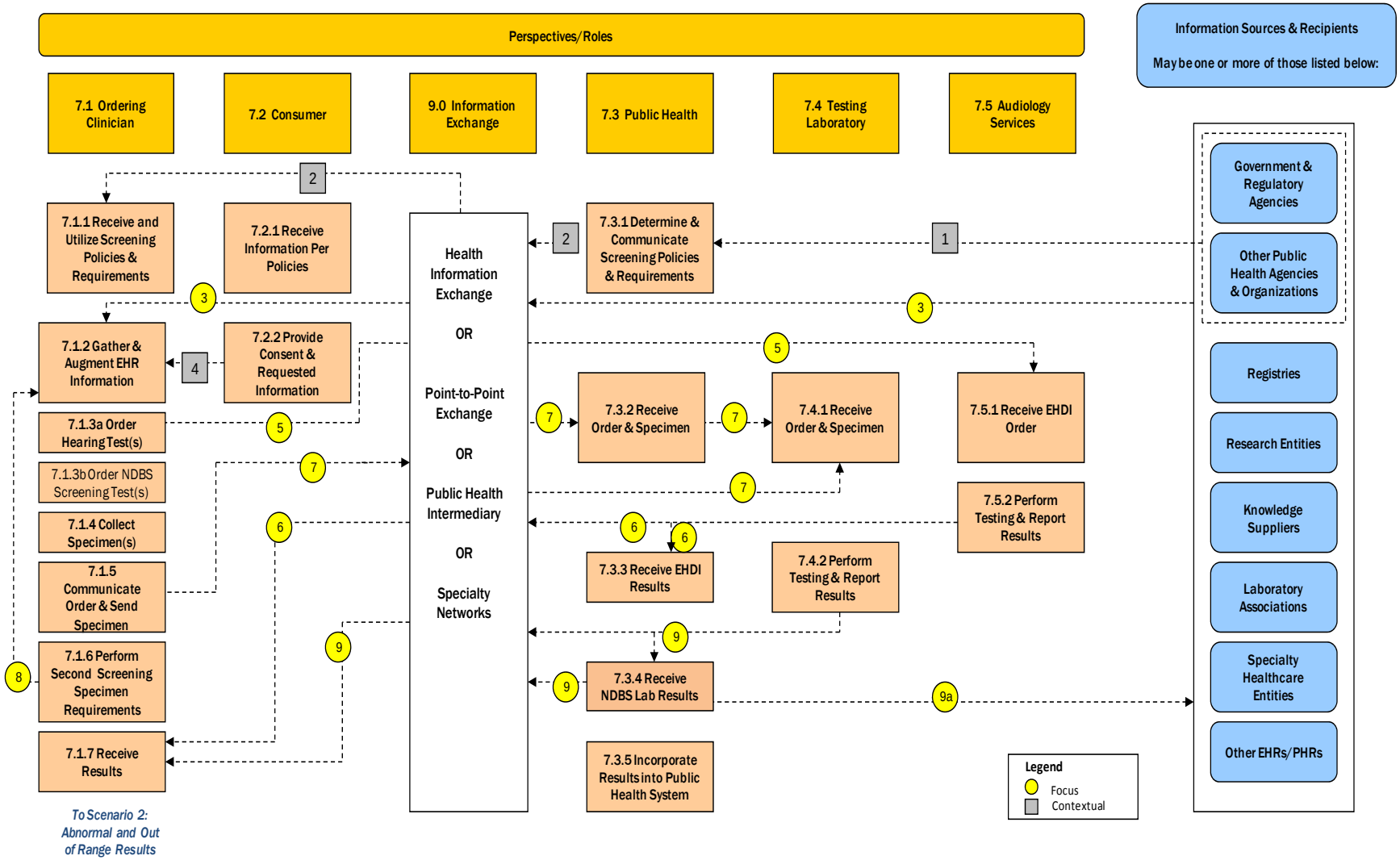

Figure 7-2. Ordering and Resulting Scenario Flows

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 sent to Ordering Clinician from the EHR.
4. Gather and augment EHR information within.
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.
7. Order and patient information that may accompany the specimen may be sent to Public Health who, in turn, sends it to the testing laboratory. In some instances, the order and patient information, and specimen may be sent directly to the testing laboratory. This order often includes the results of the EHDI.
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 newborn screening to manage screening 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:** Inform patients and/or obtain consent from parents/current guardian(s).	In most states and locales informing the consumers prior to the newborn screening is considered sufficient. However, in certain states consent or refusal are obtained at this stage and are documented. Some states mandate that consent is obtained before proceeding.
7.1.2	**Event:** Gather and Augment EHR Information	**Figure 7-1, Focus Flows 4 and 8**
7.1.2.1	**Action:** Obtain key pieces of information.	Clinician(s) at the birthing facility may need to gather certain discrete pieces of information and incorporate into patient record in EHR. 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 the date and time of the birth, gestational age, birth weight, ethnicity and other pertinent data. In some cases, the pediatric clinician who will provide care for the infant after discharge is not known. In certain settings and in some states there are methods such as web sites in place for a pediatric clinician to obtain the results of the screening at a later date. There may be a need for capture of demographic information that changes after hospital discharge, such as surname of infant.
7.1.3a	**Event:** Order Hearing test(s)	**Figure 7-1, Focus Flow 5**

Code	Description	Comments
7.1.3a.1	**Action:** A clinician at the birthing facility orders EHDI testing.	An order is written for the EHDI. The initial hearing screen is typically performed in the hospital or birthing center. Results of the EHDI are usually available within 24 hours after birth. Patient attributes should travel along with order and result. Many of these interactions have been detailed in the 2008 General Lab Orders Use Case which can be found at the following location: http://www.hhs.gov/healthit/usecases.
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 Newborn Screening Coding and Terminology Guide to this use case includes details on NDBS testing including a matrix showing the intersection of conditions and testing required. Many of these interactions have been detailed in the 2008 General Lab Orders Use Case which can be found at the following location: http://www.hhs.gov/healthit/usecases.
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 center associating the infant and the NDBS specimen in order to facilitate follow-up. This log may be kept on paper or may be kept electronically based on the standard operating procedures of the particular institution. It is important to note the time and date of birth along with the time and date of the specimen, since in some cases, such as in 7.1.4.1a below, the time of the blood draw may be non-standard.

Code	Description	Comments
7.1.4.1a	**Alternative Action:** Specimen obtained at a non-standard time, due to medical or logistical reasons.	In most situations, infants who are born prematurely or are cared for in the Neonatal Intensive Care Unit (NICU) for any other reason will have their blood drawn on day 1 and sometimes day 3 of life. However, since this is not always the case, this raises the possibility that the screening is not done. In addition, NICU babies may be transferred to and from different hospitals. Generally, the pediatric clinician who is providing care for the infant, just as in a midwife or home birth, is responsible for obtaining the blood spot and referring the family for any other relevant screening when it is deemed appropriate.
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 either the testing laboratory or Public Health.	The NDBS is either sent to the Public Health department or directly to the testing laboratory along with the test order requisition and all required information. The orders for laboratory results 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 EHDI are often available and sent along with the test orders and blood spot. Many of these interactions have been detailed in the 2008 General Lab Orders Use Case which can be found at the following location: http://www.hhs.gov/healthit/usecases.
7.1.6	**Event:** Perform Second Screening Specimen Requirements	**Figure 7-1, Focus Flow 8**

Code	Description	Comments
7.1.6.1	**Action:** Second specimen collected.	In some states and locales a second specimen is always required. A second specimen may also be drawn due to circumstances at the request of the clinician such as a medical condition taking precedence. In some instances, the initial NDBS was collected too early (less than 24 hours after birth), or in other instances, an initial NDBS was never collected. Methods must be used to ensure that a specimen can be unambiguously identified and a second sample/specimen identifier to ensure that all data generated from laboratory testing are associated with this repeat specimen and not the original specimen.
7.1.6.2	**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.
7.1.6.3	**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 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 center.
7.1.7	**Event:** Receive Results	**Figure 7-1, Focus Flows 6 and 9**
7.1.7.1	**Action:** Clinician or birthing facility receives test results.	The results of the NDBS and EHDI testing are sent to the birthing facility and/or the ordering clinician. These results may come directly from the testing laboratory or from the Public Health. If the results are normal, the parent or current guardian(s) are informed and 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 parents or current guardian(s) 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 Public Health. 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, if required by state	**Figure 7-1, Contextual Flow 4**
7.2.2.1	**Action:** Consumer is informed and may provide consent for screening tests.	In most states and locales informing the consumers prior to the newborn screening is considered sufficient. However, in certain states consent or refusal are obtained at this stage and are documented. Some states mandate that consent is obtained before proceeding.
7.2.2.2	**Action:** Consumer provides other requested information.	The consumer may need to augment the medical record at the hospital or birthing center by identifying the pediatric clinician.

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

Code	Description	Comments
7.3.1	**Event:** Determine & Communicate Screening Policies & Requirements	**Figure 7-1, Contextual Flows 1 and 2**
7.3.1.1	**Action:** Public Health determines newborn screening policies and requirements.	Public Health determines and disseminates newborn screening policies and requirements. These state level requirements are 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.
7.3.2	**Event:** Receive Order and Specimen	**Figure 7-1, Focus Flow 7**
7.3.2.1	**Action:** Public Health receives the testing order(s).	The order and specimen for metabolic screening testing from the NDBS may be sent through Public Health before it is sent on to the testing laboratory. The laboratory may be part of Public Health 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 Public Health or as a proxy for Public Health. In some circumstances, the testing laboratory may even be in a different state than Public Health. However, the local health department may be responsible for data collection and accurate resulting of the mandated tests in the state where the birth took place.
7.3.3	**Event:** Receive EHDI Results	**Figure 7-1, Focus Flow 6**
7.3.3.1	**Action:** Results of the EHDI are received by Public Health.	Results of the hearing screen may be sent directly to Public Health from the auditory services facilities associated with the birthing facility.
7.3.3.1a	**Alternative Action:** EHDI results are received with the NDBS specimen and testing order.	In at least 17 states, the results of the hearing screen test are sent to Public Health or the Public Health department laboratory along with the testing orders for the NDBS.

7.3.3.1b	**Alternative Action:** EHDI results are noted on the birth certificate.	In a few states the results of the hearing screen are noted on the certificate of birth which is forwarded to Public Health.
7.3.3.1c	**Alternative Action:** EHDI results are received at the time of the NDBS results.	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.
7.3.4	**Event:** Receive NDBS Lab Results	**Figure 7-1, Focus Flows 9 and 9a**
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 sent to Public Health. The report typically indicates positive or negative results, rather than specific values. There may be a need for two views of the data, either summary or more specific to be available to the clinician upon request in the lab report. This concept is covered in more detail in the Newborn Screening Coding and Terminology Guide and is found at the Newborn Screening Use Case website located at http://www.hhs.gov/healthit/usecases/nbs.html.
7.3.4.1a	**Alternative Action:** Public Health department receives results from birthing facility.	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.
7.3.5	**Event:** Incorporate Results into Public Health System	**Figure 7-1**
7.3.5.1	**Action:** Results are incorporated into the Public Health system.	All results of newborn screening tests including EHDI and NDBS may be incorporated into the data systems within Public Health 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 registries or organizations.	The results of the newborn screening tests, both EHDI and NDBS may be sent to various research entities or other Public Health registries. The data may be de-identified at this stage if used for research purposes 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 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 facility.
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 center, the specimen and order may be sent directly from the ordering 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 this document and the Newborn Screening Coding and Terminology Guide. Many of these interactions have been detailed in the 2008 General Lab Orders Use Case which can be found at the following location: http://www.hhs.gov/healthit/usecases.

7.4.2.2	**Action: The lab** reports the 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 discrete lab results available for clinicians. The details of these reports are outlined in Section 10 of this use case and in the Newborn Screening Coding and Terminology Guide.

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

Code	Description	Comments
7.5.1	**Event:** Receive EHDI Order	**Figure 7-1, Focus Flow 5**
7.5.1.1	**Action:** Audiology receives order for EHDI.	Audiology services associated with the birthing facility are informed of the birth and are given the clinician-generated state-mandated order for hearing screening.
7.5.2	**Event:** Perform Testing & Report Results	**Figure 7-1, Focus Flow 6**
7.5.2.1	**Action:** Audiology performs testing in a facility associated with the hospital or birthing center.	Hearing screening may be done at the bedside or in an audiology center associated with the hospital or birthing center.
7.5.2.2	**Action:** EHDI results are reported by Audiology.	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.
7.5.2.2a	**Alternative Action:** Second test is required.	If the results of the EHDI are abnormal 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).

8.0 Scenario 2: Abnormal and Out of Range Results

Figure 8-1 Abnormal and Out of Range Results

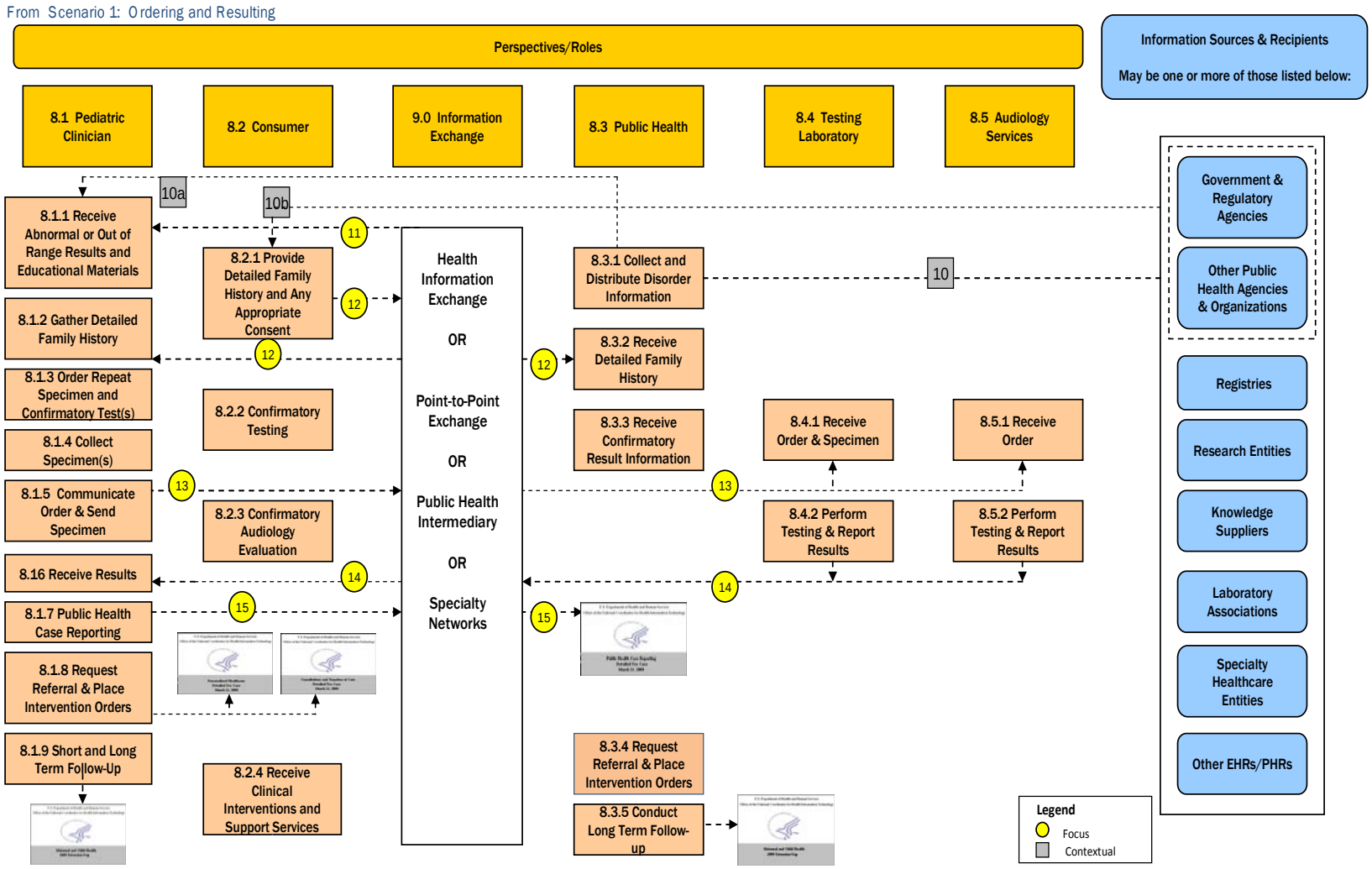

Figure 8-2 Abnormal and Out of Range Results Scenario Flows

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 is collected 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 are communicated 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.

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 or STFU team may be informed of the results from the testing laboratory or from auditory services. The actions under this event (8.1.1) may often occur in parallel.
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, Action (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	**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 EHDI testing, the clinician(s) may need to gather a more detailed family history along with the infant's health status. 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 Public Health. The detailed family history is typically taken after referral to a clinical geneticist or genetic counselor but may be taken prior to or after the confirmatory testing.
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 EHR or the consumer's PHR. As detailed in the 2008 Personalized Healthcare Use Case, a specific tool entitled "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 Test(s)	**Figure 8-1**
8.1.3.1	**Action**: A repeat specimen or repeat audiological evaluation is ordered.	In response to abnormal or out of range results on either the NDBS or EHDI screening tests, specific test(s) may be ordered if required to confirm or extend the initial screening results. However, in some instances, intervention may begin prior to a confirmatory result.
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 (the STFU team) 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 lab 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. The specimen may not always be a blood specimen, for example, a sweat test may be done to confirm Cystic Fibrosis.
8.1.5	**Event:** Communicate Order and Send Specimen	**Figure 8-1, Focus Flow 13**
8.1.5.1	**Action:** The order is 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 audiology evaluation. 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 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 infant 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 is done.	If genetic testing is part of the confirmatory testing, specimens are sent to a genetic testing laboratory. This specimen is usually sent to the genetic lab by a genetic specialist clinician but may be sent by a more general pediatric clinician.
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. The genetic testing details have been described in the 2008 Personalized Healthcare Use Case which can be found at http://www.hhs.gov/healthit/usecases.
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 Public Health. The detailed family history may be a part of the case report. 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 genetic 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. The parents may also be referred to a parent support group focused on the particular genetic disorder identified. If congenital hearing loss is confirmed, various molecular genetic tests may be needed to help identify the etiology of the hearing loss. This may require referral to a genetic specialist. Many of these interactions have been detailed in the 2008 Consultations and Transfers of Care Use Case and/or the 2008 Personalized Healthcare Use Case which both 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 Public Health. 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 Use Case, 2008 Public Health Case Reporting Use Case, and/or the 2008 Personalized Healthcare Use Case, all of 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 guardian(s) receive instructions. This may involve several hours or days of inpatient 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.
8.1.9	**Event:** Short Term and Long Term Follow-Up	**Figure 8-1**

Code	Description	Comments
8.1.9.1	**Action:** Children with various disorders require periodic follow-up visits.	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 provide management options (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. Both this action and the following action 8.1.9.2 may in some states be carried out by the STFU team or the clinical care coordinator.
8.1.9.2	**Action:** Families are referred for education and/or social services.	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.
8.1.9.3	**Action:** Continue to monitor patient for LTFU activities.	From the end of the STFU period, defined by the Association of Maternal and Child Health Programs as “the commencement of treatment or intervention until the child reaches young adulthood,” the pediatric clinician(s) and various agents of Public Health may be involved in aspects of LTFU. During this important period of time, clinicians and others involved in long term management assure that ongoing, high quality, medical management including specialty care and care coordination when needed, are provided. In addition, there should be a smooth transition from pediatric to young adult medical care, periodic assessment of the patient and a contribution to data collection by Public Health. These topics are explored in greater detail in the Maternal and Child Health Extension/Gap document and can be found at the following location: http://hhs.gov/healthit/usecases/mch.html.

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 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 as existing electronic data either from the provider's EHR or the patient's 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:** Confirmatory Testing	**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 results 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 Audiology Evaluation	**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.
8.2.4	**Event:** Receive Clinical Interventions and Support Services	**Figure 8-1**

Code	Description	Comments
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, parents or current guardian(s) 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 8.1.8.4.

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 Flows 10 and 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 documents or brochures, or may in certain cases come from direct communication between Public Health and the birthing facilities.
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**
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 Public Health either electronically or on paper from the pediatric clinician.
8.3.3	**Event:** Receive Confirmatory Result 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 2008 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**

Code	Description	Comments
8.3.4.1	**Action:** Public Health requests a referral.	In certain circumstances, Public Health 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**
8.3.5.1	**Action:** Public Health conducts LTFU of the screening results.	Following the receipt of the Public Health case report, as described in the 2008 Public Health Case Reporting Use Case, Public Health conducts LTFU 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. Many of the actions in this section are explored in greater detail in the Maternal and Child Health Extension/Gap document and can be found at the following location: http://hhs.gov/healthit/usecases/mch.html.
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. Linking of newborn screening to registries (e.g. the birth defects registry) is an important aspect of LTFU.
8.3.5.3	**Action:** Hearing evaluation follow-up.	LTFU of hearing evaluation 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 Rights and Privacy Act (FERPA).

Code	Description	Comments
8.3.5.4	**Action:** Information is used to track key parameters of the newborn screening process.	Once information has been incorporated 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) tracking the number of infants with conditions identified with newborn screening, 5) measuring rates of false positives, and 6) maybe using data 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 centers, medical homes, clinicians, early intervention providers and the family. Normal results should be considered as well as abnormal or out of range results.
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 Order & 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 Public Health, contracted by Public Health, or a private laboratory. The laboratory may be a genetic testing laboratory as detailed and described in Sections 8.1.6.4 and 8.1.6.5.
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 pediatric clinician and in some instances, the ordering clinician or organization. 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 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 services perform 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. The functional capabilities may be provided fully or partially by a variety of organizations including: health information exchange organizations, 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

Code	Capability	Comments
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. Data matching will need to be able to resolve non-standard situations, such as those described in Section 7.1.4.1a of Events & Actions.
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.

Code	Capability	Comments
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, and 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. In certain cases, this data may need to be either anonymized or pseudonymized before being sent to external database systems for confidentiality.

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 purposes 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.

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 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. These examples are not intended to be inclusive of all activities in this area.

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

A. Dates and Events
B. Birth History
C. Newborn Information Required for Screening Order
D. Analytes
E. Conditions
F. Hearing Screenings
G. Ethnicity

Dates and events for HITSP consideration may include:

A. Date of Newborn Screening
B. Date of Diagnosis
C. Diagnosis
D. Date of Referral
E. Type of Referral
F. Date of Enrollment in Treatment Plan/Services
G. 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. A high-level listing which represents the hierarchy of analytes and conditions expressed in the Newborn Screening Coding and Terminology Guide can be found below.

A. Tandem Mass Spectrometry
    i. American College of Medical Genetics (ACMG) Primary Targets
        a. Amino Acids
        b. Fatty Acid Oxidation
        c. Organic Acid
    ii. ACMG Secondary Conditions
        a. Amino Acids
        b. Fatty Acid Oxidation
        c. Organic Acid
    iii. ACMG Other Conditions
        a. Amino Acids
        b. Fatty Acid Oxidation
        c. Organic Acid
B. Non-Tandem Mass Spectrometry Conditions
    i. Endocrine
        a. Thyroid
        b. Adrenal
    ii. Hemoglobin
        a. Hemoglobinopathies
        b. Hemoglobinopathy Traits
    iii. Galactose
    iv. Cystic Fibrosis
    v. Infectious Disease
C. Early Hearing Detection and Intervention (EHDI)
    i. Hearing Loss, Bilateral
    ii. Hearing Loss, Right
    iii. Hearing Loss, Left
    iv. 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 regarding the development and adoption of health information technology.

Aliquot: A measured portion of a sample taken for analysis. One or more aliquots make up a sample.

American College of Medical Genetics (ACMG); The American College of Medical Genetics is an organization composed of biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other health care professionals committed to the practice of medical genetics. The ACMG provides education, resources and a voice for the medical genetics profession.

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, they serve as a reference for healthcare, 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.

Certification Commission for Healthcare Information Technology (CCHIT): 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).

Early Hearing Detection and Intervention (EHDI): Programs set up by U.S. states and territories to help make sure that infants and children with hearing loss are found and receive intervention as soon as possible.

Electronic Health Record (EHR): The electronic health record is a longitudinal electronic record of patient health information generated in one or more encounters in any care delivery setting. This information may include patient demographics, progress notes, problems, medications, vital signs, past medical history, immunizations, laboratory information, and radiology reports.

Electronic Health Record (EHR) / Personal Health Record (PHR) System Suppliers: Organizations which provide specific EHR and/or PHR solutions to clinicians, consumers, 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.

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 functional capability to exchange information between networks related to the health of individuals or populations. 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), integrated care delivery networks, provider organizations, health record banks, Public Health networks, specialty networks, and others supporting these capabilities.

Health Information Exchange 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 health information exchange organizations including integrated delivery networks, health record banks, and others.

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.

Knowledge Suppliers: Entities that use data, vocabulary, technology, and/or industry standards to provide information and tools to entities delivering health care. Examples of newborn screening knowledge suppliers include the American College of Medical Genetics (ACMG), the American Academy of Pediatrics (AAP), and the American College of Obstetrics and Gynecology (ACOG).

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.

Laboratory Information System (LIS) Suppliers: Organizations which provide specific laboratory information system solutions. A laboratory information system is a class of software which handles receiving, processing, transmitting, and storing information generated by medical laboratory processes. These systems often must interface with instruments and other information systems such as hospital information systems. An LIS is a highly configurable application which is customized to facilitate a wide variety of laboratory workflow models.

Long Term Follow-Up (LTFU): Actions commencing after confirmed diagnosis in an affected individual to ensure the screening program can evaluate the effectiveness of their follow up program that may include the process of ensuring availability of ongoing

intervention services and support to affected individuals throughout their lives. LTFU begins after short-term follow-up ends; that is, with the initiation of treatment.

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.

Newborn Dried Blood Spot (NDBS): Dried blood spot specimens are clinical specimens collected for the purpose of genetic testing.

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, and "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): A health record that can be created, reviewed, annotated, and maintained by the patient or the caregiver for a patient. The personal health record may include any aspect(s) of the health condition, medications, medical problems, allergies, vaccination history, visit history, or communications with healthcare providers.

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: 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.

Public Health Knowledge Providers: Associations of Public Health individuals/organizations who provide technical advice and assistance to state and local health agencies in a broad range of areas including: occupational health, infectious diseases, immunization, environmental health, chronic diseases, injury control, and maternal and child health. These associations may include Council of State and Territorial

Epidemiologists (CSTE), Association of Public Health Laboratories (APHL), and Association of State and Territorial Health Officials (ASTHO).

Public Health Systems Suppliers: Organizations which provide specific Public Health 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.

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.

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).

Short Term Follow-Up (STFU): Includes those activities that ensure all infants are screened, abnormal results are appropriately and expediently handled, and affected infants are promptly identified, appropriately referred and treatment initiated where applicable.

Social Service Agencies: Agencies such as social services and law enforcement that help facilitate the newborn screening process and provide information and support for patients and consumers.

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: Medical testing and confirmatory laboratories, either within a hospital, ambulatory, or clinician office environment and/or operating as a free-standing entity, which meet regulatory standards for clinical laboratories and analyze specimens as ordered by providers to assess the health status of patients.