Public Health Informatics Institute

Newborn Dried Bloodspot Screening Business Process Analysis

Report of the NDBS Workgroup:
Screening through transition to Long-term Follow-up

2008

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Executive Summary

The newborn dried bloodspot screening (NDBS) program is mandated by state legislation throughout the United States to identify infants who are at high risk of particular congenital and hereditary conditions, and who would be likely to benefit from early diagnosis and treatment. Most states have individual protocols for the acquisition and analysis of dried bloodspot specimens, dissemination of screening results, and the mechanism of data input and information exchange. This does not allow for the easy flow and exchange of critical information between the public and private sectors within the newborn screening system. Current public health practice and research is thus focused on achieving the seamless integration of clinical and public health information systems. Within this framework, NDBS records represent an essential dataset. Newborn screening programs have developed information systems that support their programmatic efforts to ensure that all children are screened and followed-up appropriately. At the same time, state, federal, and health industry initiatives are guiding the evolution of these systems into interoperable interfaces that bring disparate records within technological reach at the point of care. The ultimate goal of an integrated health information system is to improve the quality of care.

Designing information systems to support program activities requires an analysis of *what* is done and *by whom* throughout the process. To this end, the Public Health Informatics Institute (the Institute) convened a workgroup in February and March 2008 to carry out the business process analysis (BPA) of NDBS from birth through long-term follow-up. This work is supported by the Institute in partnership with the Health Resources and Services Administration, Maternal and Child Health Bureau (HRSA/MCHB) and the Genetics and Newborn Screening Region 3 Collaborative (Region 3).

Executive Summary (continued)

The NDBS Workgroup sought to clarify the roles and activities conducted by public health programs, primary care specialists, sub-specialists, social services, and others. The NDBS Workgroup believes that the activities described herein will lead to the better integration of information systems, which will in turn support comprehensive, continuous, culturally appropriate care that includes public health involvement coordinated through a medical home. Adjunct tasks of the NDBS Workgroup were to build on the emerging concept of the long-term follow-up (LTFU) team for newborn screening by describing the care coordination functions as well as to describe the process of intervention management (long-term follow-up).

The NDBS Workgroup applied the BPA methodology to describe the core activities within the NDBS system. Defining these core activities is the initial step in defining requirements for intrastate and interstate information systems. These information exchange systems can support the informational needs of all the stakeholders involved in the overall NDBS system. Such an analysis also contributes to the development of future information systems that will conform to National Health Information Network (NHIN) interoperability standards.

Introduction

Newborn Dried Bloodspot Screening (NDBS) is the process of collecting a heel stick blood sample onto a filter paper collection device within two-to-three days of birth, analyzing the specimen using approved laboratory methods, and reporting results to healthcare providers and families. The NDBS program is mandated by state legislation throughout the United States to identify infants who are at high risk of particular congenital and hereditary conditions, and who would be likely to benefit from early diagnosis and treatment.

Most states have individual protocols for the acquisition and analysis of dried bloodspot specimens, dissemination of screening results, and the mechanism of data input and information exchange. This does not allow for the easy flow and exchange of critical information between the public and private sectors within the newborn screening system. Current public health practice and research is thus focused on achieving the seamless integration of clinical and public health information systems. Within this framework, NDBS records represent an essential dataset.

Newborn screening programs have developed information systems that support their programmatic efforts to ensure that all children are screened and followed-up appropriately. At the same time, state, federal, and health industry initiatives are guiding the evolution of these systems into interoperable interfaces that bring disparate records within technological reach at the point of care. The ultimate goal of an integrated health information system is to improve the quality of care.

Objectives of the NDBS Workgroup

Designing information systems to support program activities requires an analysis of *what* is done and *by whom* throughout the process. To this end, the Public Health Informatics Institute (the Institute) convened a workgroup in February and March 2008 to carry out a business process analysis (BPA) of the NDBS process from birth through long-term follow-up. This work is supported by the Institute in partnership with the Health Resources and Services Administration, Maternal and Child Health Bureau (HRSA/MCHB) and the Genetics and Newborn Screening Region 3 Collaborative (Region 3). These entities recognized the value of establishing a viable baseline of the NDBS system as well as in developing its key business processes with regard to long-term follow-up.

The Institute defines BPA as, "the effort to understand an organization and its purpose while identifying the activities, participants, and information flows that enable the organization to do its work. The output of the business process analysis phase is a model of the business processes consisting of a set of diagrams and textual descriptions to be used for design or redesign of business processes."(1) Applied within the context of the NDBS program, BPA addresses the practical realities of screening by describing the responsibilities of all involved entities as well as the necessary work flow essential to the smooth and comprehensive exchange of information that can lead to a better standard of care. The NDBS Workgroup recognized that equally important to the quality of care is a sound business process that clearly outlines the steps and individuals involved with ensuring that a child is screened, appropriately diagnosed, and managed in the health care network throughout his or her lifetime.

Newborn dried bloodspot screening (NDBS) programs in state health departments have typically followed children from screening through diagnosis and initiation of care, but have not extended into management and long-term monitoring. Recently, the Department of Health and Human Services Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) issued a statement that defines long-term follow-up and identifies the health department's role in that follow-up. Using this definition as a starting point, the NDBS Workgroup sought to clarify the roles and activities conducted by public health programs, primary care specialists, sub-specialists, social services, and others. The NDBS Workgroup believes that the activities described herein will lead to the better integration of information, which will in turn support comprehensive, continuous, culturally appropriate care that includes public health involvement

coordinated through a medical home. Adjunct tasks were to build on the emerging concept of the long-term follow-up (LTFU) team for newborn screening by describing the care coordination functions as well as to describe the components of intervention management (long-term follow-up). Intervention management, as defined by ACHDNC, includes care coordination through a medical home, evidence-based treatment, continuous quality improvement, and new knowledge discovery.(2)

The work of the NDBS Workgroup complements that of the Subcommittee on Follow-up and Treatment, which is part of ACHDNC. The Subcommittee has defined the general character of long-term follow-up, to which the NDBS Workgroup has independently added the specific work activities conducted by each member of the clinical and public health teams involved with assuring comprehensive follow-up care.

The NDBS Workgroup includes leaders in public health practice and education, clinicians, laboratorians, hospital caregivers, and NDBS program staff (Appendix A). Some of these individuals are a subset of members and stakeholders of the *Connections* Community of Practice (CoP), a facilitated network of public health practitioners and stakeholders engaged in the integration of child health information systems at the state and local level, under the aegis of the Institute. The Institute facilitated a series of meetings in which the NDBS Workgroup and partners used BPA to:

- Define the collaborative activities involved in newborn dried bloodspot screening
- Describe the members of the LTFU team
- Define the full range of activities of the Clinical and Public Health care coordinators (as part of the LTFU team, the care coordinators ensure that children identified through screening receive needed services and continuous, comprehensive care throughout their lifetime)
- Discuss the information system support to follow-up services.

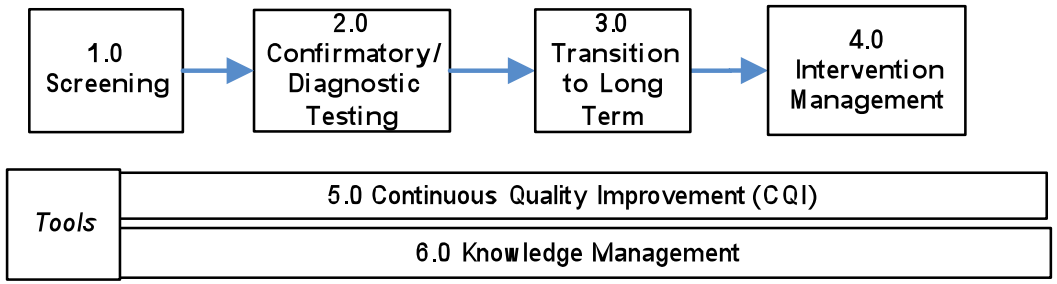
The NDBS Workgroup applied the BPA methodology to describe the core activities within the NDBS system. Defining these core activities is the initial step in defining requirements for intrastate and interstate information systems that can support the informational needs of all the stakeholders involved in the overall NDBS system. Such an analysis also contributes to the development of future information systems that will conform to National Health Information Network (NHIN) interoperability standards.

NDBS Workgroup Report

The present narrative describes the collaborative results and conclusions of the NDBS Workgroup offered in business process terms. These discussions among expert stakeholders were essential in developing a consistent view of the many activities that constitute a system of long-term follow-up. The NDBS Workgroup agreed that the many activities they carry out to support children with abnormal newborn dried bloodspot screening results can be portrayed or modeled in a standardized manner, which translates into a shared screening and information exchange system. However, not all states or organizations will follow the steps exactly as they are presented. This document attempts to show the most common and essential screening and follow-up processes (including short-term follow-up and the transition to long-term follow-up) used by a majority of states and territories, and offers a description of the potential processes involved with long-term follow-up as a best case scenario that involves collaborative health management coordinated at both the clinical and public health levels.

Figure 1 represents a conceptual diagram of the steps involved in the NDBS program. It indicates the general categories of the steps as well as the direction of the work flow involved in the program. Also shown are the tools involved in long-term follow-up that include continuous quality improvement and knowledge management. The numbers preceding each event correspond to the accompanying context diagrams, which are explained on page fourteen.

Figure 1: Conceptual Diagram, Scope of NDBS Program



This report describes the sequence of screening events and the functions of the clinical and public health teams involved with NDBS screening and follow-up care as defined by the NDBS Workgroup. Using the tools of BPA, the NDBS Workgroup created the conceptual framework of NDBS screening by generating the business process matrices, context diagrams, and task flow diagrams that detail the necessary and proposed screening events. Figure 2 depicts the Institute's business process matrix (BPM), which includes the individual business process elements that the NDBS Workgroup considered when creating the context and task flow diagrams. The BPM outlines the components that characterize the business process – the goals, objectives, triggers, inputs/outputs, business rules, and (measurable) outcomes (horizontal axis), and a representative set of business processes (vertical axis). Using the matrix to define the components, the NDBS Workgroup was able to determine whether a given activity had the appropriate structure to be classified as a business process.

Figure 2: Business Process Matrix, Public Health Informatics Institute

Business Process Name	Goal	Objective	Business Rules	Trigger(s)	Task Set	Inputs	Outputs	Measurable Outcomes
Definitions	The major goal that the process supports. The goal is the end state to be achieved by the work of the agency, and should be defined in terms of the benefits to the community.	A concrete statement describing what the business process seeks to achieve. A well-worded objective will be SMART: Specific, Measurable, Attainable/Achievable, Realistic, and Time bound.	A set of criteria that defines or constrains some aspect of the business process. Business rules are intended to assert business structure or to control or influence the behavior. For example, laws, standards, guidelines, etc.	Event, action, or state that initiates the first course of action in a business process. A trigger may also be an input, but not necessarily so.	The set of activities that are carried out in a business process.	Information received by the business process from external sources. Inputs are not generated within the process.	Information transferred out from a process. The information may have been the resulting transformation of an input, or it may have been information created within the business process.	The resulting transaction of a business process that indicates the objectives have been met.

Context diagrams are used to illustrate the participants as well as the flow of information within the work environment. Context diagrams consist of two graphical elements: circles and arrows. The circles represent entities (a person or group of persons who perform one or more tasks involved in the process depicted). Arrows represent transactions that involve the exchange of information among entities. Task flow diagrams capture the basic temporal flow of tasks as well as the individual or groups involved in each task. Graphical elements in a task flow diagram depict inputs, processes, and results for each step that make up a task. These graphical elements include the following:

- Oval – beginning or end of task series
- Square – task
- Double-edged square – predefined task series
- Diamond -- decision
- Home plate – transitional point
- Arrows – transaction

These elements are displayed across horizontal areas of the task flow diagram, which are referred to as swim lanes. The swim lanes represent the individual or groups (entities) involved in each task. The graphical elements (inputs, processes, and results) may remain in one swim lane, indicating that the task is confined to that person or group, or may cross two or more swim lanes, in which case the task that these elements depict may be carried out by any of the entities displayed in those swim lanes.

This report represents the written complement to the details provided in the task flow and context diagrams, and is provided to readers to assist in assessing the validity of the NDBS Workgroup's output. The Institute will also disseminate the findings of the NDBS Workgroup to the broader audience of the public health and medical communities through peer-reviewed publications. This information should serve to inform and define information system requirements to support programs.

The NDBS Workgroup has described the steps and entities involved with the following business process activities:

- First Screening (including routine screening and requested repeat screening)
- Confirmatory/Diagnostic testing
- Transition to long-term follow-up
- Intervention Management

Table 1 provides the sequence of screening events involved with first screening through confirmatory/diagnostic testing. Please note that the numbers preceding each event correspond to the accompanying task flow diagrams.

Table 1: Sequence of screening/testing events

1.0.1 Acquire blood spot	The physician of record and/or birth site staff informs the family about the newborn dried bloodspot screening program and the screening process. Parental consent is solicited with a consent form, if required by state law. In most states screening is done by default, unless the family declines via a dissent form, in which case the refusal is filed either by the department of public health or the NDBS lab and no further testing steps are taken. The birth site (i.e. hospital, birthing center, etc.) then identifies the newborn and completes the pre-numbered identification form, which also records demographic information. The first bloodspot sample is drawn, placed on the blood spot paper, and sent to the screening laboratory. Some institutions also transfer a patient's demographic information electronically. The birth site documents that the specimen was sent.
1.0.2 Routine second screen	Some states or facilities routinely conduct repeat screenings as part of their established protocol (e.g., if the patient is in the NICU).
1.0.3 Requested Repeat Screen	If abnormal (out-of-range) results are produced by a previous screen, or the initial sample is unsatisfactory, the initial specimen may be reprocessed or another sample taken.
1.1.1 Testing specimen	Specimens are sent directly to the screening lab by the birth site.

1.2.1 Normal (in-range) results	For normal (in-range) results, the birth site checks off the results on the specimen log, files results in the patient record or electronic health record, and forwards the results to any or all of the following: the physician of record, the PCP, or NICU. The results are recorded by each; in addition the department of health cross checks the results with vital records. The family is notified by the birth site or PCP.
1.2.2 Abnormal (out-of-range) results	The PCP or physician of record confers with a specialist when necessary (e.g., confirmatory results for a particular condition) and with the STFU team, which shares information among all groups. The specialist confirms the diagnosis and enlists the members involved with long-term follow-up. In some states, it is the specialist who initiates confirmatory testing after being informed by the PCP or screening lab of the initial results.
2.0 Confirmatory/Diagnostic Testing	Diagnostic testing is conducted to confirm abnormal (out-of-range) screening results.

NDBS Screening, Testing, Care Coordination, and Intervention Management (1.0 – 6.1.1)

The following sections detail the screening events as well as the entities and individuals involved in the screening, testing, and follow-up processes. Each is introduced by the business process matrix that corresponds to that event. The matrices provide the conceptual framework from which the NDBS Workgroup derived the screening events, as evidenced in the task flows and context diagrams.

1.0 First Screening

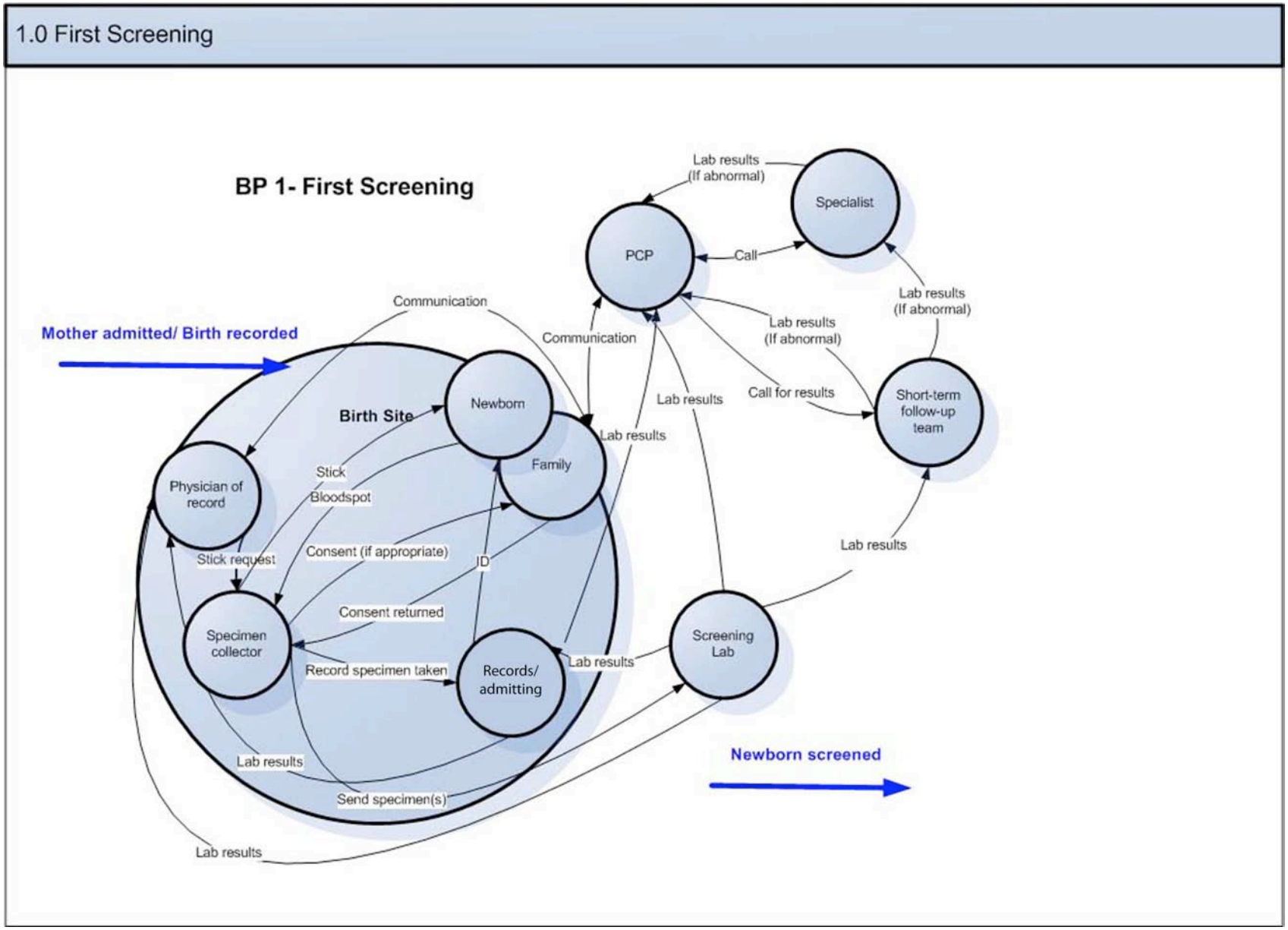
Newborn dried bloodspot screening involves parent and provider education, dried bloodspot specimen collection, and initial screening. The process is triggered by one of two scenarios: a birth is recorded at the birth site or the mother is admitted in labor.

Figure 3: Business Process Matrix, 1.0 First Screening

Business Process Name	Goal	Objective	Business Rules	Triggers	Task Set	Input	Output	Measurable Outcomes
First Screening	Prevent complications from detectable congenital and hereditary disorders.	Identifying infants at risk of disorders.	• State laws, rules, and regulations. • State advisory recommendations. • Professional recommendations and guidelines. • Hospital/state protocol (repeat testing). • Abnormal results reported to: ○ Birth site ○ PCP ○ Specialist ○ Family.	• Mother admitted in labor. • Infant is born in the U.S. at a hospital or birthing center (home births may be different).	• Consent (if needed). • Blood spot taken. • Card sent to lab. • Lab processing. • Lab reports results to: ○ Birth site ○ Physician of record ○ PCP ○ Specialist (if out-of-range).	Birth report. Blank collection kits (filter paper) sent from state lab to birth site.	Report of first screening test.	• Proportion of newborns screened. • Proportion of unsatisfactory samples. • Proportion of all test results reported per state requirements. • Proportion of out-of-range results reported to STFU team. • Percent of screenings completed in a stated time based on best practices. • Proportion lost to follow up. • Interval between births reported results and intervention.

Screening begins within the first 48 hours after the infant's birth. The screening infrastructure usually includes: the birth site (including the admissions department, the physician of record, the specimen collector, and the medical records department), the primary care physician (PCP), the short-term follow-up (STFU) team, and the screening laboratory. The screening process generally occurs as described in the following sections.

Figure 4: Context Diagram, 1.0 First Screening



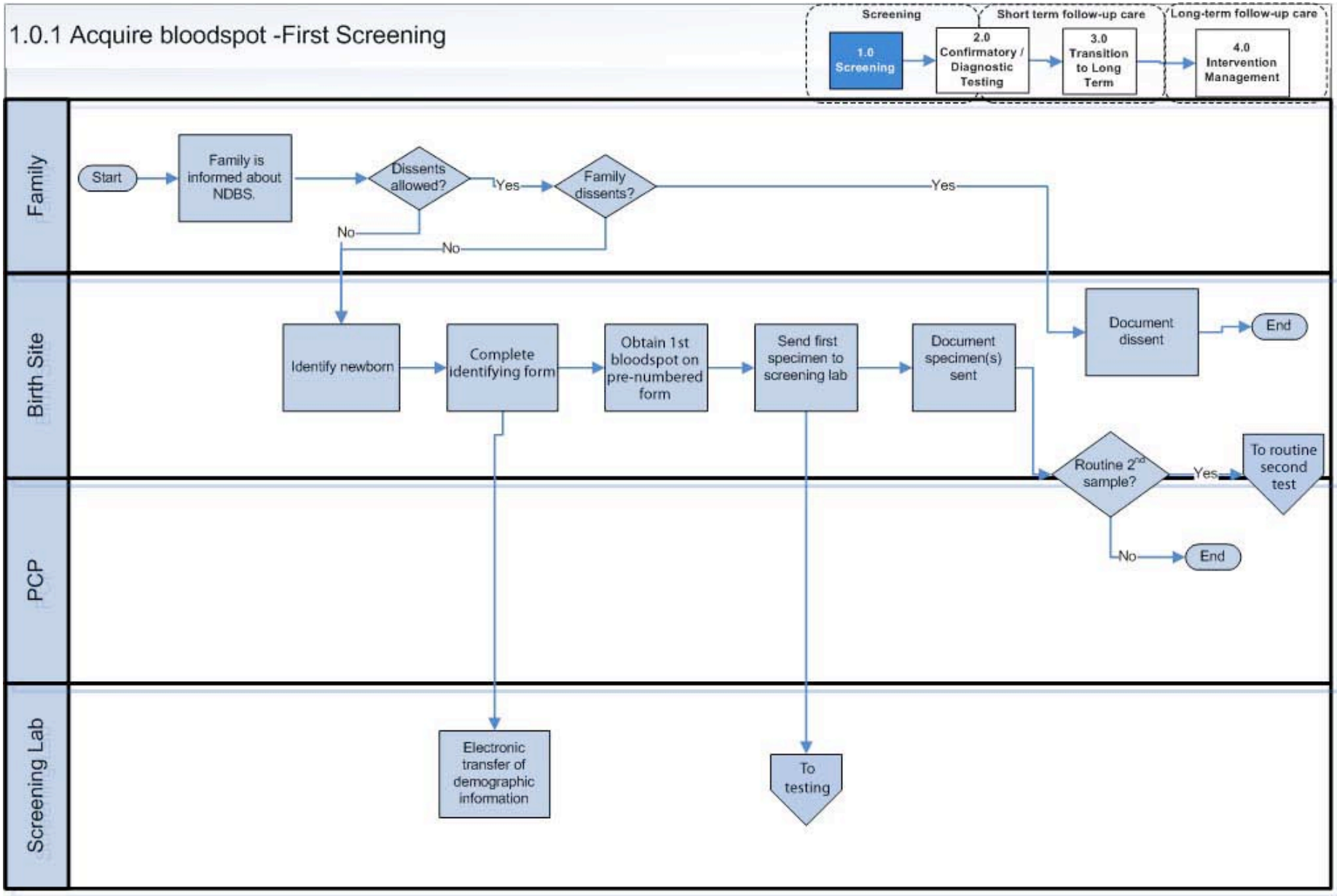
1.0.1 Acquire the blood spot

The physician of record/birth site staff informs the family about the newborn dried bloodspot screening program and the screening process through literature and/or discussion. At this point, a few states require parental consent, according to state law. These states proceed with screening only after the family signs a consent form. In most other states, screening is done by default, unless the family declines for personal or religious reasons via a dissent form, in which case the refusal is filed either by the department of public health or the NDBS lab, and no further testing steps are taken.

The birth site (i.e. hospital, birthing center, etc.) identifies the newborn and completes the pre-numbered identification form, which also records demographic information. The first bloodspot sample is drawn, placed on the blood spot paper, and sent to the screening laboratory. Some facilities also transfer a patient's demographic information electronically. The birth site documents that the specimen was sent. When screening of the initial specimen produces an urgent abnormal (panic) value, the STFU team will enlist a clinician/specialist to begin treatment/intervention prior to receiving the confirmatory results.

Note: It is standard practice for labs to reprocess initial specimens in duplicate as per Clinical and Laboratory Standards Institute (CSLI) protocol and quality assurance procedures.

Figure 5: Task Flow Diagram, 1.0.1 Acquire the blood spot



1.0.2 Routine Second Screening

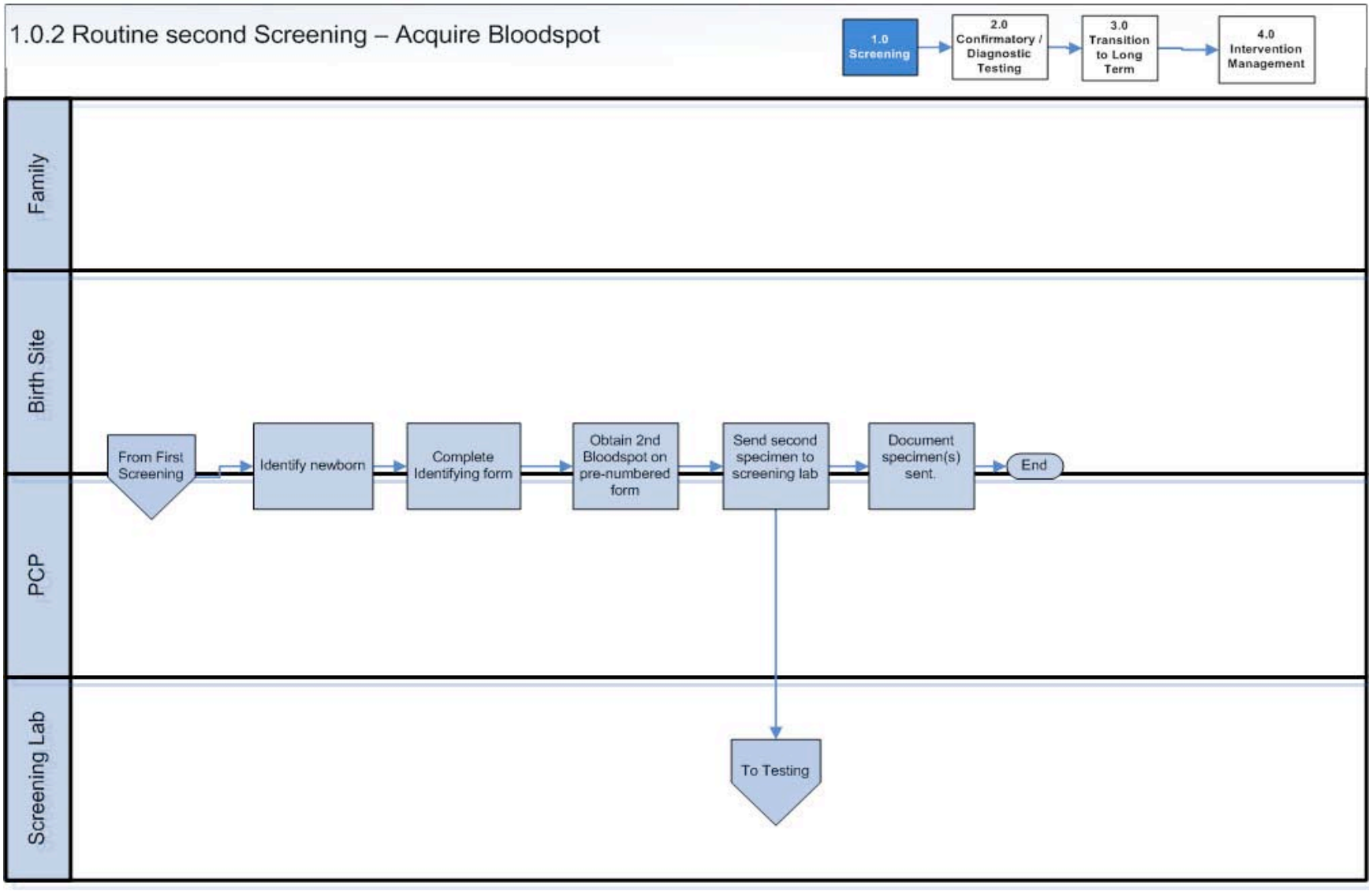
Some states or facilities routinely conduct repeat screenings as part of their established protocol (e.g., if the infant is in the NICU) to catch missed first screenings, to screen infants not detected during the first screening time frame, or to screen for disorders that have abnormalities detected at later time frames.

Figure 6: Business Process Matrix, 1.0.2 Routine Second Screening

Business Process Name	Goal	Objective	Business Rules	Triggers	Task Set	Inputs	Outputs	Measurable Outcomes
Routine second screening	Prevent complications from detectable congenital and hereditary disorders. Catch missed first screens. Pick up infants not detected on first screen. Pick up abnormalities detected at later time frames.	Identify infants at risk of disorders.	• State laws (mandated in every state). • Professional recommendations and guidelines. • Birth site/state protocol (repeat testing). • Abnormal results reported to: o Birth site o PCP o Specialist o Family.	Infant is born in the U.S. at a hospital or birthing center (home births may be different).	• Blood spot taken. • Card sent to lab. • Lab processes specimen. • Lab reports results to: o Birth site o Physician of record o PCP o Specialist (if out-of-range).	Birth report. Blank collection kits (filter paper) sent from state lab to birth site.	Report of routine second screening test.	• Proportion of newborns screened. • Proportion of unsatisfactory samples. • Proportion of all test results reported per state requirements. • Proportion of out-of-range results reported to STFU team. • Percent of screenings completed in a stated time based on best practices. • Discordance between first and second screen.

Once an additional sample is taken, the steps are repeated as described in section, 1.0. First Screening. However, no additional permission from the family is solicited because permission was obtained and documented with the first sample.

Figure 7: Task Flow Diagram, 1.0.2 Routine Second Screening



Note: The routine second screen should not be confused with the requested repeat screen, which occurs when the initial sample is unsatisfactory or produces out-of-range results as explained in, 1.0.3, Requested Repeat Screen.

1.0.3 Requested Repeat Screen

If abnormal (out-of-range) results are produced by a previous screen or the initial sample is unsatisfactory, the initial specimen may be reprocessed or another sample taken. Note: Low birth weight or NICU babies may be tested more than once per the facility or state protocol.

Figure 8: Business Process Matrix, 1.0.3 Requested Repeat Screen

Business Process Name	Goal	Objective	Business Rules	Triggers	Task Set	Inputs	Outputs	Measurable Outcomes
Requested Repeat screen	Prevent complications from detectable congenital and hereditary disorders.	Identify infants at risk of disorders when first screening is out-of-range.	• State laws (mandated in every state). • Professional recommendations and guidelines. • Birth site/state protocol (repeat testing). • Abnormal results reported to: ○ Birth site ○ PCP ○ Specialist ○ Family.	Infant is born in the U.S. at a hospital or birthing center (home births may be different).	• Blood spot taken. • Card sent to lab. • Lab processes specimen. • Lab reports results to: ○ Birth site ○ Physician of record ○ PCP Specialist (if out-of-range).	Birth report. Blank collection kits (filter paper) sent from state lab to birth site.	Report of repeat screening test.	• Proportion of newborns screened. • Proportion of unsatisfactory samples. • Proportion of all test results reported per state requirements. • Proportion of out-of-range results reported to STFU team. • Percent of screenings completed in a stated time based on best practices. • Reasons for additional request. • Interval between request and screen. • Interval between request and reported.

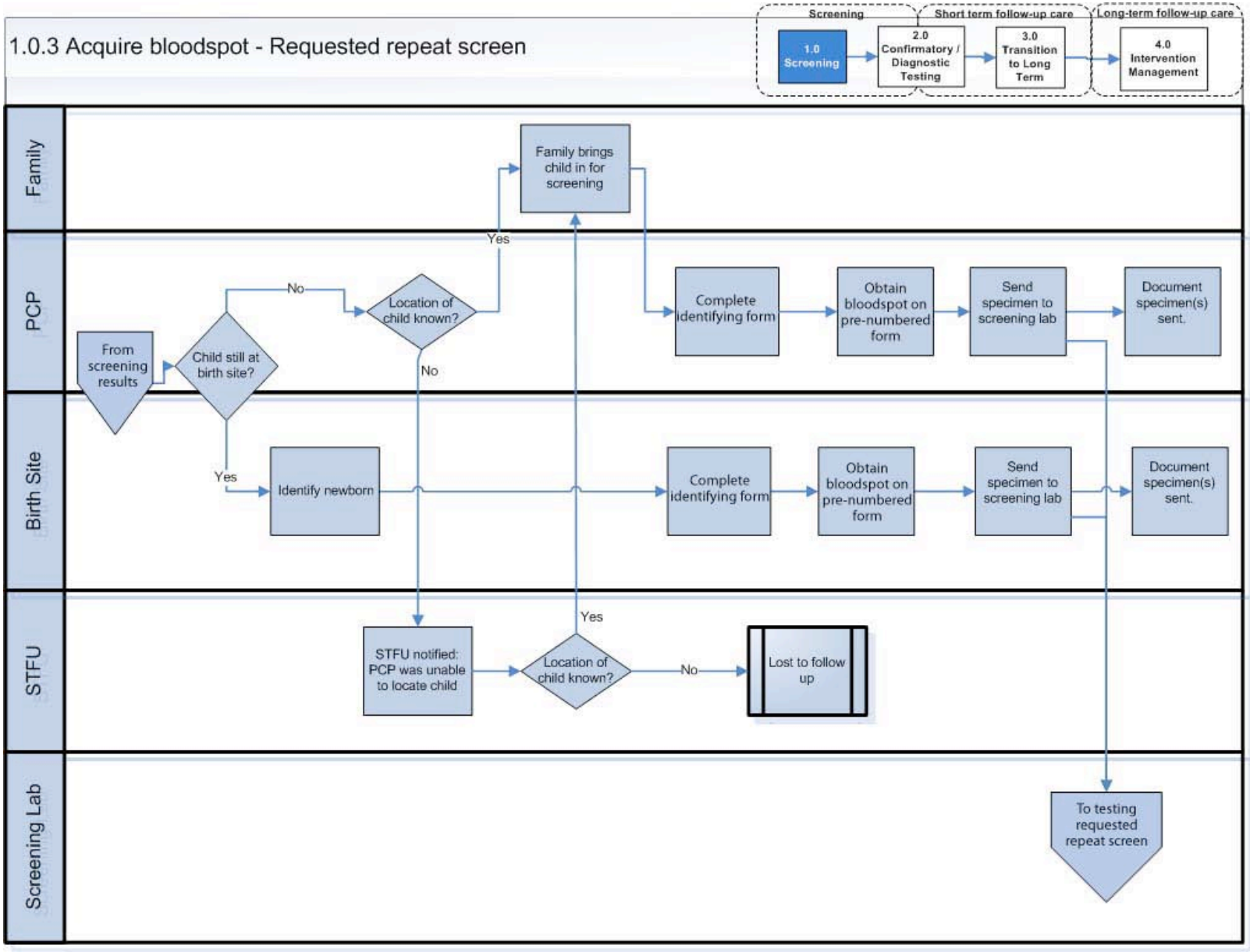
The STFU team notifies the PCP that an additional sample is required. However, individual state protocol identifies any of the following parties as responsible for contacting the family and discussing care options: the birth site, PCP, or NICU. Oversight of the repeat screening process is conducted by either the birth site or the PCP.

The requested repeat screening process occurs as follows:

- the family arranges to provide another sample (either with the birth site or the PCP)
- the identification form is completed
- the bloodspot is obtained on a pre-numbered form
- the specimen is sent to the screening lab
- the results are returned to the physician of record at the birth site, STFU team, or PCP
- the results are disseminated and recorded
- the specimen is stored for up to two weeks, or, in some states, indefinitely. (Note: the last step requires a separate business process that is outside the scope of this document.)

Each of these screening results prompts the involvement of the STFU team to coordinate retesting, diagnostic testing, and/or referral of the newborn to a specialist.

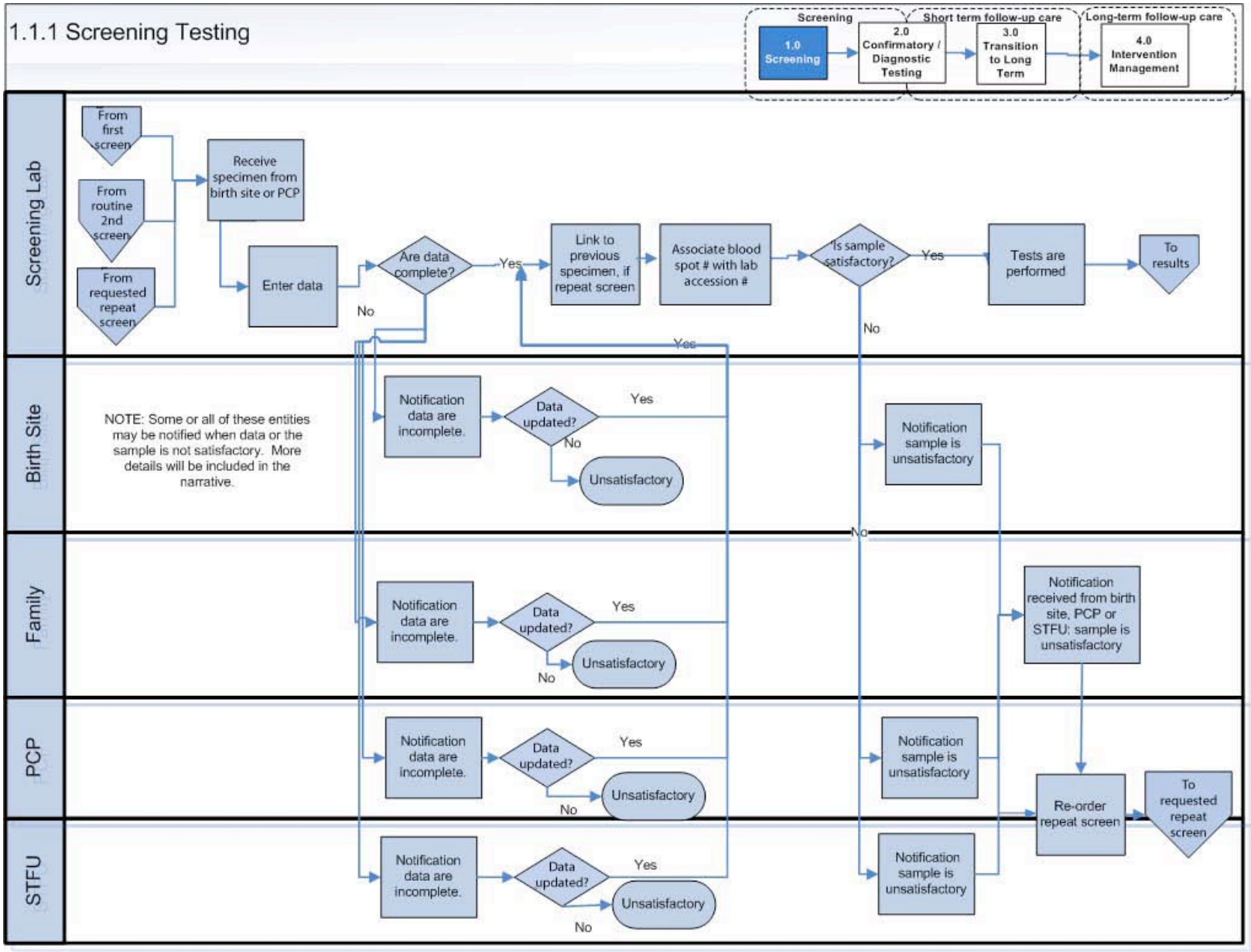
Figure 9: Task Flow Diagram, 1.0.3 Requested Repeat Screen



1.1.1 Screening Testing

Specimens obtained from the first screen, routine second screen, and/or requested repeat screen are sent directly to the screening lab by the birth site or PCP, which may notify the lab in advance of any incoming screens (either electronically or by phone). The practice of notifying the lab of incoming samples is not standard. In general, birth sites have no verification process in place to confirm that the specimen was delivered to the lab. In a few states, the birth site compares the laboratory information with birth records to ensure that all newborns are captured for the screening. This process is documented by the birth site, and serves as a tracking mechanism to assure that universal screening takes place and to verify that a test was completed by the screening lab. Notification/verification practices differ among birth sites based on state laws and individual facility protocols.

Figure 10: Task Flow Diagram, 1.1.1 Screening Testing



Once the specimen is received, the screening laboratory completes the following steps prior to processing the specimen:

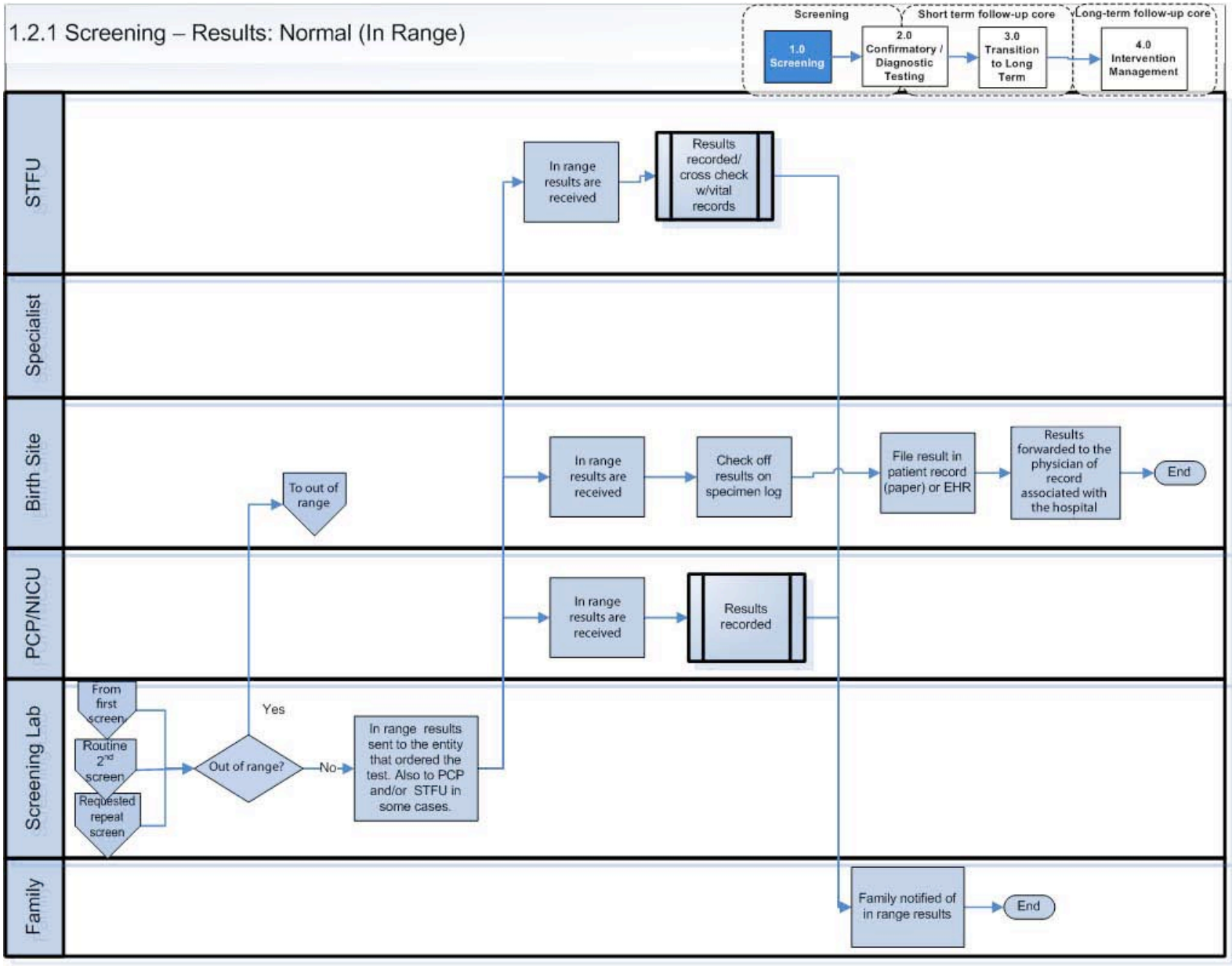
- verification that the sample is satisfactory
- verification that the demographic data received from the sampling entity (i.e. birth site, STFU team, PCP) are complete
- entry of the data
- link to the first specimen when not the initial screen
- comparison of the blood spot number with the lab number
- verification that the sample is satisfactory

In some states, the blood sample may be discarded or only tested for critical values (e.g., to check for galactosemia) when it is inadequate or otherwise unsatisfactory. The screening lab will notify some or all of the following entities of an unsatisfactory and/or inadequate sample, depending on state protocol: birth site, PCP, or STFU team. The family is notified by the birth site or PCP that an additional sample is required. The PCP re-orders a requested repeat screen when required.

1.2.1 Screening – Normal (in-range) results

For normal (in-range) results, the birth site checks off the results on the specimen log, files results in the patient record or electronic health record, and forwards the results to any or all of the following: the physician of record, the PCP/NICU, and STFU team. The results are recorded by each; in addition, the department of health cross checks the results with vital records. The family is notified by the birth site or the PCP.

Figure 11: Task Flow Diagram, 1.2.1 Screening – Results: Normal (in-range)

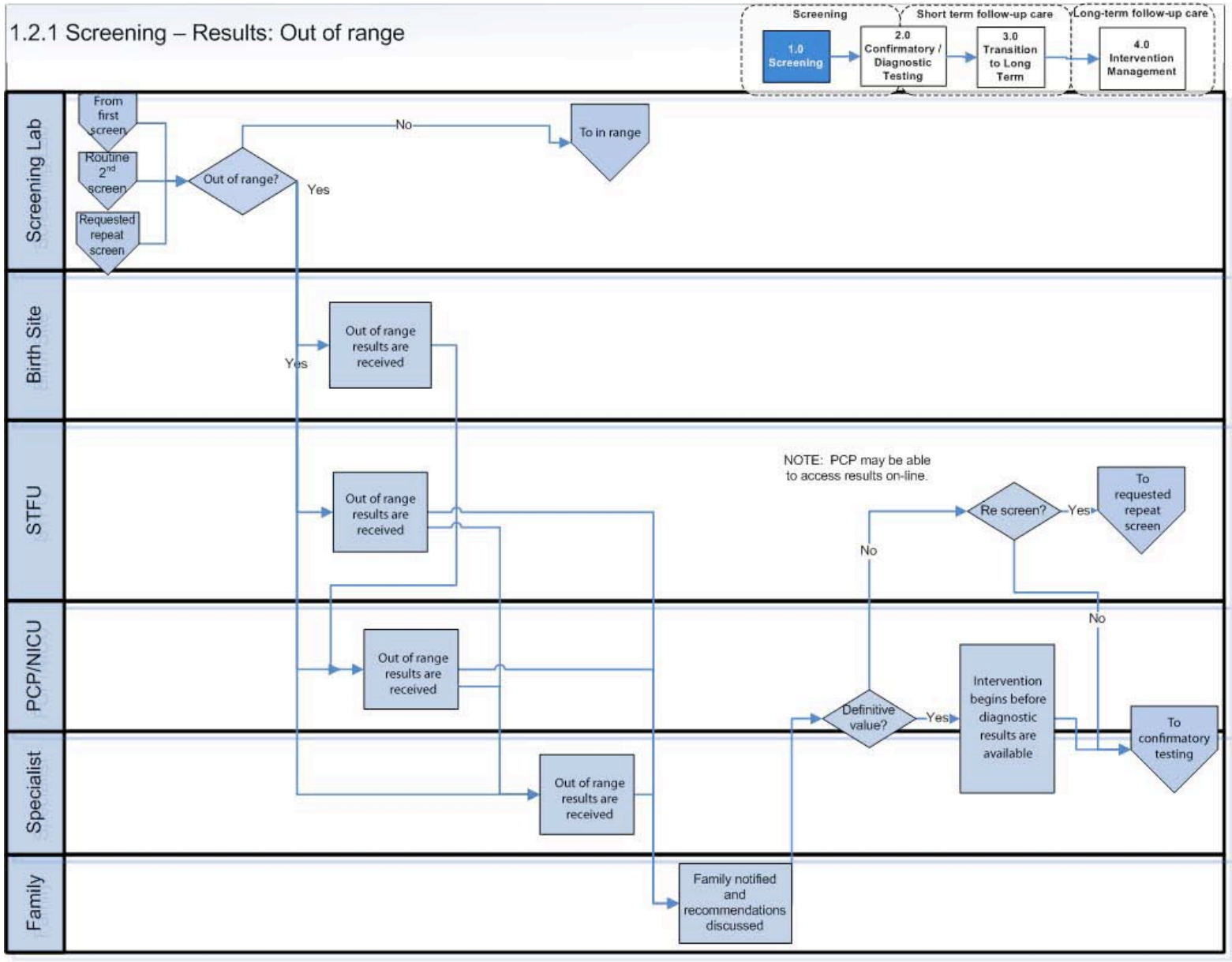


- **1.2.2 Screening – Abnormal (out-of-range) results**

An abnormal (out-of-range) screen can be designated as:

- Urgent out-of-range (panic) value – draw blood and do confirmatory testing
- Definitive value
- Borderline, requiring re-screen

Figure 12: Task Flow Diagram, 1.2.2 Screening – Results: Abnormal (out-of-range)



The STFU team informs the PCP and specialist that the screening result is abnormal (out-of-range). Depending on state protocol, the birth site, PCP, or specialist does the following:

- notifies the family that the infant needs to be seen and further testing is necessary
- if the family agrees to testing, will request the diagnostic test
- procures the additional specimen from the newborn/child
- sends the specimen to the diagnostic lab

The PCP or physician of record confers with a specialist when necessary and with the STFU team, which is responsible for sharing information among all the groups. The PCP or specialist, in turn, confirms the diagnosis (in some states, it is the specialist who initiates confirmatory testing after being informed by the PCP or screening lab of the initial results), and at this point enlists the members involved with long-term follow-up. Note: the PCP may be able to access results on-line. More information about confirmatory testing is provided in the section, 2.0 Confirmatory/Diagnostic Testing.

Short-term follow-up

The clinical team involved with coordinating the repeat screening and confirmatory/diagnostic testing processes includes the PCP, specialist, and the STFU team, which is typically considered to be part of (or contracted by) the state health department. Members of the STFU team include: coordinator, relevant state public health department staff, and specialist (e.g., geneticist). The STFU team identifies the newborn and notifies the PCP of abnormal (out-of-range) results. In the event that the birth site is unable to locate the family, the child is recorded as lost to follow-up. The STFU team is also responsible for informing the PCP and/or specialist about the need for follow-up, and communicates with these individuals as necessary. Due to its role, the STFU team is considered to be the coordinating entity. Note: In some states, it is the birth site, PCP, or specialist that contacts the family as well as the diagnostic lab to request that a repeat screening or diagnostic test take place.

2.0 Confirmatory/Diagnostic Testing

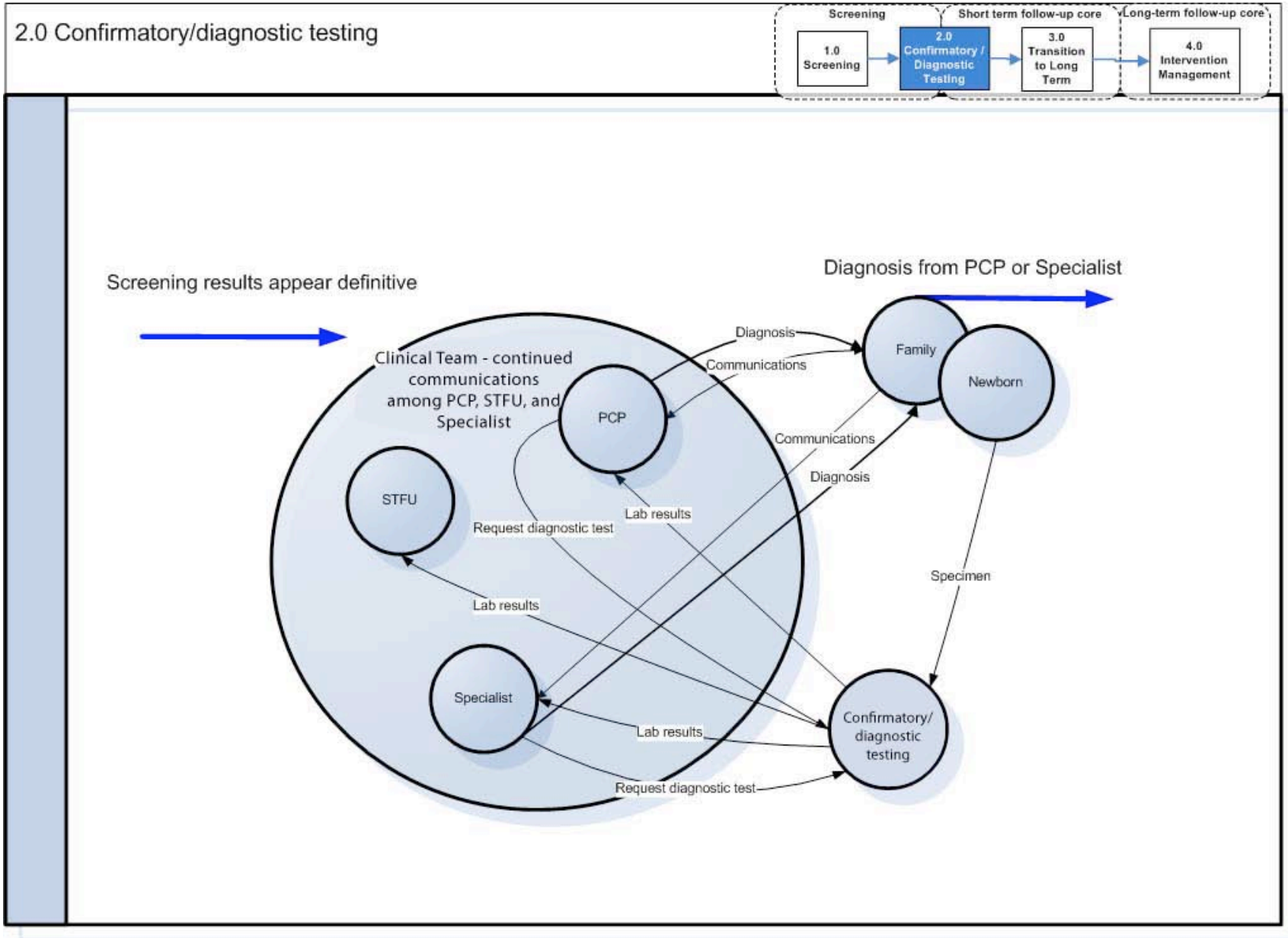
Based on the screening results and according to program protocol, diagnostic testing is conducted to determine the condition involved.

Note: hearing screening follows a somewhat different process.

Figure 13: Business Process Matrix, 2.0 Confirmatory/Diagnostic Testing

Business Process Name	Goal	Objective	Business Rules	Triggers	Task Set	Inputs	Outputs	Measurable Outcomes
Confirmatory /Diagnostic Testing	Prevent complications from detectable congenital and hereditary disorders.	Timely confirmation of diagnosis in infants with out-of- range screens.	• Diagnostic protocols. • Professional recommendations and guidelines. • Referral to specialists if diagnosed. • Certified lab. • State laws, rules, and regulations. • PCP, specialist, and STFU team must communicate.	• Out-of-range screening result (appears definitive). • Family history. • Clinical manifestations before results. • Prenatal diagnosis.	• Diagnostic testing. • Report results to PCP and specialist and/or STFU team. • Family notified. • QA: information exchange between screening and diagnostic lab when results differ. • Arrange payment – get prior authorization from insurance company or other appropriate source. • Refer children with special health care needs to long-term follow-up.	Test request from STFU team, specialist or PCP.	• Report of diagnostic test. • Family brought in. • Referral to ancillary services and long-term follow-up.	• Percent of diagnoses completed in a stated time based on best practices (condition-specific). • Proportion of those referred by confirmed diagnosis. • Percent lost to follow-up. • Percent identified with secondary or additional disorders.

Figure 14: Context Diagram, 2.0 Confirmatory/Diagnostic Testing

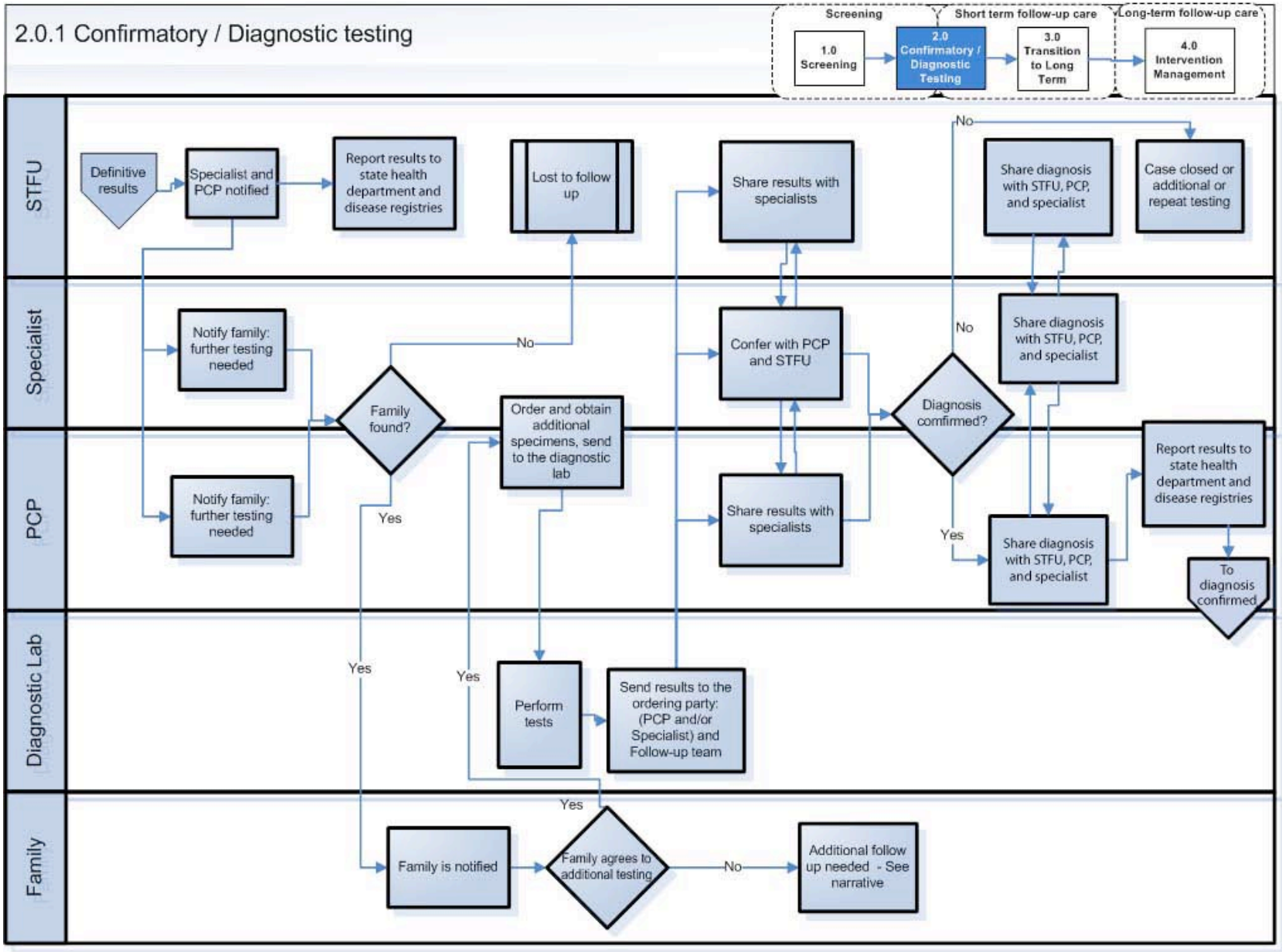


The parties involved in the clinical team include the PCP, specialist, and STFU team. The STFU team is considered the coordinating entity as the team is responsible for maintaining communication among all members involved in the clinical team.

2.0.1 Confirmatory/Diagnostic Testing

Diagnostic testing is conducted to confirm abnormal (out-of-range) screening results. Once the test is complete, the lab shares results with the ordering party (i.e., STFU team, PCP, or specialist). If a diagnosis is confirmed, the ordering party shares the results with the other members of the team. At this point, the case is transitioned to intervention management (long-term follow-up). In some cases, additional repeat testing is requested. During the confirmatory testing period, the STFU team continues to coordinate the exchange of information with the PCP and specialist. The STFU team is also responsible for providing a report of the results to the appropriate state health department registry of diseases and referring the affected infant to the available services. Note: If the family is not found after concerted effort, the infant is considered lost to follow-up. If the family refuses testing, the STFU team may follow-up in an attempt to convince the family to allow the testing.

Figure 15: Task Flow Diagram, 2.0.1 Confirmatory/Diagnostic Testing



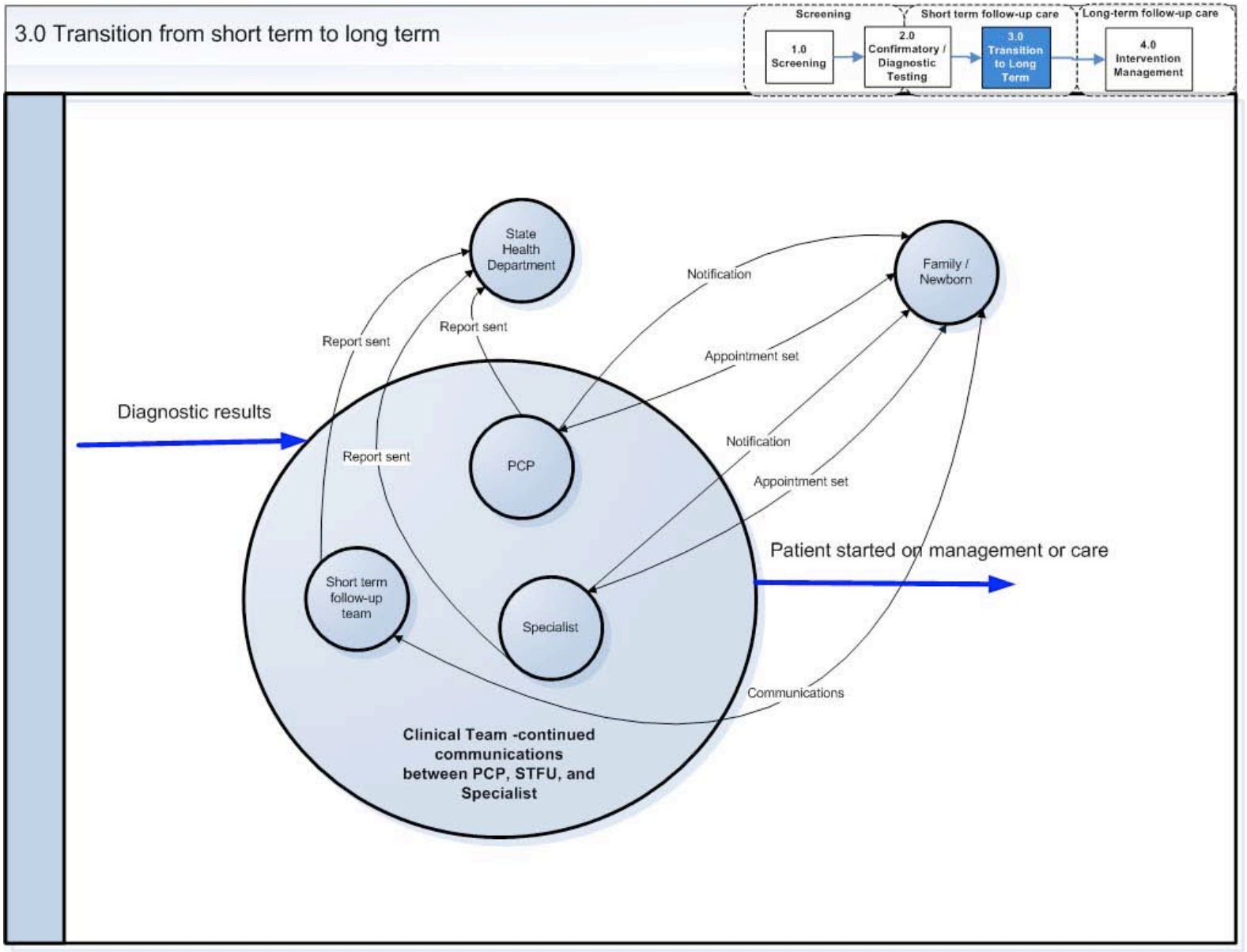
3.0 Transition to Long-Term Follow-Up

The important aspects of transition from short-term to long-term follow-up are that the child begins immediate care and the arrangements for long-term intervention and management are initiated.

Figure 16: Business Process Matrix, 3.0. Transition from Short-term to Long-term Follow-up

Business Process Name	Goal	Objective	Business Rules	Triggers	Task Set	Inputs	Outputs	Measurable Outcomes
Transition from short-term to long-term follow-up	Prevent complications from detectable congenital and hereditary disorders.	Timely and accurate transfer of patient and information to the proper care provider. Care management.	• Privacy policy (HIPAA). • Reportable conditions (e.g., birth defects). • State laws, rules, and regulations. • Identify medical home. • Professional recommendations.	• Confirmed diagnosis. • Referral to long-term follow-up.	• Confirmed diagnosis shared with specialist, PCP, STFU team, specialist, and family. • PCP or specialist accepts patient. • Appointment set with patient. • Develop communications plan. • Develop intervention/management plan. • Identify source of payment. • Establish baseline health status/record. • Identify medical home. • Assess patient/family satisfaction.	• Diagnostic test results. • Interpretation by PCP, specialist, or lab. • Information on payment mechanism.	• Communicate with family. • Report to NDBS program.	• Timely and appropriate treatment of diagnosed child. • Every child having a treatment/management plan and medical home. • Proportion of children with abnormal diagnosis results who have appropriate management. • Proportion of children with confirmed diagnoses who receive follow-up care from PCP or specialist. • Proportion of children who have followed their care plan. • Proportion lost to follow-up.

Figure 17: Context Diagram, 3.0 Transition from Short-term to Long-term Follow-Up

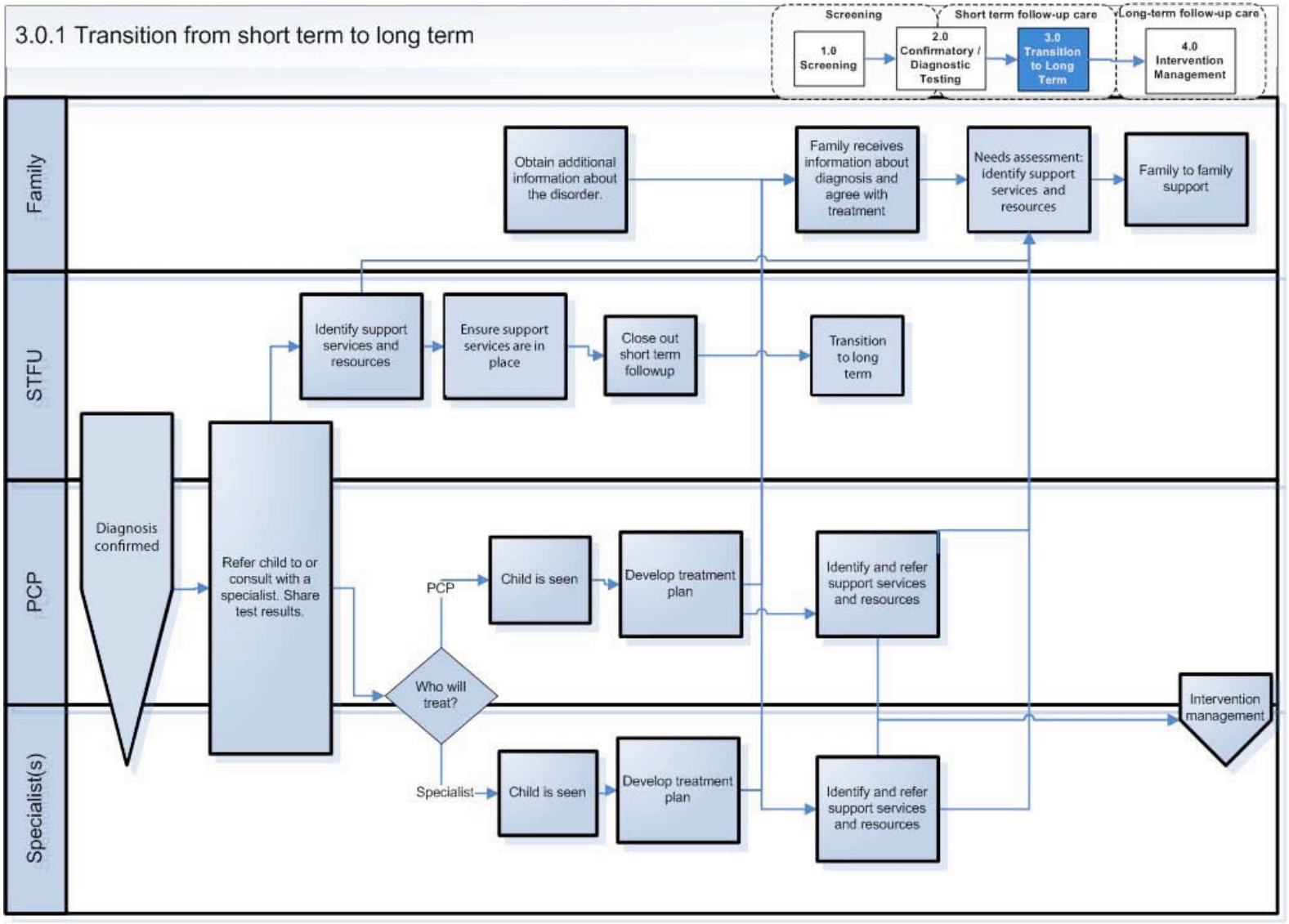


Once results confirm a child's disorder, the STFU team, the PCP, and the specialist are all key entities in facilitating the transition of the infant and family into long-term intervention and management. In most states, the STFU team takes the lead in arranging care as well as with sharing information among all members of the child's care team.

3.0.1 Transition from Short-term to Long-term Follow-up

When tests have confirmed a diagnosis, the PCP or specialist provides the results to the child's family. If the child's family agrees to treatment, either the specialist or the PCP (depending on state protocol) will develop and begin a treatment plan. In the meantime, the STFU team (and in some states the PCP or specialist) identifies necessary support services and resources (including family-to-family support) and ensures that these support services are in place. At this time, the STFU team will close out short-term follow-up. In all states, the state health department is notified of abnormal confirmatory results (by the STFU team, PCP, or specialist, depending on state policy), and provided with a report.

Figure 18: Task Flow Diagram, 3.0.1 Transition from Short-term to Long-term Follow-Up



4.0. Intervention management – Care Coordination

The LTFU team is an essential component of the intervention and ongoing treatment of the child throughout his or her lifespan. The components of intervention management as described by the NDBS Workgroup (in accordance with ACHDNC definitions) include the following:

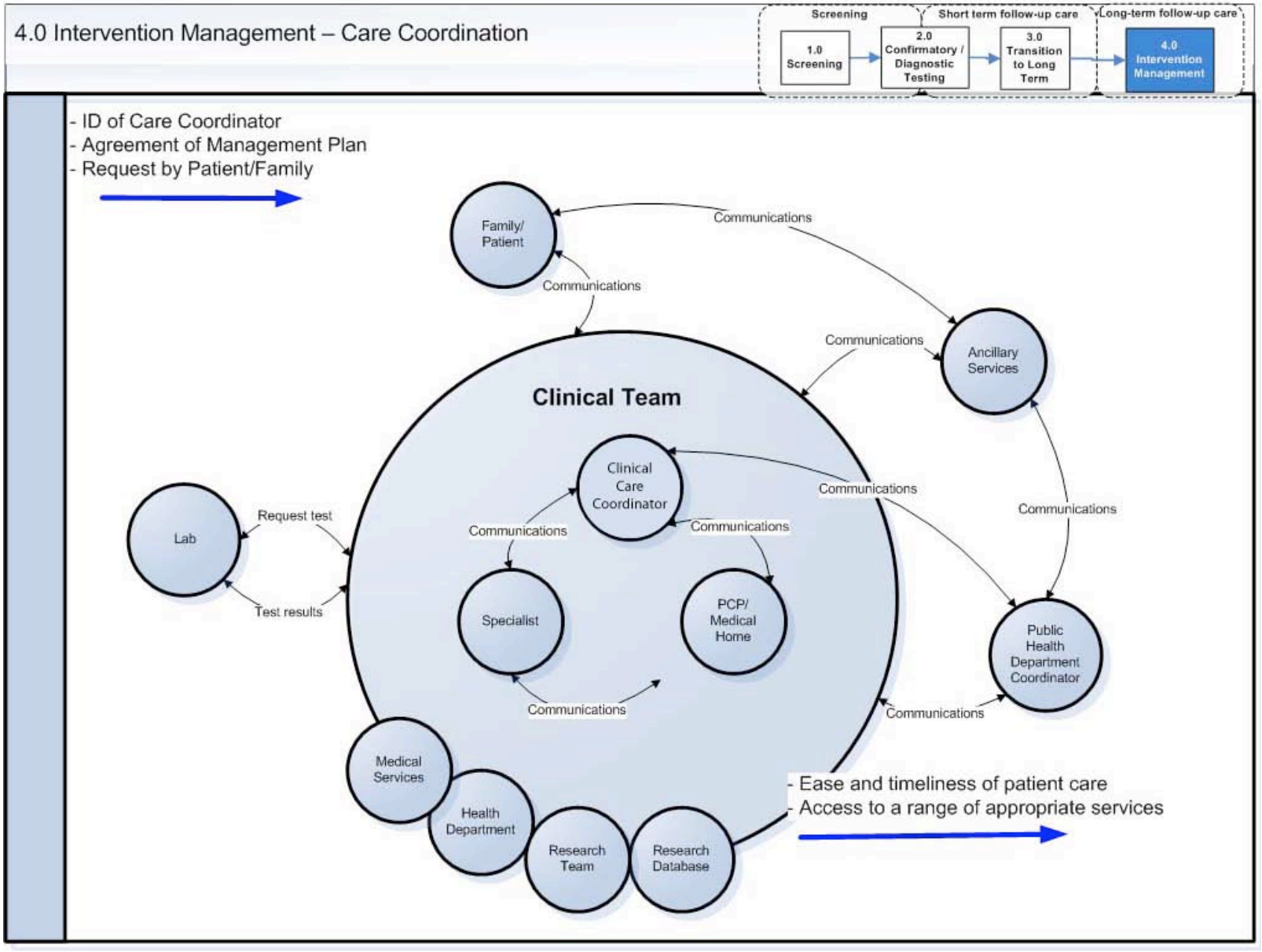
- Care coordination/Ongoing Treatment
- Continuous Quality Improvement
- Knowledge Generation
- Knowledge Management and Dissemination

The members of the clinical team involved with intervention management include: the PCP, the specialist, and the care coordinators. The Clinical Care Coordinator (CCC) is a member of the medical home. (3) The Public Health Care Coordinator (PHCC) is a member of the state health department.

Both are in place to provide continuity of care to the affected child and his/her family, and to ensure the consistent exchange of information among the medical home, public health entities, and ancillary services. The ultimate goal of the care coordinators is to ensure the best possible outcome for the child.

The medical home is responsible for reviewing and updating the management plan, performing and monitoring clinical tests, recommending hospitalization when necessary, requesting lab tests and ancillary services, and reviewing test results. The lab is involved to periodically conduct additional tests upon request from the medical home. Ancillary services are provided on an ongoing basis through adulthood.

Figure 19: Context Diagram, 4.0 Intervention Management – Care Coordination



Clinical Care Coordinator

The Clinical Care Coordinator (CCC) at the medical home facilitates the linkage of the child and his/her family with appropriate services and resources in a coordinated effort. The CCC is responsible for ensuring that the affected child receives the range of appropriate services from the point of diagnosis through adulthood. The activities of the CCC are to:

- Conduct a comprehensive assessment with identification of needs, including psychosocial
- Develop a care plan jointly with the child's family
- Implement the care plan with the following provisions:
  - Provide care and form clinical team
  - Connect child with appropriate tests (e.g. thyroid values)
  - Provide timely and appropriate referral
  - Coordinate medical and ancillary services
  - Assist family with identification and referral to community services, including family support groups
  - Maintain central record of pertinent medical information and communications
  - Facilitate communication (family, clinical team)
  - Help the patient/family transition to adult care without losing medical home
- Disseminate/educate appropriate management/treatment
- Conduct regular and periodic evaluations
  - Monitor care coordination and service needs
  - Monitor child's health status

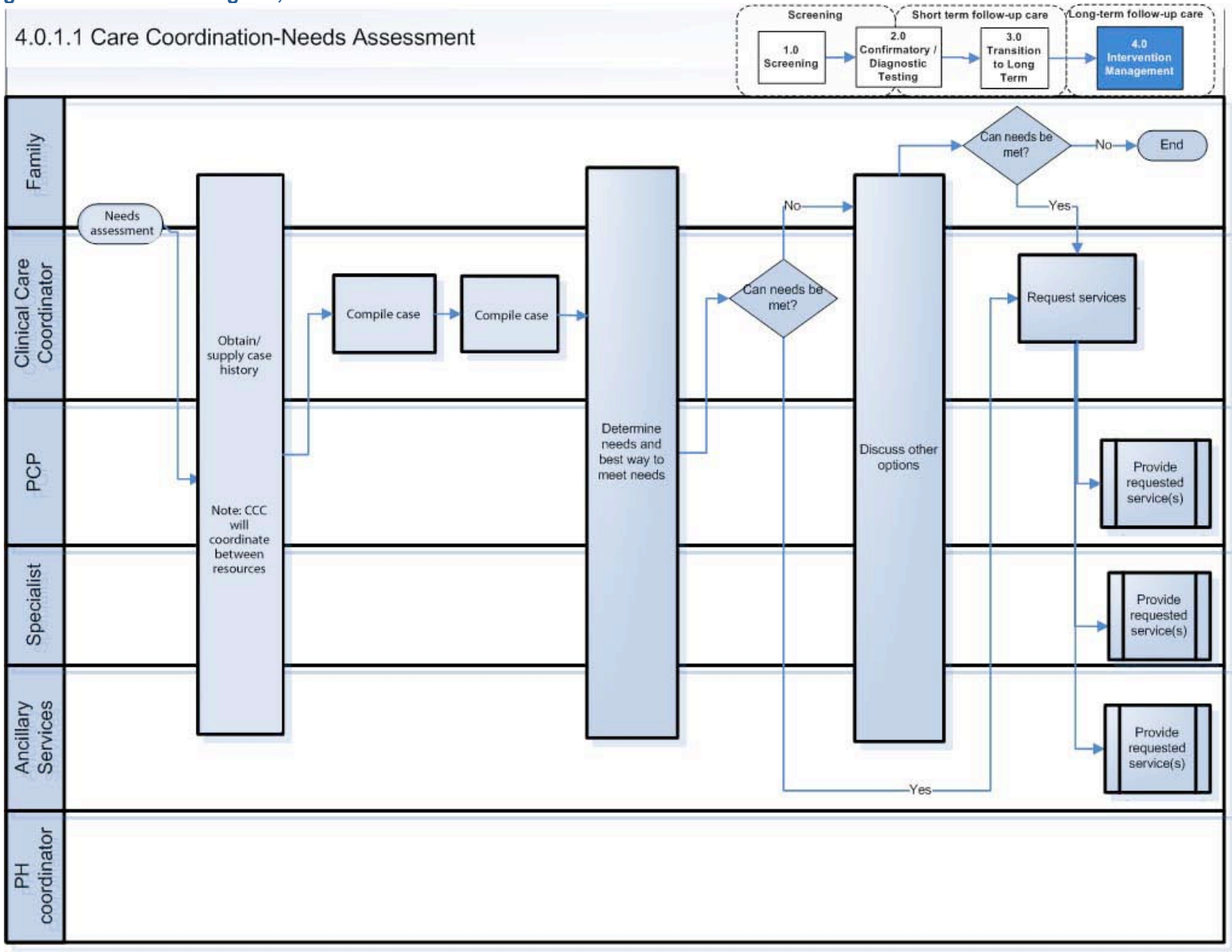
4.0.1.1 Needs assessment

As the services are provided, the CCC will evaluate the family to ensure that all needs are met, update the management plan, and provide reports to providers and relevant entities. The needs assessment process proceeds as follows:

- Family is approached about needs evaluation
- Family agrees to needs assessment and services
- Family provides medical history and expresses medical and social needs
- CCC obtains case history and reviews case
- CCC determines needs and best way to meet needs
- CCC, working with the PCP, provides clinical services within the medical home
- CCC requests social services from appropriate ancillary group
- If needs cannot be met will discuss other options with family
- PCP, specialist, and ancillary groups provide needed services

The CCC also provides updates to the PCP, specialist, and ancillary services (this, in addition to traditional social services, may include support groups, such as those for sickle cell disease).

Figure 20: Task Flow Diagram, 4.0.1.1 Needs Assessment



Another essential role of the CCC is to manage the exchange of information among the other members of the team as well as with services outside the medical home. The entities involved in information sharing include:

- Family/patient
- Medical services (e.g. pharmacy)
- Research team, when relevant
- Research database and clinical database, when relevant
- Public health department and/or PHCC
- Ancillary services

The NDBS Workgroup feels it is essential that the CCC have the education and professional background necessary to communicate easily with both internal and external members of the clinical care team.

Public Health Care Coordinator

As proposed by the NDBS Workgroup, the Public Health Care Coordinator (PHCC) provides oversight of long-term follow-up by monitoring the child and family's needs and assisting in addressing service gaps. This includes requesting needed services from members of the medical home as well as ancillary services. In describing the functions of the PHCC, the NDBS Workgroup sought to acknowledge the need for an active public health presence in fulfilling the coordination and provision of care. Overall, the PHCC assesses the completeness of care and provides assurance of the delivery of care. As the monitor of long-term follow-up, the various activities of the PHCC as described by the NDBS Workgroup are as follows:

- Ensure that periodic testing takes place (blood measurements, bio-markers, lab assessments)
- Assess whether the child is adhering to needed services
- Monitor and document the provision and coordination of services
- Coordinate information about the child and share relevant information with those entities that need the information, including the medical home, ancillary, and community services
- Assess health outcomes (including school developmental/psychological assessments)

- Facilitate medical homes' timely access to test results and relevant public health information
- Report outcomes to clinicians, research entities, health plans, and community groups
- Participate in program and policy developments

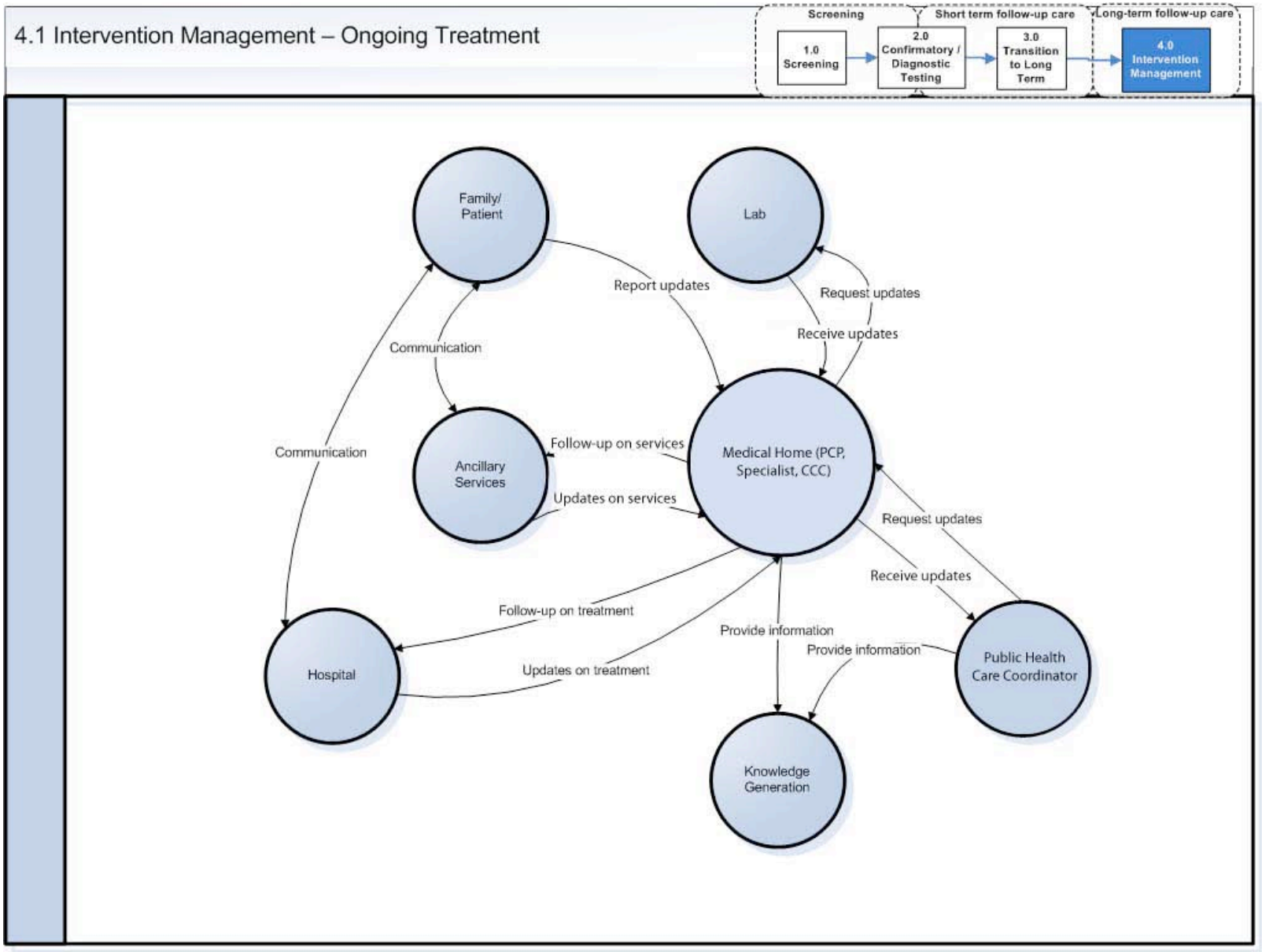
The PHCC opens a case once notified of a child's abnormal result. The CCC and PHCC work closely together to monitor the care that the child receives. The CCC is actively involved in the patient's management on an ongoing, day-to-day basis, while the PHCC has a broad range of responsibilities.

The PHCC regularly interfaces with the CCC to gather information about the clinical services provided to the child as a means of program evaluation. The PHCC may also periodically interface with the family and assess the quality of the care that the child receives. This evaluation could take the form of a patient survey, the results of which are reported to the public health department. The PHCC uses information from the CCC, medical home, and family to update the child's management plan from the public health perspective, i.e. lab and ancillary services. In some states (such as Florida) local health departments have designated individuals who conduct activities similar to those of a PHCC as described in this report.

4.1 Intervention Management

Ongoing treatment of an infant extends beyond the members of the medical home (i.e., PCP, CCC, and specialist) to include the PHCC, ancillary services, birth center, screening laboratory, and entities involved with knowledge generation.

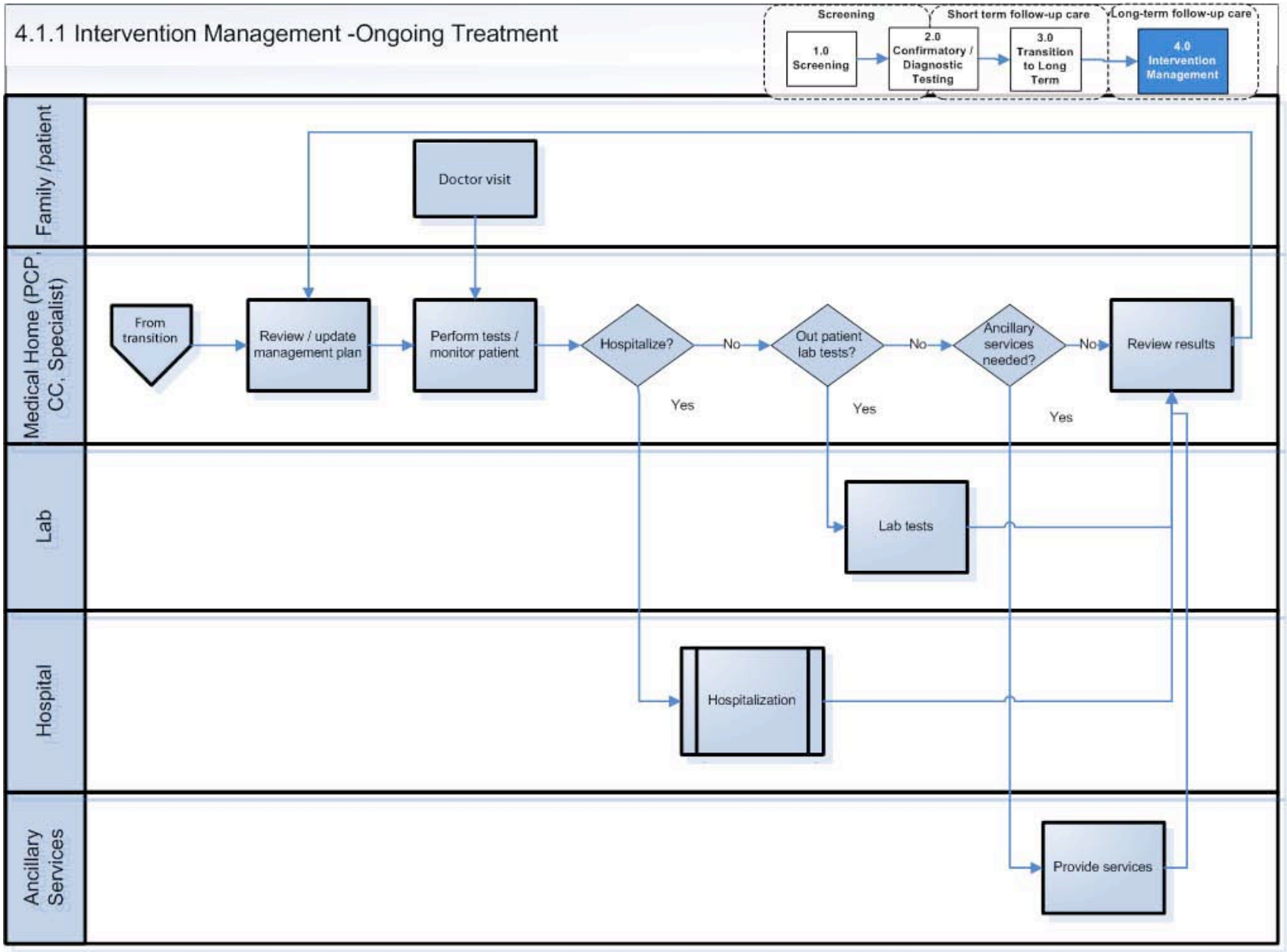
Figure 21: Context Diagram, 4.1 Intervention Management



4.1.1 Intervention Management – Ongoing Treatment

The medical home reviews the ongoing test results and clinical exams to update the management plan accordingly. This entity is also responsible for performing the tests that monitor the infant's ongoing state of health and recommending appropriate care, which can include hospitalization, outpatient lab tests, and ancillary services as needed.

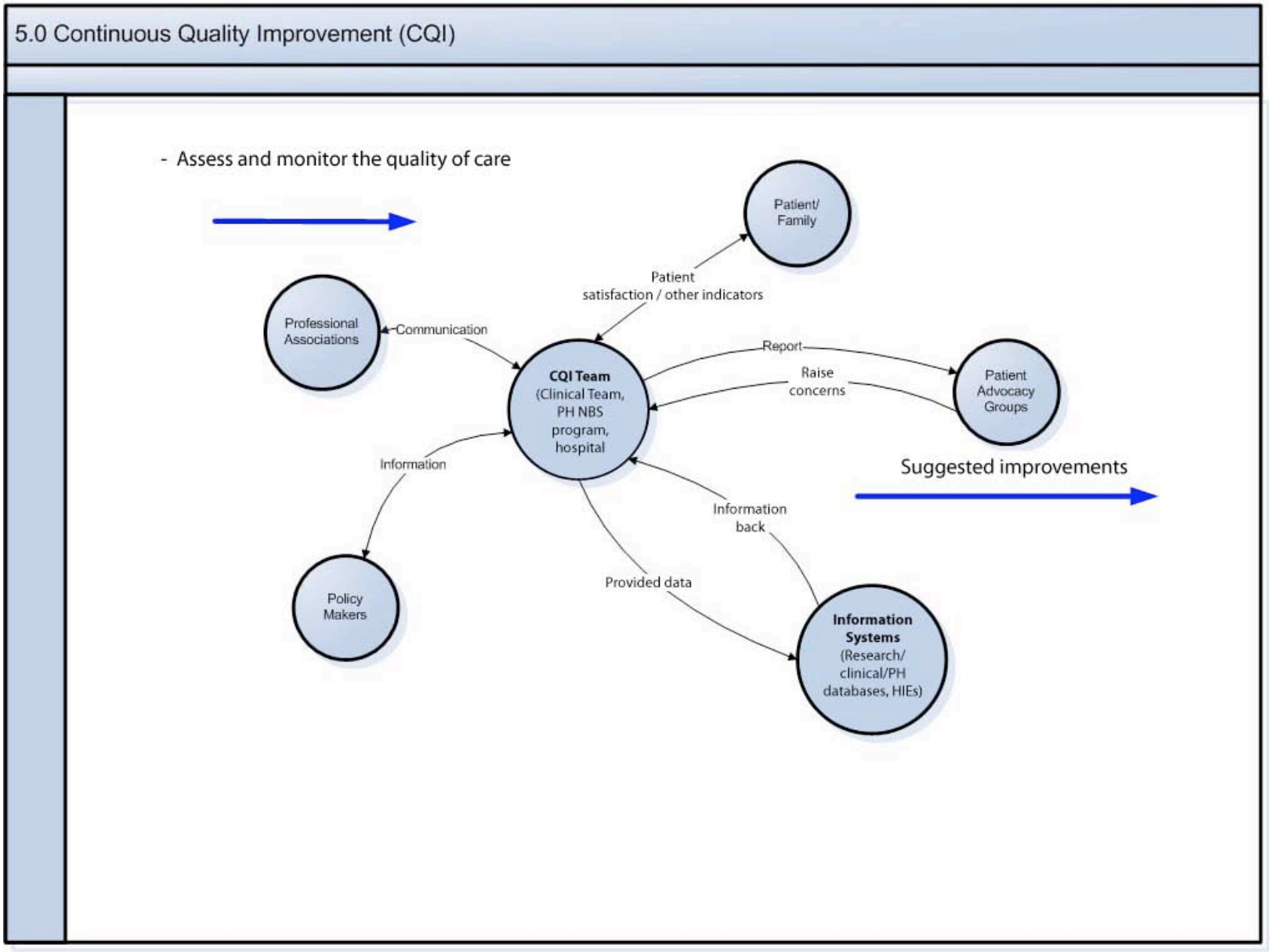
Figure 22: Task Flow Diagram, 4.1.1 Intervention Management – Ongoing Treatment



5.0 Continuous Quality Improvement (CQI)

The clinical team, birth site, and public health newborn screening program form the core of the CQI team involved with intervention management. In an effort to assess and monitor the quality of care, the CQI team assesses patient satisfaction, informs policy makers, communicates with professional associations, and provides data to information systems (contributing data to clinical, research, and public health information systems as well as health information exchanges). In addition, the CQI team provides reports to patient advocacy groups.

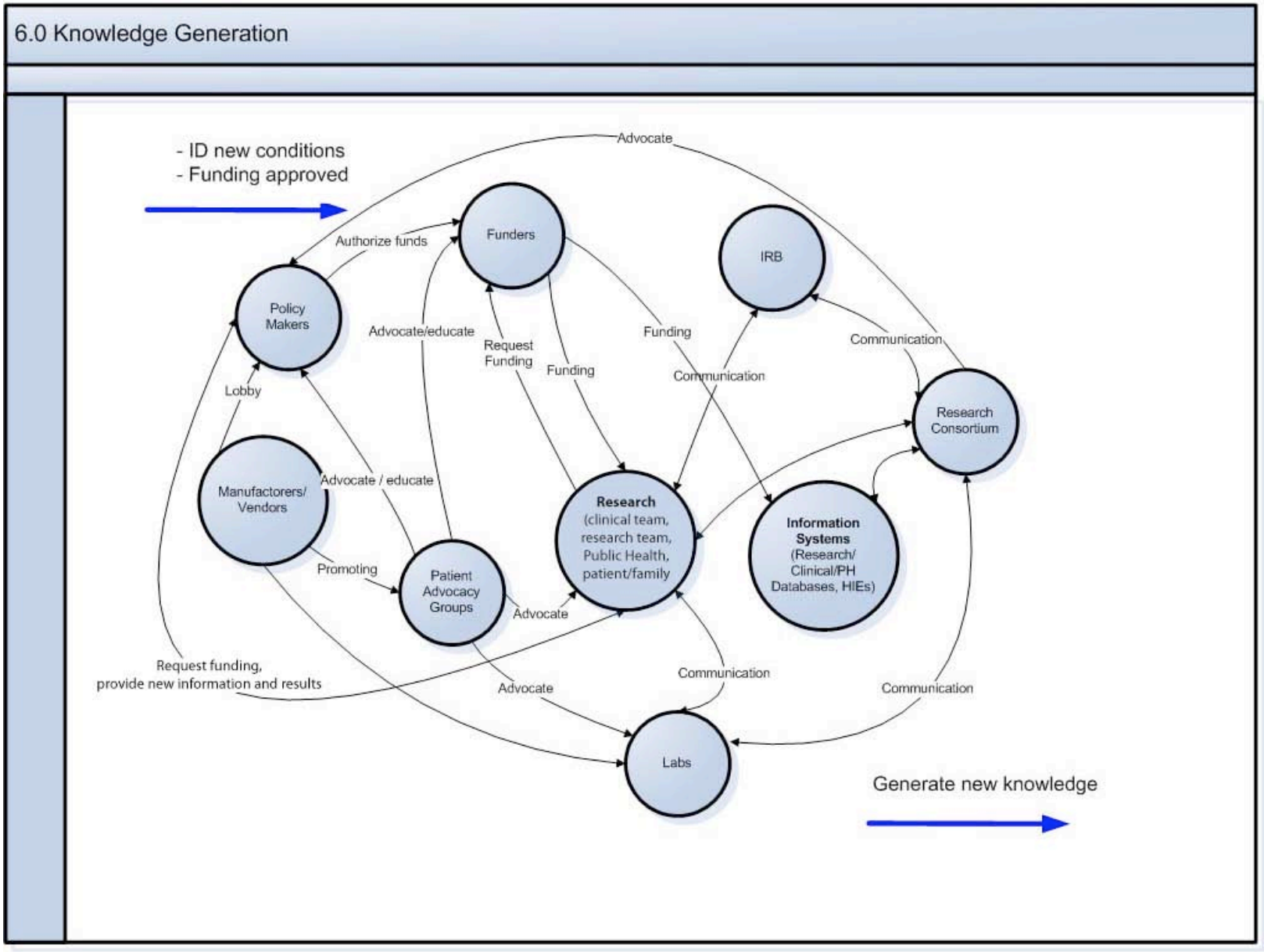
Figure 23: Context Diagram, 5.0 Continuous Quality Improvement (CQI)



6.0 Knowledge Generation

The purpose of this component is to promote the exchange of information among the clinical, research, and public health communities. This exchange also includes funders and policy makers, vendors, laboratories, patient advocacy groups, and research consortiums.

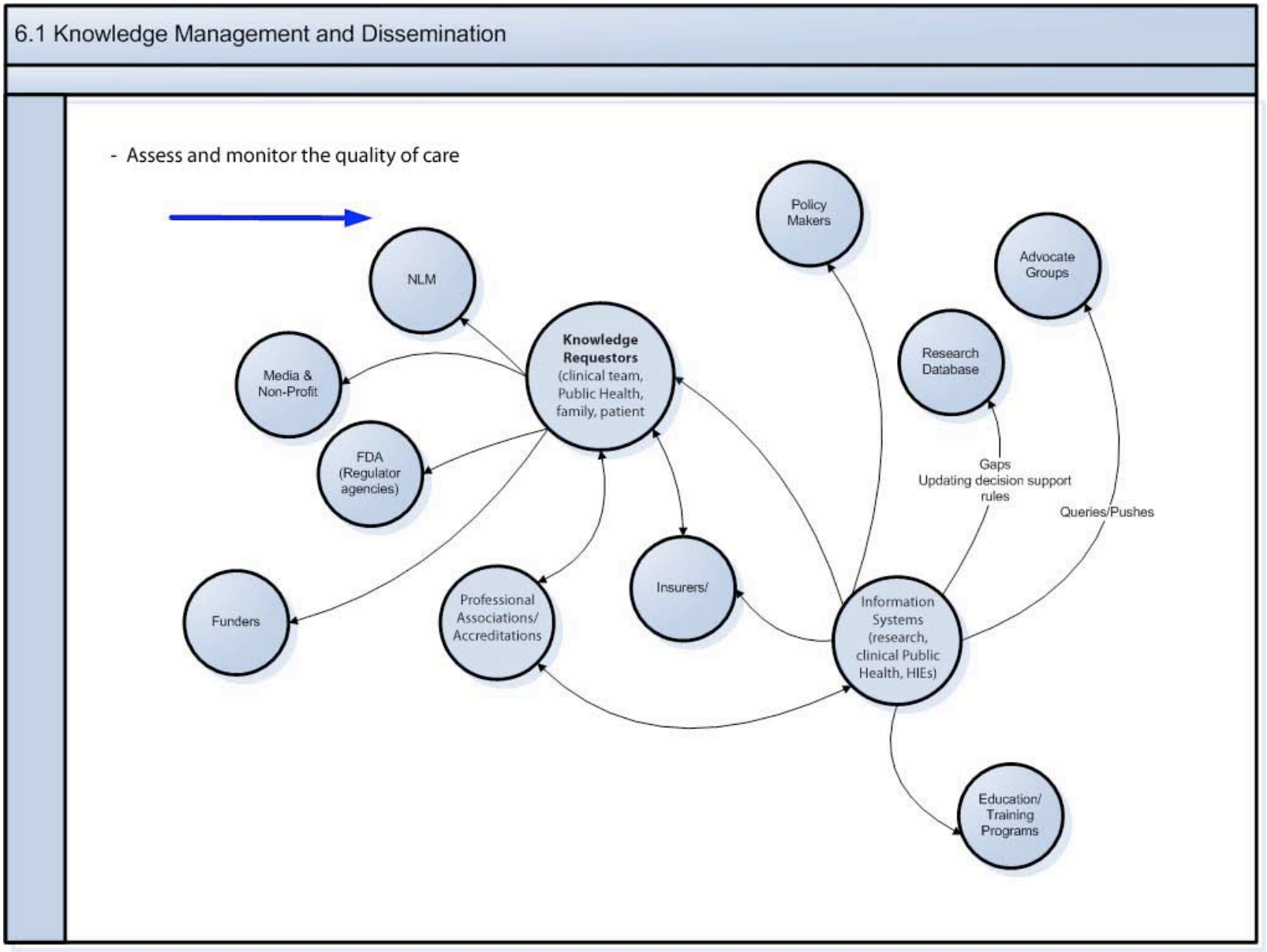
Figure 24: Context Diagram, 6.0 Knowledge



6.1. Knowledge Management and Dissemination

Data generated by clinical, research, and public health entities are now shared within the scientific community through publications distributed to academic and federal entities as well as outside the scientific domain to include advocacy groups and the media.

Figure 25: Context Diagram, 6.1.1 Knowledge Management and Dissemination



Next Steps: Describing the Process of Intervention Management (Long-term Follow-Up)

As completed, the output of the NDBS Workgroup provides a sound basis for continued efforts to define a cohesive approach among all the entities involved in long-term follow-up of children with special health care needs. The care coordination functions as outlined by the NDBS Workgroup contribute to the notion of a collaborative approach among clinicians and public health entities – a much needed, but largely absent component of care. As stated in the January 2007 Association of Maternal and Child Health Program Report, Newborn Screening Long-term Follow-Up Assessment, "Activities occurring after screening and after diagnosis which limit the health consequences of confirmed disorders should be examined to strengthen the overall newborn screening infrastructure. Such activities collectively comprise newborn screening long-term follow-up (LTFU) and have long been an under-funded and comparatively neglected component of the newborn screening system. Nonetheless, LTFU activities are essential to both realize the full public health benefits of newborn screening and document them." The report continues, "Without systems in place to ensure effective disease management after diagnosis, the full promise of newborn screening – the prevention of illness, short-and long-term disability, and premature death – cannot be achieved."[4] In response to the need for a cohesive approach to LTFU for children with special health care needs, the Southeast NBS Genetics Collaborative (Region 3) and the Public Health Informatics Institute convened a subsequent workgroup of experts in pediatric care and public health to describe the clinical and public health roles and responsibilities of all entities involved in LTFU. The scope of this work also included detailing the requirements involved in building the specifications for health information systems that will support care from birth through adulthood, and firmly establish the guidelines for an information system that links individual clinical care with population health and surveillance.

References

[1] Public Health Informatics Institute. *Taking Care of Business. A Collaboration to Define Local Health Department Business Processes*. 2nd ed. Decatur, GA: Public Health Informatics Institute; 2008:25.

[2] Kemper AR, Boyle CA, Aceves J, et al. Long-term follow-up after diagnosis resulting from newborn screening: statement of the US Secretary of Health and Human Services' Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children. *Genet Med*. 2008 Apr; 10(4):259-61.

[3] Medical Home Initiatives for Children with Special Needs Project Advisory Committee. American Academy of Pediatrics. The Medical Home. *Pediatrics*. 2002 Jul; 110 (Part 1 of 1):184-6.

[4] Newborn Screening Long-term Follow-Up Assessment. Washington DC: Association of Maternal and Child Health Programs; 2007. Available at: www.amchp.org. Accessed May 23, 2008.

Glossary of Terms

Business Process

A set of related work tasks designed to produce a specific desired programmatic (business) result. The process involves multiple parties – internal or external – to the organization, and frequently transcends organizational boundaries.

Business Process Analysis

The effort to understand an organization and its purpose while identifying the activities, participants, and information flows that enable the organization to do its work. The output of the business process analysis phase is a model of the business processes consisting of a set of diagrams and textual descriptions to be used for design or redesign of business processes.

Business Process Matrix

The business process matrix depicts the components that characterize a business process – the goals, objectives, triggers, inputs/outputs, business rules, and outcomes (horizontal axis) and a representative set of NDBS business processes (vertical axis) from the master list that was analyzed using the matrix as a tool. Using the matrix to define the components, the NDBS Workgroup members were able to determine whether a given activity had the appropriate structure to be classified as a business process.

Business Rule

A statement that defines or constrains some aspect of the business process. Business rules are intended to assert business structure or to control or influence the behavior of the health agency (business).

Glossary of Terms

Confirmatory/diagnostic testing*

Test to prove or disprove the presence of a specific condition identified by screening tests (for NDBS screening, this testing is from a specimen other than the screening specimen).

Context Diagram

A non-technical graphical tool for recording context information. It consists of the following elements: (1) entity – a person or group of people (e.g., accounts payable clerk or accounts payable department) who performs one or more tasks involved in a process; and (2) transaction – information exchanges between entities. Entities are represented by circles and transactions are represented by arrows. A context diagram may involve all the transactions of a single user of a system or of multiple users. Usually, single-user diagrams are attempted first (for ease), but multi-user diagrams are needed to get a good look at an entire process.

Clinical Care Coordinator (CCC)

As a member of the long-term follow up team, the clinical care coordinator is responsible for ensuring that the patient receives the range of appropriate services from the point of diagnosis through adulthood.

Entity

A person, group, organization, or system that interacts through transactions. Entities are the participants in a process, and are represented by circles in the context diagrams.

*Clinical and Laboratory Standards Institute

Glossary of Terms

Goal

The major health goal that the business process supports. The goal is the end state to be achieved by the work of the health agency and should be defined in terms of the benefits provided to the community/population or individual/client.

Input

Information received by the business process from external sources. Inputs are not generated within the process.

Objective

A concrete statement describing what the business process seeks to achieve. The objective should be specific to the process such that one can evaluate the process or reengineer the process and understand how the process is performing towards achieving the specific objective. A well-worded objective will be SMART (Specific, Measurable, Attainable/Achievable, Realistic, and Time-bound).

Outcome

The resulting transaction of a business process that indicates the objective has been met. Producing or delivering the outcome satisfies the stakeholder of the first event that triggered the business process. Often, measures can be associated with the outcome (e.g., how much, how often, decrease in incidents, etc.). Please note that an outcome can be, but is not necessarily, an output of the process.

Glossary of Terms

Output

Information transferred out from a process. The information may have been the resulting transformation of an input, or it may have been information created within the business process.

Public Health Care Coordinator (PHCC)

As a member of the long-term follow up team, the public health coordinator assesses the completeness of care and provides assurance of the delivery of care.

Stakeholder

A person, group, or business unit that has a share or an interest in a particular activity or set of activities.

Task

A definable piece of 'work' that can be done at one time. A business process is made up of a series of work tasks.

Task Flow Diagram

A graphical tool used to capture the basic flow of tasks as well as the exception flow(s) identified through decision points. The graphical description of tasks shows inputs, processes, and results for each step that makes up a task.

Transaction

An information exchange among entities. Transactions are represented by arrows in context diagrams.

Trigger

Event, action, or state that initiates the first course of action in a business process. A trigger may also be an input, but not necessarily so.

Appendix A
NDBS Workgroup

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Preparation of this report was supported by a grant conferred to the Public Health Informatics Institute from the Maternal and Child Health Bureau, Health Resources and Services Administration, U.S. Department of Health and Human Services.