



Texas Newborn Screening Program Gaps and Barriers Summary Report May 2008

Texas Newborn Screening Performance Measures Project

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1 TNSP Gaps and Barriers Summary Report

1.1 Background Information on the Summary Report

The *Texas Newborn Screening Program Gaps and Barriers Summary Report May 2008* represents the first phase of the ongoing Texas Newborn Screening Performance Measures Project (TNSPMP). The project goal is to develop evidence-based performance measures that link the quality of patient care of affected newborns with the quality of the pre- and post- analytical stages of the newborn screening process.

The TNSPMP project is funded by the Centers for Disease Control and Prevention (CDC) grant (CDC-CI07-710).

The report summarizes gaps and barriers in the Texas Newborn Screening System that were identified by several sources, as outlined below.

It builds upon themes expressed in the National Newborn Screening and Genetics Resource Center (NNSGRC) consultative report, which was produced from a March 2005 review of the Texas Newborn Screening Program (TNSP). The review was at the request of Eduardo J. Sanchez, MD, MPH, past Commissioner of the Texas Department of State Health Services (DSHS).

The report also incorporates internal findings from the Texas Newborn Screening Continuous Quality Improvement (TNSCQI) team using the Program Evaluation and Assessment Scheme (PEAS). The PEAS is a tool for assessing newborn screening programs developed as part of a cooperative agreement between Health Resources and Services Administration, Maternal and Child Health Bureau, Genetics Services Branch, NNSGRC, and The University of Texas Health Science Center at San Antonio - Department of Pediatrics.

The *Texas Newborn Screening Program Gaps and Barriers Summary Report May 2008* also includes documented observations from the Texas Newborn Screening Performance Measures Project External Stakeholder Team (TNSPMP EST). The TNSPMP EST is a 22-member team of stakeholders representing all stages of the newborn screening process including individuals from birthing facilities, primary care physicians, pediatric specialists, medical societies and associations, managed care organizations, parents, other state newborn screening program experts, CDC and subject matter experts.

The report is intended to lay the groundwork for making informed decisions on the development of performance measures using evidence-based approaches. In particular, the report may be used as an information tool to determine literature search efforts.

Although the summary report provides a current status of gaps and barriers as of May 1, 2008, it should be noted that this report is not intended to provide solutions or interventions. Aspects of addressing gaps and barriers will be explored in future objectives of the TNSPMP.

Finally, this report represents a snapshot of the Texas Newborn Screening System that may or may not be the total depiction of the entire system. The Texas Newborn Screening System consists of all the people, organizations, and entities that participate in serving infants born in Texas and providing services to those who have screened positive for newborn screening disorders.

1.2 Framework of the Summary Report

The framework of the *Texas Newborn Screening Program Gaps and Barriers Summary Report May 2008* categorizes gaps and barriers from the following Newborn Screening (NBS) System components:

- Education
- Specimen Collection and Transport
- Timely and Universal Screening
- Laboratory Reporting
- Case Management
- Medical Management
- Program Evaluation
- Program Administration and Finance
- Personnel
- Information Systems
- Contingency Planning

1.3 Identified Gaps

The definition of a gap, as used throughout this document, is a disparity or a significant difference between two situations, attitudes, or perceptions.

For each gap listed in the *Texas Newborn Screening Program Gaps and Barriers Summary Report May 2008*, a letter indicator is provided to indicate the source or sources of the gap or barrier identified.

Each letter represents a different source:

T N P

- T indicates that the observation was noted by the TNSPMP EST.
- N indicates that the finding was noted in the NNSGRC consultative report.
- P indicates that the finding was identified by the TNSCQI Team through conducting a review using the PEAS.

If the gap was noted from several sources, this is indicated by listing multiple letters, such as seen above.

1.4 Identified Barriers

The definition of a barrier, as used throughout this document, is a condition that makes it difficult to make progress or to achieve an objective.

As expected, there is a set of known overarching barriers which makes it difficult to achieve newborn screening system objectives. The barriers identified for each component are unique and specific to that component. Overarching barriers not listed in each individual component are shown below:

- Adequate financing is not in place to effectively fund all newborn screening system components.
- A state size of over 268,500 square miles with 1250 miles of shared border with Mexico poses challenges to administering the NBS program including lack of adequate state resources in rural areas, locating families who have moved across the Mexico border, and ensuring adequate and timely specimen transport.
- Some NBS system components are difficult to manage due in part to the large number of health care facilities/providers who collect specimens and/or follow children diagnosed with disorders. Difficulties include providing rapid communication, timely and ongoing education,

- and monitoring processes of a large and variable provider population.
- Texas has a high percentage of children (per capita) relative to other states.
- For fear of being deported, parents without legal US status sometimes provide insufficient or inaccurate contact information, making it difficult for case management staff to locate them for notification of an abnormal screen

1.5 Status Update

The status update section for each component provides the current status of issues related to each particular topic as of May 1, 2008 and reflect changes made since the NNSGRC consultative review in early 2005. The bulleted points in the status update section are not one-to-one relationships with the gaps identified in the corresponding respective sections.

1.6 Acronym List

Acronyms used throughout this document are:

ACMG	American College of Medical Genetics
CDC	Centers for Disease Control and Prevention
CEU	Continuing Education Units
DSHS	Department of State Health Services
ECI	Early Childhood Intervention Services
EST	External Stakeholder Team
HL7	Health Level 7
IT	Information Technology
LIMS	Laboratory Information Management System
MS/MS	Tandem Mass Spectrometry
NBHS	Newborn Hearing Screening
NBS	Newborn Screening
NNSGRC	National Newborn Screening and Genetics Resource Center
NNSIS	National Newborn Screening Information System
PCP	Primary care physicians
PEAS	Program Evaluation and Assessment Scheme
PKU	Phenylketonuria
TNSCQI	Texas Newborn Screening Continuous Quality Improvement
TNSP	Texas Newborn Screening Program
TNSPMP	Texas Newborn Screening Performance Measures Project

1.7 Acknowledgements

The *Texas Newborn Screening Program Gaps and Barriers Report* benefited from the contributions of various individuals and teams.

A special thanks to the following for contributing, gathering, and compiling information for this report.

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2 Assessment of the Education Component

2.1 Introduction to Education

Newborn screening is a complex system involving screening, short-term follow-up, diagnosis, treatment/management, and evaluation. An active, documented, education plan is vital to the success of the program and should be specifically geared to healthcare workers and families at each phase of the screening process. The plan should be documented and evaluated annually to ensure the effectiveness of the education efforts.

2.2 Education: Identified Gaps

The following gaps in the newborn screening system were identified:

- Providers are not aware of resource materials available from the NBS Program. T
- Educational materials may not consistently address each disorder. N P
- Language appropriate materials vary, thus reducing the ability to impact larger audiences through video, audio and literature. Further, educational materials may not be culturally and linguistically competent for all audiences. N P
- The NBS Education Plan is not reviewed annually to assess overall effectiveness and quality. T P
- Various training needs were identified in the following areas: T N
 - Submission procedures for mailing and courier activities,
 - Submitter and parent understanding of roles and responsibilities,
 - Accessing DSHS or national information on newborn screening, and
 - Accessing information on disorders not tested in Texas.
- Primary care physicians have been overlooked as a resource for providing formal input on education needs as well as input on the overall Newborn Screening Education Plan. N
- The lack of understanding and awareness across the newborn screening system may influence the lack of value for the newborn screening program. T
- Additional outreach methods such as providing public service announcements, distributing disease specific newsletters, and distributing a Practitioner's Manual (which serves as an instructional guide for medical specialist on treating specific disorders) are not in place. T N P
- There is a lack of adequate training for regional staff. T
- Educational materials provided by other entities are not reviewed for accuracy and consistency to ensure they do not conflict with TNSP educational materials. P
- A proactive approach to assist and provide support activities for parents with affected infants is needed. Further, confirming that parent education was received is needed when infants are diagnosed. T N P
- Organizations external to DSHS (including managed care organizations, obstetricians, prenatal hospital case managers, and PCP's) need to align with education efforts in terms of the importance of newborn screening and education efforts on newborn screening results. T
- Hospital and birthing center management may not know about TNSP report cards, which provide feedback to providers on specimen quality and transit time. The report cards may not be reaching management staff responsible for quality and T

educational improvements at their respective site(s).

- Parents are not actively informed via distributed educational or information brochures on where to obtain additional screening. N
- Healthcare professionals often refer to newborn screening as the “PKU test” which causes problems and confusion for case management staff when following up on other non-PKU specific disorders. In some cases, the “PKU” terminology is coded into information systems. T P
- Educational efforts may be missing obstetricians, gynecologists and other prenatal groups. N P

2.3 Education: Identified Barriers

- Employee turnover in health care provider offices makes it difficult to keep provider staff current with training and available educational materials.
- Some physician offices do not have personal computers for staff to download online training.
- Healthcare professionals have time constraints negatively impacting their ability to complete online training.
- Due to the expanded panel of disorders now screened and new knowledge based on research and new technology, protocols/treatments continue to be developed and consensus on educational content for materials from physicians varies.
- Due to the size of the state and number of healthcare providers, it is difficult to monitor the accuracy of educational materials provided by other entities to ensure they do not conflict with program objectives. Additionally, beyond providing education and guidance, the TNSP has no authority to control the content of materials distributed by private facilities.
- Regional staff is also tasked with multiple responsibilities outside of the newborn screening program.
- Language barriers reduce the impact made in captivating larger audiences through video, audio, and literature.

2.4 Education: Status Update

- NBS Brochures are available in English, Spanish and Vietnamese and were revised to ensure reading at a fifth grade level.
- Ad hoc committees are utilized for soliciting disorder-specific feedback for educational activities.
- A survey was administered in summer 2005 to providers for program feedback following the NNSGRC review.
- The distribution of a Practitioners Manual has been discontinued. Resources for health care providers, submitters, physicians, specialists etc. are available on the website including:
 - ACT and FACT sheets
 - Disease specific information that includes comprehensive materials on disorders from various resources.
 - Links to obtain information from other resources outside of Texas
 - Public Service Announcements in both Spanish and English
 - In Fiscal Year 2007, there were 460,409 website visits, 980 requests for literature, and 423,527 pieces of marketing materials distributed.
 - There is an online provider training module on newborn screening that enables participants to receive free CEU's, CME's, etc. Presently, both a Sickle Cell Trait and Genetic module are in the process of being developed for the website.
- Two full time educators were hired to assist with healthcare providers' training needs including:
 - Educators work with the laboratory in identifying hospitals that may demonstrate the need for additional onsite training.
 - Educators promote the DSHS website and provide submitters with a CD that includes a training presentation on specimen collection and other useful links. Additionally, DSHS made these CDs available at the website in March, 2008. As of May 1st, 2008, there have been 140 requests for CDs from the website.
 - Educators have identified a need for parental training and are currently offering in-services to expectant mothers. The NBS website has a link specific to parent concerns.
 - Upon request, the NBS Program provides on-site education as well as education via the website, health fairs, nurse/parent educators, materials, and by other means.
 - An Education Plan was developed for the expansion of newborn screening. In addition, DSHS has hired an additional trainer who will play a significant role in the evaluation of educational efforts.

3 Assessment of the Specimen Collection and Transport Component

3.1 Introduction to Specimen Collection and Transport

Proper specimen collection and information handling are essential to the screening process. Heelstick blood collection processes and the collection device should conform to the most current CLSI/NCCLS Standards. Specimen submission practices should be monitored to provide ongoing education and feedback to submitters on their ability to ensure that each child is successfully screened (with documented refusals when necessary). Important education points to emphasize include ensuring specimens are collected within the desired timeframe, ensuring patient data/demographic information is complete, accurate, and legible and ensuring specimens are dried and mailed within 24 hours.

3.2 Specimen Collection and Transport: Identified Gaps

The following gaps in the newborn screening system were identified:

- Staff at collection facilities needs periodic training and testing on proper specimen collection and transport processes. T N P
- Specimen transit time should be minimized to minimize the effects of extreme environmental conditions such as summer heat and humidity. T N
- Data collected on NBS cards should be reviewed for quality and analyzed such that submitter performance can be evaluated and improved. N P
- A written procedure is needed regarding improvement of submitter performance relative to specimen quality. P
- A mechanism is needed to rapidly notify submitters of unsatisfactory specimens. P
- Newborn screening refusals should be documented systematically according to program guidelines with information provided back to the NBS Program for evaluation. P
- A system should exist for transmission of electronic data between providers and the NBS Program. N
- Specimen collection cards should indicate that they comply with CLSI guidelines. P
- Time of receipt of specimens in the laboratory should be captured in addition to date of receipt. P
- Newborn screening staff at hospitals and birthing centers may not know how to collect and store specimens. T

3.3 Specimen Collection and Transport: Identified Barriers

- Current staffing is inadequate to follow-up on unsatisfactory specimens and to ensure infants are screened.
- Current staffing is inadequate to link first, second and subsequent screens, identify missing specimens, and contact providers for follow-up specimens.
- A change in rules would be needed to collect refusal data.
- There is no effective courier system to ensure timeliness of specimen transport and specimen delivery to the state laboratory.

3.4 Specimen Collection and Transport: Status Update

- The Case Management website is now the major source for provider education and has been expanded to include collection videos and an online Newborn Screening training module offering CEU's upon completion.
- Case Management hired two full-time educators to provide onsite training based on submitter request or significant problems with unsatisfactory specimens.
- Newborn Screening 'Report cards', which provide summaries of specimen rejections and specimen transit times, are mailed quarterly to providers who have submitted specimens for the current calendar year.
- An automated system with web-based remote data entry and direct HL7 file transfer capabilities are scheduled for implementation in the fall of 2008. Several large birthing facilities will be included in the initial pilot phase.
- Newborn screening collection devices meet Clinical Laboratory Standards Institute (CLSI) guidelines although not stated on the device.
- Specimens submitted on expired cards were rejected beginning May 16, 2005.

4 Assessment of the Timely and Universal Screening Component

4.1 Introduction to Timely and Universal Screening

A goal of the newborn screening program is to screen every child born in Texas at two different time intervals; with screening specimens collected once between 24 and 48 hours of age and again at 7 to 14 days of age. These are considered optimum times for specimen collection to ensure proper detection of disorders with minimal false positive rates. Overall, timely screening is critical to provide early identification and intervention for affected newborns prior to the onset of negative health outcomes. Assessment tools and continuous monitoring is needed to ensure those goals are being met.

4.2 Timely and Universal Screening: Identified Gaps

The following gaps in the newborn screening system were identified:

- Enforcement of NBS laws and rules is not covered in the law. N
- Newborn screening staff at hospitals and birthing centers may not be taking specimens at recommended times; first screen at 24 to 48 hours, second screen at 7 to 14 days. T
- Manual or automated mechanisms are not in place to ensure babies born within Texas are screened twice in the appropriate timeframes. N P
- DSHS does not receive notification of refusals. P
- Specimen collection and transport should be documented and tracked by the submitting facility. N P
- Transmittal of specimens to the laboratory after collection should be timely, and the laboratory should monitor transit times. Special attention is needed in rural areas for timely submissions. T P
- Hearing screening is not universally required as a result of certain exemptions reflected in the Texas Administrative Code. However, the Texas Administrative Code, Chapter 37, Rule 37.03 (h), states that if a birthing facility is not required by the Texas Health and Safety Code, Chapter 47, to offer newborn hearing screening, the facility must refer the parents of each newborn delivered in the facility to another birthing facility which offers newborn hearing screening. N
- In some cases, patients are admitted to the neonatal intensive care unit or a hospital with symptoms before the screen results reach the primary care physician. T
- For older children adopted from other countries which may not have a newborn screening program, a timeframe is needed for when NBS is appropriate. It is not clear at what age the screening values are no longer valid. T
- ACMG recommended secondary targets are not included in the Texas newborn screening panel. T

4.3 Timely and Universal Screening: Identified Barriers

- Timely results of confirmatory testing may depend on availability of laboratories to perform the particular test(s). Further, some laboratories may not accept specific insurance or Medicaid.
- Collection of refusal data would require a rule change.
- Current staffing is inadequate to follow-up with submitters on unsatisfactory specimens and to ensure infants are screened.
- Current staffing is inadequate to ensure first, second, and subsequent screens are linked in the database system.
- Military bases or Indian nations are not bound by law to submit newborn screens. Currently, there is no system for assessing the number of these specimens sent to other laboratories or states.

4.4 Timely and Universal Screening: Status Update

- Clarification was provided in the Texas Administrative Code (37.55) that the healthcare provider is the responsible party for ensuring the newborn screen is collected.
- A process was initiated to link all screens (first, second, etc.). As of May 1st, 2008, automatic linking by the software was approximately 70 percent effective for follow-up specimens. With additional manual linking, the number of follow up specimens linked increases to 84 percent.
- Most birthing facilities (greater than 95 percent) submit hearing screening data. All rural hospitals' obstetric units are now reporting to Newborn Hearing Screening.
- The feasibility and costs of integrating newborn screening data with hearing screening and Vital Statistics is currently being evaluated. Database integration would require additional funding.

5 Assessment of the Laboratory Reporting Component

5.1 Introduction to Laboratory Reporting

Final screening results must be accurate and reported in a timely manner. Procedures should be in place to quickly notify case management staff of an abnormal result that indicates a child has one of the screened disorders. In cases where the abnormal result indicates a potential clinical emergency, extra measures are needed to ensure the report is received, including documentation of reporting and report receipt. Submitters should also be notified as quickly as possible if the specimen received in the laboratory is unsuitable for testing.

5.2 Laboratory Reporting: Identified Gaps

The following gaps in the newborn screening system were identified:

- The laboratory does not currently have a method to verify correct delivery of laboratory result reports to submitters. P
- The laboratory does not have a method to verify that laboratory result reports are forwarded from the submitter to the infant's primary care physician. P
- Data collected on NBS cards should be reviewed for quality and analyzed such that submitter performance can be evaluated and improved. N P
- A mechanism is needed to rapidly notify submitters of unsatisfactory specimens. P

5.3 Laboratory Reporting: Identified Barriers

- The current staffing level in the NBS Laboratory is inadequate to take on additional responsibilities.

5.4 Laboratory Reporting: Status Update

- A query for the Laboratory Information Management System is currently being developed, which will verify that specimens received are accessioned. Once specimens are accessioned, queries are run to verify that specimens are reported.
- Full testing and result reporting is performed Monday through Saturday effective February 10, 2007.
- The voice response/fax system was upgraded to include the expanded panel and has been operational since May 2007.
- An automated system with web-based remote data entry and direct Health Level 7 (HL7) file transfer capabilities are scheduled for implementation in the fall of 2008. Many large birthing facilities will be included in the initial pilot phase.

6 Assessment of the Case Management Component

6.1 Introduction to Case Management

DSHS newborn screening follow-up comprises short-term and long-term activities. Short-term follow-up activities by case management staff include locating infants with an abnormal screen and providing the family with resources to ensure infants are promptly seen by a physician. Activities also include ensuring the completion of confirmatory testing, helping with referrals to appropriate specialists, obtaining final diagnosis, and initiation of treatment. Case management staff may also serve as a subject matter resource for the family and primary care physician. Long-term follow-up activities include ongoing tracking of diagnosed patients in order to monitor health outcomes and gauge overall program effectiveness.

- Short-term follow-up activities include notification of the abnormal screen results to medical providers and parent/legal guardian with suggestions on next steps and ends with diagnosis of a disorder or clearing the case.
- Long-term follow-up includes the tracking of diagnosed patients to monitor health outcomes.

Health care providers outside of DSHS also provide case management services to assist families needing ongoing care and services for affected children. Their services typically include assisting families in setting up appointments with specialists, resolving insurance issues, and ongoing access to medical foods, medications, and other services.

By identifying affected infants with potentially catastrophic disorders, contacting guardians, and actively coordinating the infant's care, the affected infant can receive optimal care and secure a better quality of life.

6.2 Case Management: Identified Gaps

The following gaps in the newborn screening system were identified:

- | | |
|---|-------|
| ▪ Improved coordination is needed between case management departments of managed care organizations/insurers and case management at the Texas Newborn Screening program in locating difficult to find patients. | T |
| ▪ Information system does not have complete functionality for data management of all processes, and follow-up data are not systematically retained. | N |
| ▪ Funding is inadequate to cover many areas of the NBS system including travel for consultants, services for families, many case management services, counseling for families, follow-up on unsatisfactory specimens, and other services. | N P |
| ▪ Standardized information regarding long-term outcomes including physical and mental development of children with NBS disorders is sparse or lacking. | T N P |
| ▪ There have been concerns that case management procedures are not well documented (e.g. closing cases, assigning lost to follow-up and updating National Newborn Screening Information System (NNSIS)). | N |
| ▪ There appears to be no formal partnerships or relationships with community based organizations or professional organizations that might assist with family services in general. | N |
| ▪ Feedback to the TNSP from the medical community appears to be an ongoing problem particularly in regards to providing results of biochemical confirmatory tests and final diagnoses on affected infants. | T N P |
| ▪ Case management staff have difficulty contacting providers regarding critical | T |

abnormal results on the weekends and having providers act upon those results,

- Reports from the specialist community indicated concerns with case management following procedures on faxing reports, leaving telephone messages, and being difficult to contact. N P
- There is little or no follow-up of traits beyond initial notification. N

6.3 Case Management: Identified Barriers

- Mobility of families as well as inaccurate contact information poses a challenge to follow-up on screening results.
- It is difficult to track and monitor children transferring across state lines and the Mexican border.
- In some cases, patients cannot be located in order to determine final diagnosis. Case management cannot be conducted if patients cannot be found.
- Hospitals may not have adequate staff to handle case management.
- Physicians have to deal with ramifications of using staff, time, and resources to follow-up on higher numbers of false positives. For example, there is an increase in the number of abnormal results for Congenital Adrenal Hyperplasia and Hypothyroidism in premature infants.

6.4 Case Management: Status Update

- Follow-up data are updated frequently and are retained according to written protocols approved by specialist consultants.
- Only formal advisory committee members can receive funding for travel, and the current statute limits the use and availability of such committees. Despite lack of travel funds, three ad hoc specialist committees continue to meet and provide guidance to the program.
- Funding for Case Management has been increased. Additional funding includes ten percent of the NBS fee that is now allocated to case management. Projected budget figures for year 2008 estimates a funding ratio of laboratory to case management at approximately 5:1 versus the 20:1 funding ratio in 2005.
- Additional follow-up staff has been hired including an ombudsman to assist families, two educators, two nurse team leaders, six nurses, seven additional public health technicians, 16 regional case managers, and two managers.
- Protocols have been established and/or reviewed by program consultants, and orientation processes for new case management staff have been implemented. Written protocols are provided to new staff, which require their signature at the completion of the orientation period.
- In 2005 following the NNSGRC consultative report, an independent survey of stakeholders conducted by TNSP case management staff did not indicate problems with procedures being followed or difficulty contacting staff.
- Case management staff has been provided with dual line phones to minimize the need for customers to leave messages.
- Beginning December 3, 2007, NBS Case Management began notifying families by certified letter of children identified with sickle cell trait. They also receive educational materials and a resource list of sickle cell associations and resources.
- Two formal contracts with community-based sickle cell organizations (Austin and Dallas) have been initiated. Sickle cell associations are notifying families in their catchment areas that have infants born with sickle cell trait.
- Letters are sent to the primary care physician asking that the mother be tested when a baby is confirmed with PKU.

7 Assessment of the Medical Management Component

7.1 Introduction to Medical Management

The newborn screening system extends beyond the Texas Department of State Health Services' Newborn Screening Program to primary care physicians (PCPs), parents, disorder specialists, nutritionists, confirmatory testing laboratories, genetic counselors, and others. Medical management covers the full scope of interventions and resources necessary to provide ongoing care of affected newborns and support for their families.

7.2 Medical Management: Identified Gaps

The following gaps in the newborn screening system were identified:

- | | | | |
|--|---|---|---|
| ▪ Current funding for NBS does not incorporate comprehensive newborn screening system costs. | T | N | P |
| ▪ Counseling services are not provided through the NBS Program. | | N | P |
| ○ Qualifications of counselors are unknown | | | |
| ○ Quality/consistency/availability of counseling services provided is unknown | | | |
| ○ Counseling is not part of the follow-up case record | | | |
| ▪ There is a lack of specialists and lack of access to specialists to assist parents with proper and timely treatment of newborn screening disorders in some areas of the state, particularly rural areas. For example, infants with potential Hypothyroidism may be placed on synthroid as a temporary intervention while on a waiting list to see an endocrinologist. | T | | |
| ▪ Providers are not consistently utilizing specialty assistance. For example, an infant may be placed on therapy without ever contacting an endocrinologist. | T | N | |
| ▪ Providers do not consistently provide case information to the NBS Program. | | N | |
| ▪ There is not an active strategy (such as an advisory committee) to obtain formal input from system stakeholders on a periodic basis to evaluate and provide recommendations for the NBS Program. | T | N | P |
| ▪ The NBS Program needs to be more proactive in providing parent support activities. | | N | |
| ▪ Although ACT and FACT sheets are available, providers need clearer guidelines regarding appropriate actions for abnormal results indicating specific disorders. An ACTion (ACT) sheet describes the short term actions a health professional should follow in communicating with the family and determining the appropriate steps in the follow-up of the infant that has screened positive. These information sheets were adapted from information provided on the American College of Medical Genetics (ACMG) website. | T | | |
| ▪ Transition to adult care is needed in conjunction with the newborn screening system. | T | | |
| ▪ Long term follow-up activities within the Texas Newborn Screening program are limited thus hindering evaluation of outcomes and the effectiveness of newborn screening. | T | | |
| ▪ Timely receipt of results on hospital charts is needed at hospitals and clinics. | T | | |
| ▪ There is a lack of genetic counseling access. Appropriate referrals are not being made. | T | | |
| ▪ The program does not take an active interest in helping to mediate reimbursement issues with insurance companies. | | | N |

- There is a lack of referrals to Early Childhood Intervention Services (ECI) which is the statewide program for families with children, birth to three, with (or at risk for) disabilities and developmental delays. The Texas Administrative Code, Chapter 37, Rule 37.503 (g) notes that programs that offer outpatient screening or audiologic services shall refer an infant with confirmed or suspected hearing loss to ECI. T

7.3 Medical Management: Identified Barriers

- The DSHS newborn screening program does not have jurisdiction over genetic counseling services and medical management protocols.
- Current law limits the use of official advisory committees unless specifically named in statute.
- Missed appointments make it difficult for providers/physicians to provide proper care to patients.
- There is inadequate time and human resources to assure communication between primary care providers and specialists.
- Managed care does not ensure access to specialists.
- Timely receipt of confirmatory test results may depend on availability of laboratories for a particular test. Local draw stations may not accept specific insurance or Medicaid.

7.4 Medical Management: Status Update

- Funding has been increased for follow-up and case management activities, in addition to Medicaid/Title V and CSHCN funding. Currently, 10 percent of the NBS fee is also allocated to case management activities.
- An ombudsman was hired to assist parents and providers who are having difficulty with accessing appropriate medical care and addressing insurance issues. This person has knowledge of local community-based resources across the state and can also assist parents in that regard.
- Case Management has hired two full-time educators and educational materials have been greatly enhanced.
 - ACT sheets providing recommendations on appropriate follow-up for positive newborn screens have been adapted for Texas and made available online to healthcare providers.
 - FACT sheets to assist providers with discussing positive newborn screens with parents were developed and are available online for either providers or parents.
 - Case Management follow-up staff provides ACT and FACT sheets directly to providers caring for infants with positive screens.
 - Educational materials for parents and healthcare providers are available online, at health fairs, and from educators.
 - Online training modules have been developed for newborn screening and newborn hearing screening (NBHS). Continuing Education Units (CEUs) are provided at no cost to doctors, nurses, audiologists and social workers.
- In December 2007, a sickle cell trait notification process was implemented. This process has provided the NBS Program with opportunities to build relationships with community based sickle-cell organizations.
- The Interagency Council for Genetics, whose membership includes geneticists, consumers of genetic services or family members of consumers of genetic services, and representatives from the March of Dimes, the Department of Aging and Disability Services, and the Texas Department of Insurance, serves as a resource for the NBS Program.
- CDC grant funding for assessment of the Texas NBS system is being used to identify and document gaps and barriers in the pre- and post-analytical processes. Information collected through this grant activity could be used to improve various components of the NBS system.

8 Assessment of the Program Evaluation Component

8.1 Introduction to Program Evaluation

Ongoing evaluation and monitoring of newborn screening system components is necessary in order to determine if program goals are being met and to address any gaps or problems identified. A formal plan should be in place that defines select indicators, assigns responsibility for their monitoring, and suggests methods to remediate problems when they occur. Program evaluation should encompass both short-term and long-term activities, and annual summary reports should be shared with internal and external stakeholders.

8.2 Program Evaluation: Identified Gaps

The following gaps in the newborn screening system were identified:

- There are no established procedures for reviewing and validating DSHS laboratory results via comparisons of 1st and 2nd screen results, confirmatory results from external laboratories, or recall rates from other state programs. N P
- There does not appear to be a documented, uniform system for collecting, validating and maintaining case data, or procedures for short-term program evaluation. N P
- Although laboratory and case management data are provided to specific stakeholder groups, the program lacks a formal annual report summarizing key performance indicators for the stakeholders for each phase of the screening system. N P
- There does not appear to be a documented, uniform system for collecting, validating and maintaining long-term follow-up data, resulting in limited information on disorder specific outcomes and overall program impact. T N P
- Specialists and primary care physicians sometimes fail to submit (or delay submission of) information necessary for appropriate case management and program evaluation, including confirmatory laboratory results and information on cases detected after a normal screen. T N
- By statute, the Texas Health and Safety Code requires an ongoing roster of children diagnosed with a NBS disorder. However, the current system appears to be loosely organized and inconsistently managed. N

8.3 Program Evaluation: Identified Barriers

- A uniform database for follow-up is difficult to establish due to the different types of data needed for each specific disorder.
- There are no NBS rules related to enforcement of program initiatives.

8.4 Program Evaluation: Status Update

- Although not in real-time, a process is in place to match second screen abnormal specimens with initial specimens that were normal. A process for validating first and second screens has not been addressed.
- If there is a notable discrepancy between the first and second screen results, the laboratory and the submitter of the specimen are notified, and a review is initiated immediately to determine the cause of the discrepancy.
- The NBS rules were amended to strengthen and clarify the roles and responsibilities for physicians and providers attending the delivery of newborn in addition to the care of the newborn if the newborn is no longer in the hospital. Responsibilities include not only screening the child but also assisting the department with follow-up on positive screens and reporting all confirmed cases to the department.
- Case Management is diligent in attempting to gather all necessary results associated with an abnormal screen. NBS protocols require that case management staff continue to request confirmatory results in order to identify a diagnosis or so that a case can be cleared and closed out.
- As a result of the expansion of the program, Team Lead positions were created in NBS Case Management, and Team Leads have the responsibility for conducting quality assurance activities related to case management data. These activities include the collection, validation and the management of abnormal cases by case management staff (nurses and public health technicians). Quality assurance reports generated by the Laboratory Information Management System (LIMS) are used by the Team Leads to validate data.
- At the annual consultant meetings, feedback regarding program performance and recommendations for program revision is a means for program evaluation.
- A project to develop a database for long-term follow-up data on diagnosed cases is being pursued with DSHS information technology staff.
- The annual consultant meetings are held to share NBS program results. The data shared is a means for feedback on performance, recommendations and evaluation.
- To keep abreast of changes and updates, laboratory and case management staff share/discuss major protocol updates at weekly meetings and in preparation for annual consultant meetings.
- A Continuous Quality Improvement Team has been established consisting of Laboratory and Case Management staff that meets weekly on internal program assessment and continuous improvement efforts.

9 Assessment of the Program Administration and Financing Component

9.1 Introduction to Program Administration and Financing

In order to manage the Newborn Screening Program; there should be a plan for ongoing assessment of administrative and financial concerns. Topics to consider include formal mechanisms for major system changes, adequate newborn screening program staffing, the addition and deletion of disorders, new and evolving analytical systems, system oversight capacity, and involvement of stakeholders in addressing ongoing system needs and future growth.

9.2 Program Administration and Financing: Identified Gaps

The following gaps in the newborn screening system were identified:

- | | |
|--|-------|
| ▪ The division of laboratory and case management functions between two different organizational areas complicates program administration and financing. | N |
| ▪ Data are needed to support recommendations for legislative initiatives. | T |
| ▪ A formal process for proposing new tests to be included in the screening panel does not exist. (E.g. Cystic Fibrosis). | T N P |
| ▪ There is no mechanism for enforcement of the law or rules requiring NBS. | N |
| ▪ A cost analysis of system components including education and comprehensive follow-up services is needed to monitor cost benefits of newborn screening and determine which services the fee should cover. | N |
| ▪ Funding should adequately cover both laboratory and follow-up activities. | T N P |
| ▪ An external advisory committee does not exist to provide program recommendations. | T N P |
| ▪ Parents of affected newborns are not provided with timely assistance in dealing with financial issues, including third-party reimbursement, eligibility with special organizations, and other financial issues affecting the receipt of case-related medical services. | T P |
| ▪ Grants should be considered as opportunities for additional program funding. | N |
| ▪ To promote necessary program improvements, external stakeholders need to establish a solid relationship with the new commissioner who was assigned in 2007. | T |
| ▪ Data is needed to support recommendations for legislative initiatives. | T |
| ▪ DSHS does not screen newborns for the 29 disorders recommended by March of Dimes. | T |
| ▪ Strategic legislative initiatives need to be planned and considered (for example, the consideration of mandatory distribution of newborn screening information similar to cord blood banking). | T |
| ▪ Program finance and business plans should be inclusive of system issues such as confirmatory/diagnostic clinical services, genetic counseling services, nutritional counseling services, employee continuing education, employee recruitment and retention, mechanisms for assessing adequacy of payment for medical management, program/test development, and data integration. | P |
| ▪ There is no procedure in place for rapid communication of critical program issues to | P |

submitters or health care providers.

9.3 Program Administration and Financing: Identified Barriers

- Startup funding is required for NBS Program expansion and is typically appropriated by the state legislature.
- Available financing currently does not allow for complete expansion to the ACMG core and secondary panels endorsed by advocate groups for parents and affected newborns.
- Program administration for DSHS NBS is divided between two divisions with different administrative management.
- Current law limits the use of official advisory committees unless specifically named in statute.

9.4 Program Administration and Financing: Status Update

- House Bill (HB) 790, 79th Regular Session required newborn screening for the disorders listed on the core panel as recommended by the ACMG, or other appropriate screening panel, to the extent funding was available. This change in statute provides authority to add disorders without additional legislation given that the disorders are part of a DSHS recognized recommended panel and funding is available. The Texas Administrative Code (Rules) lists each disorder currently tested in Texas. Per protocol, testing additional newborn screening disorders requires a scope revision in the Code prior to implementation
- Newborn screening for core disorders identifiable by tandem mass spectrometry (MS/MS) and Biotinidase deficiency were implemented by early 2007 in accordance with HB790.
- Funding for implementation of cystic fibrosis newborn screening is not currently available.
- The NBS fee was increased November 1, 2006 from \$19.50 to \$29.50, approximately two months in advance of implementing screening for the new disorders. Ten percent of the NBS fee for the newborn screening kit card is allocated to Case Management.
- Funding has been increased for follow-up activities. However, follow-up costs such as diagnostic testing and follow-up visits are not covered by the fee except for indigent cases.
- With the addition of new case management and laboratory staff, DSHS is now actively seeking and applying for grant opportunities. The laboratory was recently awarded a \$300,000 per year CDC program assessment grant which will provide opportunities for growth and quality improvement in both divisions.

10 Assessment of the Personnel Component

10.1 Introduction to Personnel

Employee training is necessary to ensure qualified personnel are assigned to analytical duties in the newborn screening laboratory and to oversee follow-up activities in the case management program. A comprehensive employee training program should include new employee orientation, ongoing staff development, and documented performance assessment activities. Recruitment and retention of sufficient qualified staff to ensure program goals are met should be a program priority.

10.2 Personnel: Identified Gaps

The following gaps in the newborn screening system were identified:

- Few continuing education opportunities are available for laboratory and case management staff to keep abreast of new information, knowledge, and best practices as it relates to newborn screening. T N P
- More may need to be done to ensure employee competency is formally assessed and approved prior to independent work assignments. P
- Ensuring up-to-date training for personnel across the newborn screening system, both internal and external to the DSHS newborn screening program, is challenging considering high turnover rates across the system. N P
- DSHS staff contacting providers via telephone may not have sufficient technical knowledge or updated information to respond to questions about the test results. N
- Within the DSHS NBS program, there is neither an active process in place for the recruitment and retention of talented staff or a career ladder to provide incentive for growth and development. It has also been noted that performance measures for personnel do not provide a clear path for incentives or rewards for good work. P
- There is not sufficient staff to adequately administer the program within the DSHS NBS program. N P
- There may not be a sufficient number of specialists, specifically metabolic specialists, to assist families of affected newborns. T

10.3 Personnel: Identified Barriers

- Recruitment and retention of personnel within TNSP continues to be a limitation.

10.4 Personnel: Status Update

- Case Management staff is required to take the NBS online training.
- Nurse Team Leaders have been established to provide training, technical assistance and oversight to daily work performed by the NBS staff.
- Laboratory personnel are not approved to work independently in screening analysis until a formal assessment of competency has been completed by the lead supervisor.
- Additional laboratory and case management staff have been hired. The 17 new laboratory positions include three microbiologists for specimen accessioning, ten chemists and medical technologists for the tandem mass spectrometry and biotinidase screening teams, one public health technician for linking specimens, one program specialist for NBS special projects, and a manager for the tandem mass spectrometry screening area. Additional follow-up staff includes an ombudsman to assist families, six nurses, seven additional public health technicians, 16 regional case managers, two managers and two educators who provide onsite training based on submitter request or significant problems with unsatisfactory specimens.

11 Assessment of the Information System Component

11.1 Introduction to Information System

Information systems are the system of organizations, data records, activities, and persons that process newborn screening data and information, including manual processes or automated processes. A component of the information system includes the computerized laboratory and case management information systems which is essential in managing data generated for all phases of the newborn screening system. Information on newborn screening data becomes knowledge when the information is evaluated and interpreted for purposes of taking action or making decisions (e.g. for improving the newborn screening program, for improving quality of services, etc.).

11.2 Information System: Identified Gaps

The following gaps in the newborn screening system were identified:

- Texas Newborn Screening Program data are not integrated with health database systems such as vital records, newborn hearing, immunizations, children with special health care needs and other databases such as lead, birth defects, WIC, etc. N P
- Texas Newborn Screening Program is lacking an integrated information system with hospitals, providers, and community based organizations to exchange data including screen results, confirmatory results, diagnosis, and treatment for affected infants. T N P
- Without a complete linking process between first and second specimens, it is difficult to evaluate testing protocols and to ensure definitive testing of both first and second screen for each newborn within Texas. N P
- Laboratory and case management databases do not link screening result data and case outcome data in a linear or useful way. Data must be linked manually for purposes of program evaluation. P
- Written procedures associated with maintaining the integrity of the newborn screening information system within DSHS (network, hardware, and software components) are not easily accessible due to the complexity of the organizational infrastructure that supports computer maintenance. N P
- Information systems are not in place to link primary care physicians with specialists. N
- NBS records need to be up-to-date, secure, and easily accessible to providers (e.g. the ImmTrak system in Texas). T
- State of the art newborn screening program databases are needed to support easy access to NBS results from throughout the U.S. for when children move across state lines. T
- Standard operating procedures are lacking to govern the data entry of case management information. Therefore, the database may contain incorrect or inadequate data. P
- Current DSHS data system does not support automated downloads to national NBS database (NNSIS). T

11.3 Information System: Identified Barriers

- The design of the Laboratory Information Management System software is not patient-centric, but rather specimen centric. Thus, a direct link between laboratory testing data and case outcome data does not exist.
- Specialists and providers do not receive financial support or incentives for linkage or integration efforts.

11.4 Information System: Status Update

- Prior to and during expansion, laboratory and case management personnel actively worked with the LIMS vendor to provide input on upgrades to the LIMS.
- A process has been established to document and track LIMS issues to resolution.
- Electronic data services for NBS demographics and laboratory reports will be available via a web-based/file transfer system due for completion by the fall of 2008 to be offered to all NBS submitters. Prior to developing final specifications, large volume submitters were visited to evaluate their current systems and to determine their specific needs.
- Currently, DSHS is evaluating the feasibility and costs of integrating newborn screening data with hearing screening and vital statistics, as well as evaluating the possibility of enhancing the web-based system to include web-based options for healthcare providers to electronically submit confirmatory test results and electronically report diagnosed cases.
 - Additional funding would be required for database integration.
- A process was initiated to link all screens (first, second, etc.). As of May 1st, 2008, automatic linking by the software was approximately 70 percent effective for follow-up specimens. With additional manual linking, the number of follow up specimens linked increases to 84 percent. Continuous efforts are being made to increase the efficiency and success of the linking process.
- The maintenance contract for the LIMS includes the creation of ten reports provided annually by the vendor to meet the program's growing needs.
- Laboratory IT Application and Development staff received hands-on training from the vendor and are now better able to meet NBS Program staff needs for report development.
- The laboratory appointed two super users who have been trained on system query development and providing ad hoc reports.

12 Assessment of the Contingency Planning Component

12.1 Introduction to Contingency Planning

Continuity of operations planning (“contingency planning”) is essential for ensuring continuous operations during unplanned events such as natural disasters, power outages, network downtime, or reagent shortages. These disruptions in normal workflow may be temporary (a few hours) to disruption for an extended period of time. Planning should incorporate not only laboratory testing and program case management functions but also issues affecting system partners.

12.2 Contingency Planning: Identified Gaps

The following gaps in the newborn screening system were identified:

- DSHS does not have a comprehensive contingency plan in place to ensure continuity of the newborn screening system in case of an emergency. P
- Internal/external stakeholders have not been asked for input on contingency planning. N P
- A system for ensuring that administrative, laboratory and follow-up records are appropriately protected and maintained has not been established. P
- A method for ensuring that all newborns are screened and receive needed follow-up has not been developed. P
- There appears to be over reliance on a single commercial vendor for laboratory tests and computerization without sufficient attention to back-up should the vendor experience problems. N

12.3 Contingency Planning: Identified Barriers

- There is a lack of time and resources to devote to the rapid development and implementation of this plan.

12.4 Contingency Planning: Status Update

- Per Texas Administrative Code, Texas agencies are required to undertake business continuity planning to minimize the effects of a disaster on services and business processes. The DSHS Commissioner approved the agency’s policy for business continuity planning and disaster recovery planning in September 2006.
- Functions most critical to the immediate mission of the agency, if operations are disrupted for an extended period, were identified as Tier 1 Functions. In 2007, the Newborn Screening Program has been classified as a Tier 1 Function within the DSHS agency.
- The development of a contingency plan has been initiated for both the NBS laboratory and NBS Case Management.
- The Newborn Screening Continuous Quality Improvement Process team will be assisting with the development of a detailed contingency plan for the Newborn Screening program.

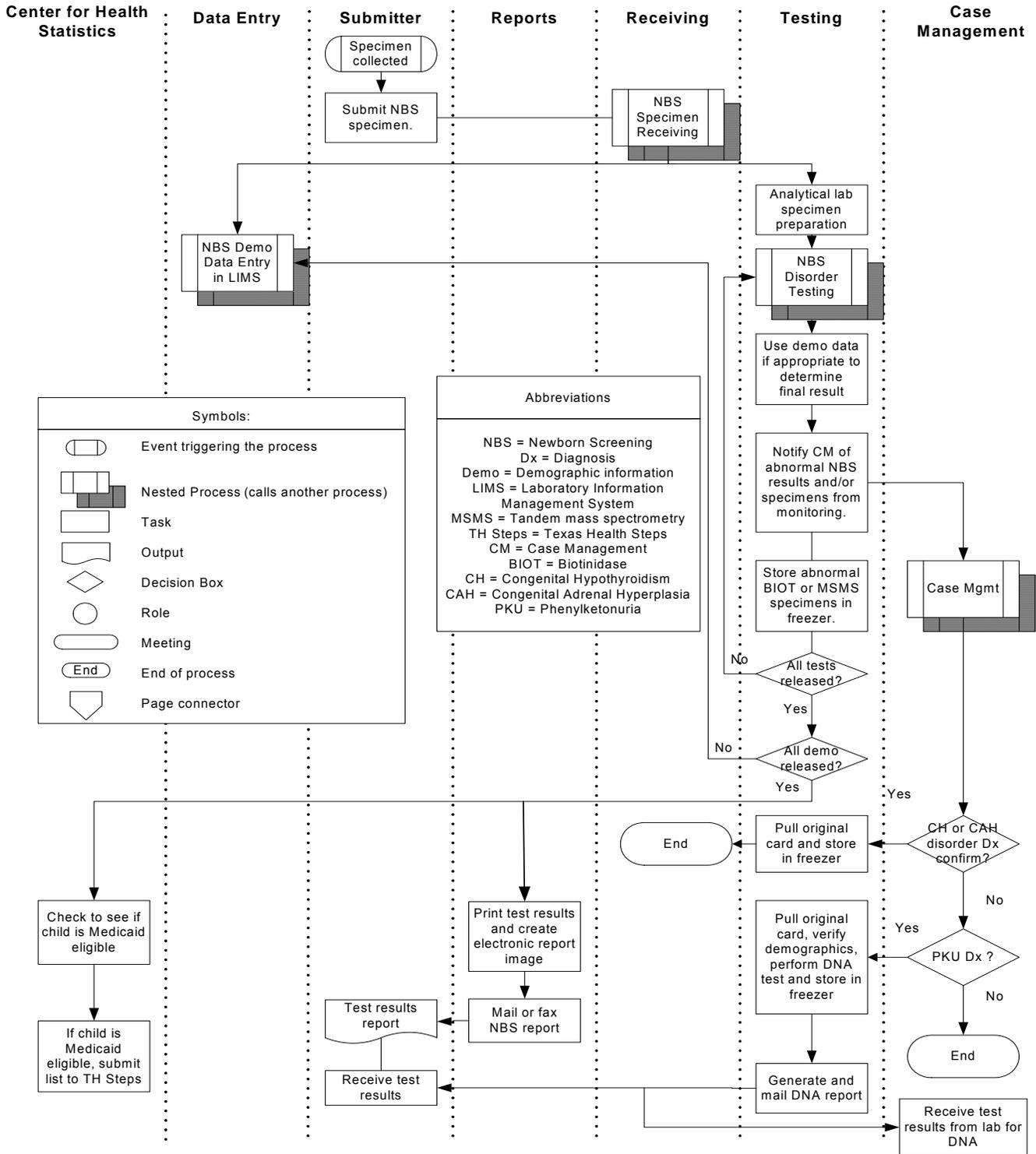
13 Newborn Screening Processes

13.1 DSHS NBS Program

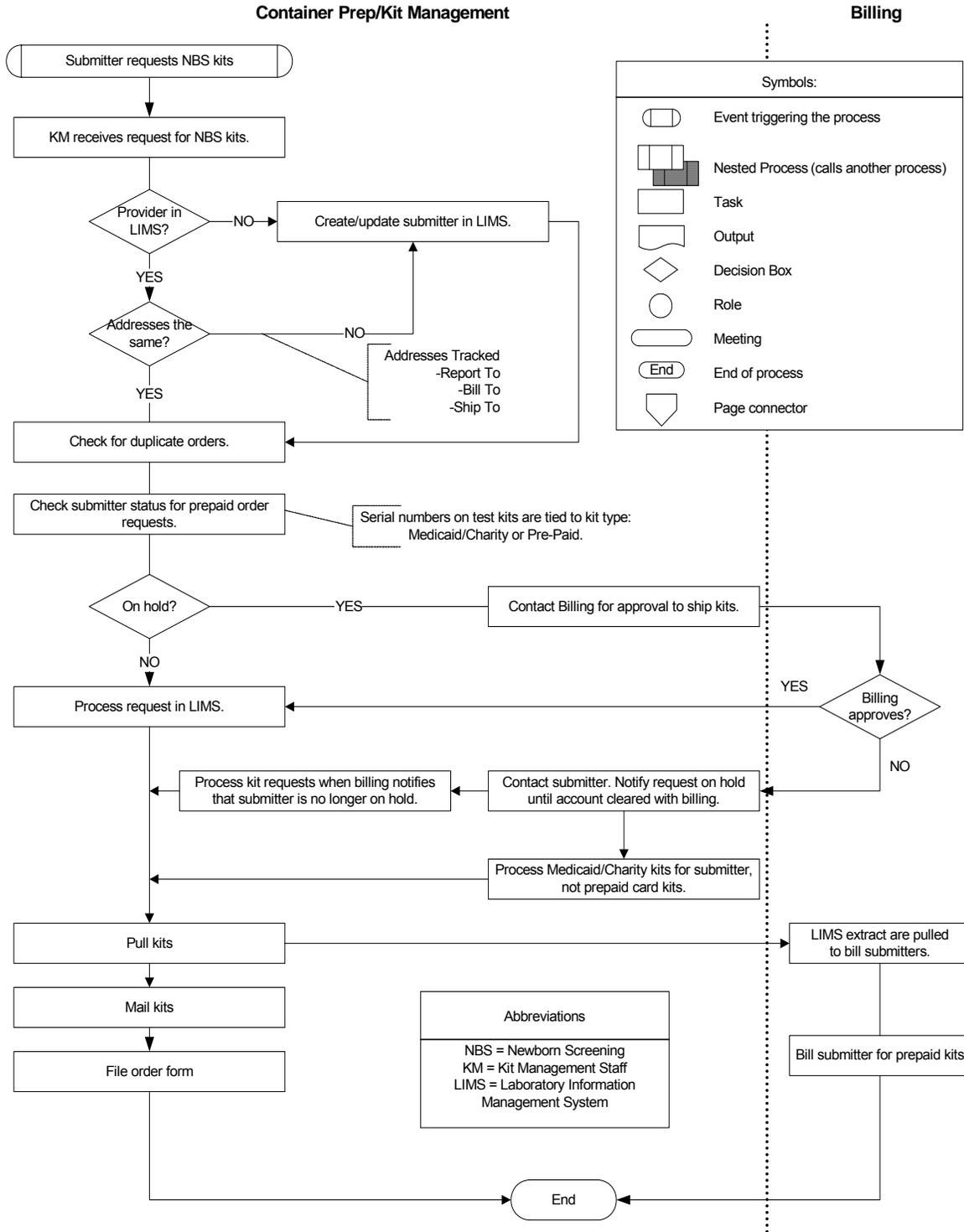
This section shows workflows for the newborn screening program within the Texas Department of State Health Services

Name	Detail
Newborn Screening Workflow	Workflow showing an overview of the entire newborn screening processes within DSHS using a cross functional depiction across various departments. Contains nested processes for NBS Specimen Receiving, NBS Demographic Data Entry in LIMS, NBS Disorder Testing, and NBS Case Management.
NBS Specimen Collection Kit Management Workflow	Overview of kit management with cross functional depiction with billing department.
Newborn Screening Receiving Workflow	Workflow showing the processes for receiving specimens at the DSHS laboratory using cross functional depiction with Data Entry and Laboratory areas. Nested processes include NBS Disorder Testing and NBS Demographic Data Entry in LIMS.
Run NBS Disorder Tests	Workflow showing testing of the various NBS disorder tests. Does not contain nested processes.
NBS Demographic Data Entry in LIMS	Workflow for demographic entry area within the laboratory.
PKU Monitoring	Workflow for PKU monitoring using cross functional depiction across the NBS System.
NBS Case Management	Overview of case management workflow. Nested process includes the initial notification of abnormal screens.
Initial Notification of Abnormal Screen	Workflow showing the steps taken to establish first contact with PCPs.
Galactosemia (GAL)	Follow-up workflow of Galactosemia with Initial Notification of Abnormal Screen as a nested process.
Hemaglobinopathy	Follow-up workflow of Hemaglobinopathy with Initial Notification of Abnormal Screen as a nested process.
Phenylketonuria (PKU)	Follow-up workflow of Phenylketonuria with Initial Notification of Abnormal Screen as a nested process.
Hypothyroidism	Follow-up workflow of Hypothyroidism with Initial Notification of Abnormal Screen as a nested process.
Congenital Adrenal Hyperplasia (CAH)	Follow-up workflow of CAH with Initial Notification of Abnormal Screen as a nested process.

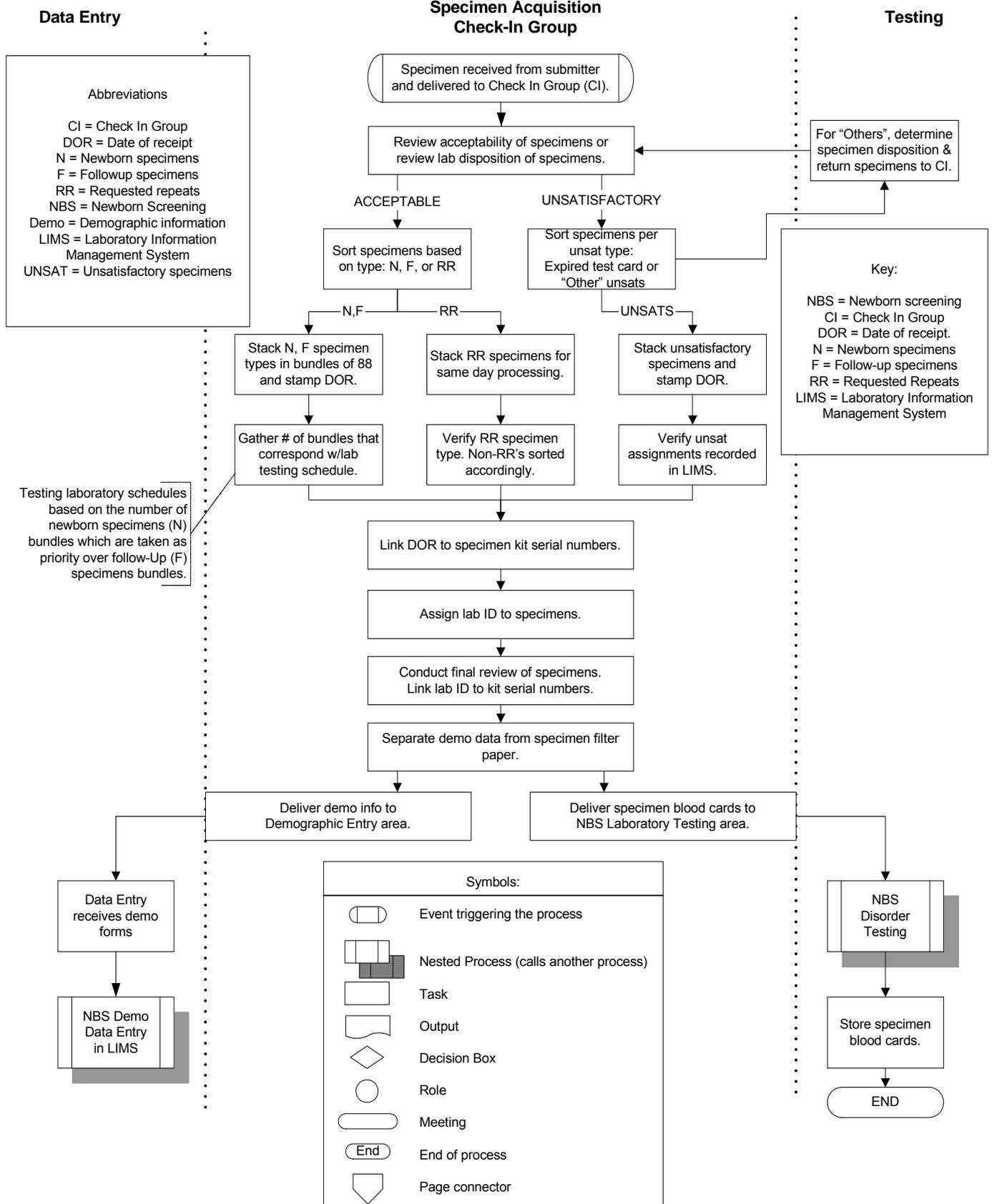
Newborn Screening Workflow



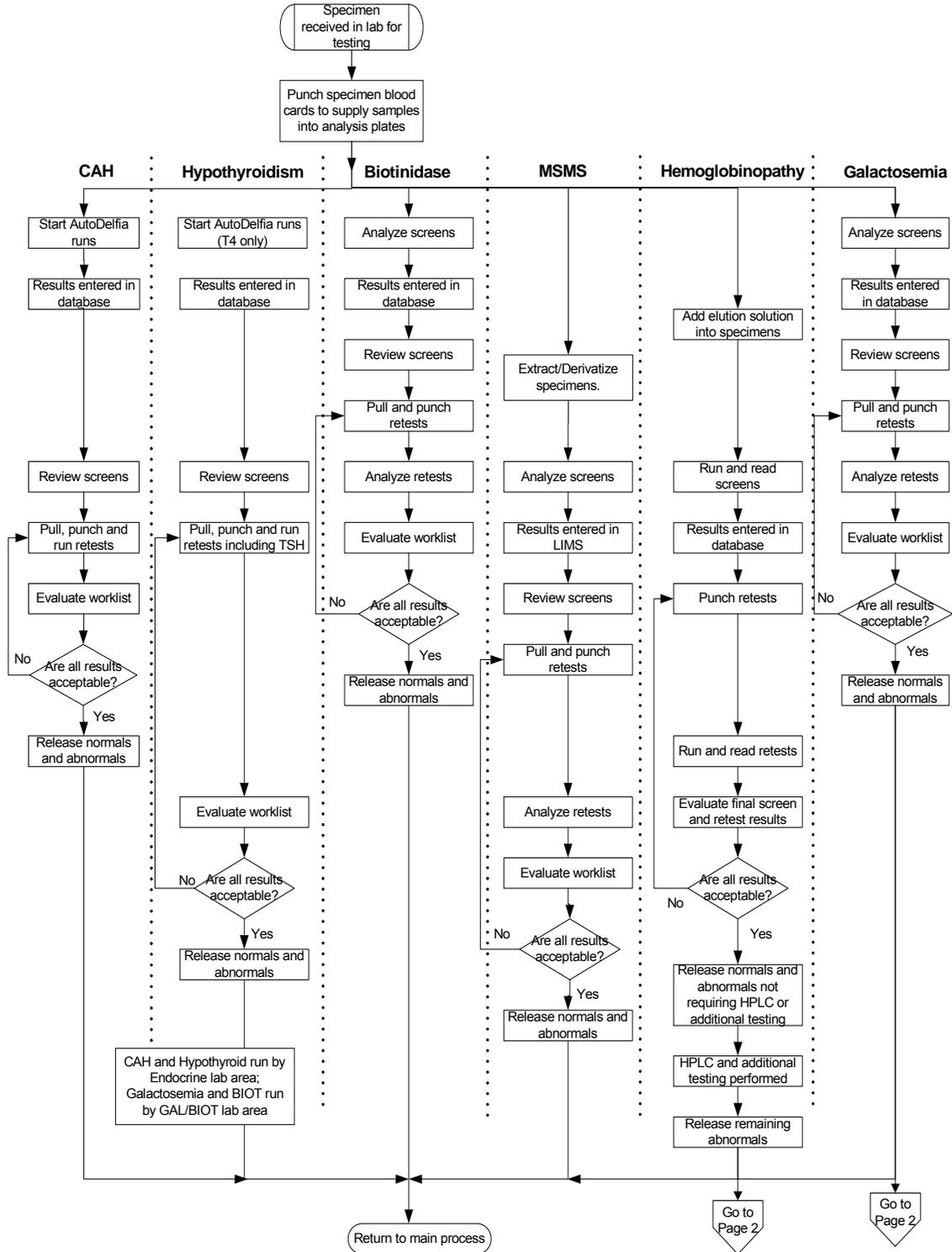
NBS Specimen Collection Kit Management Workflow



NBS Specimen Receiving Workflow



Run NBS Disorder Tests



Run NBS Disorder Tests

CAH

Hypothyroidism

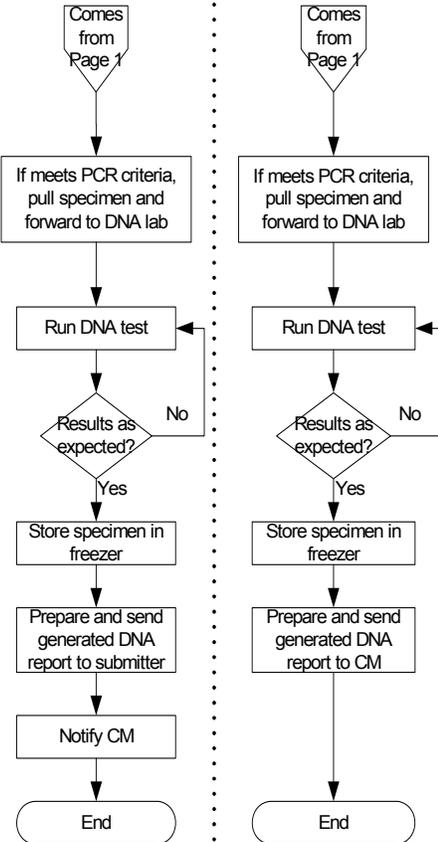
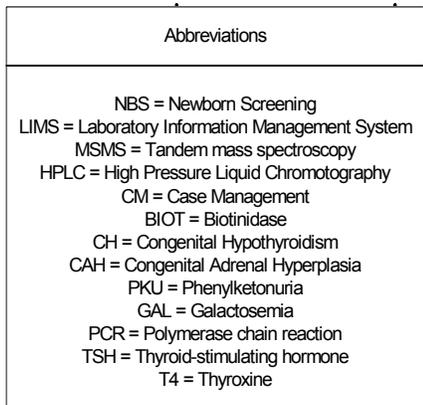
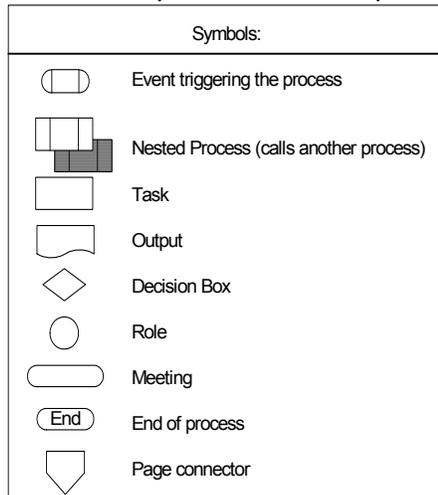
Biotinidase

MSMS

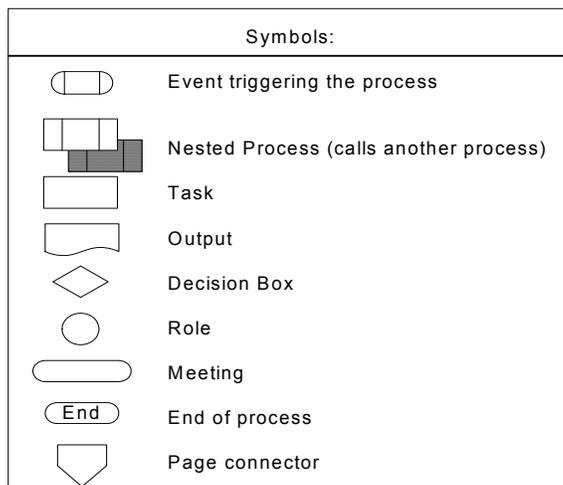
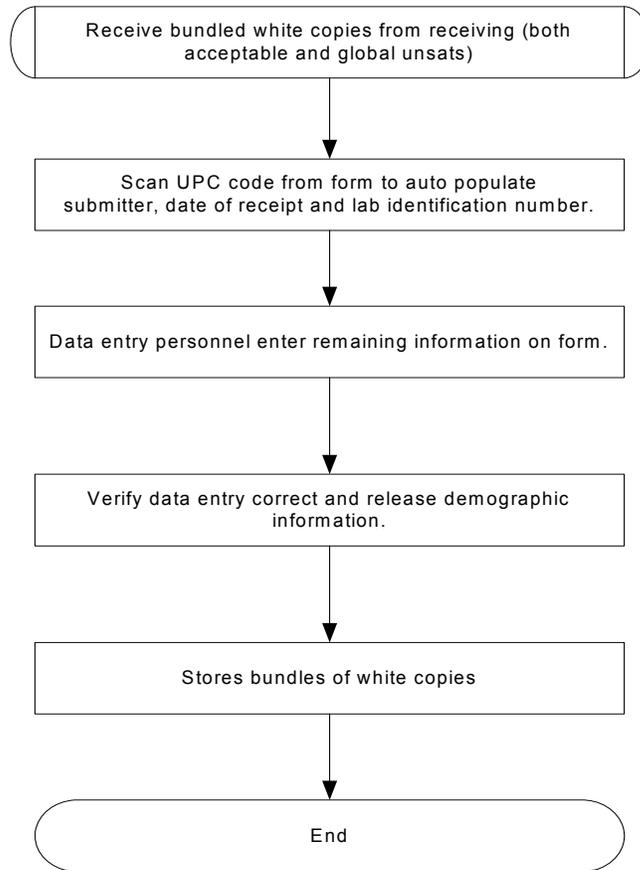
Hemoglobinopathy

Galactosemia

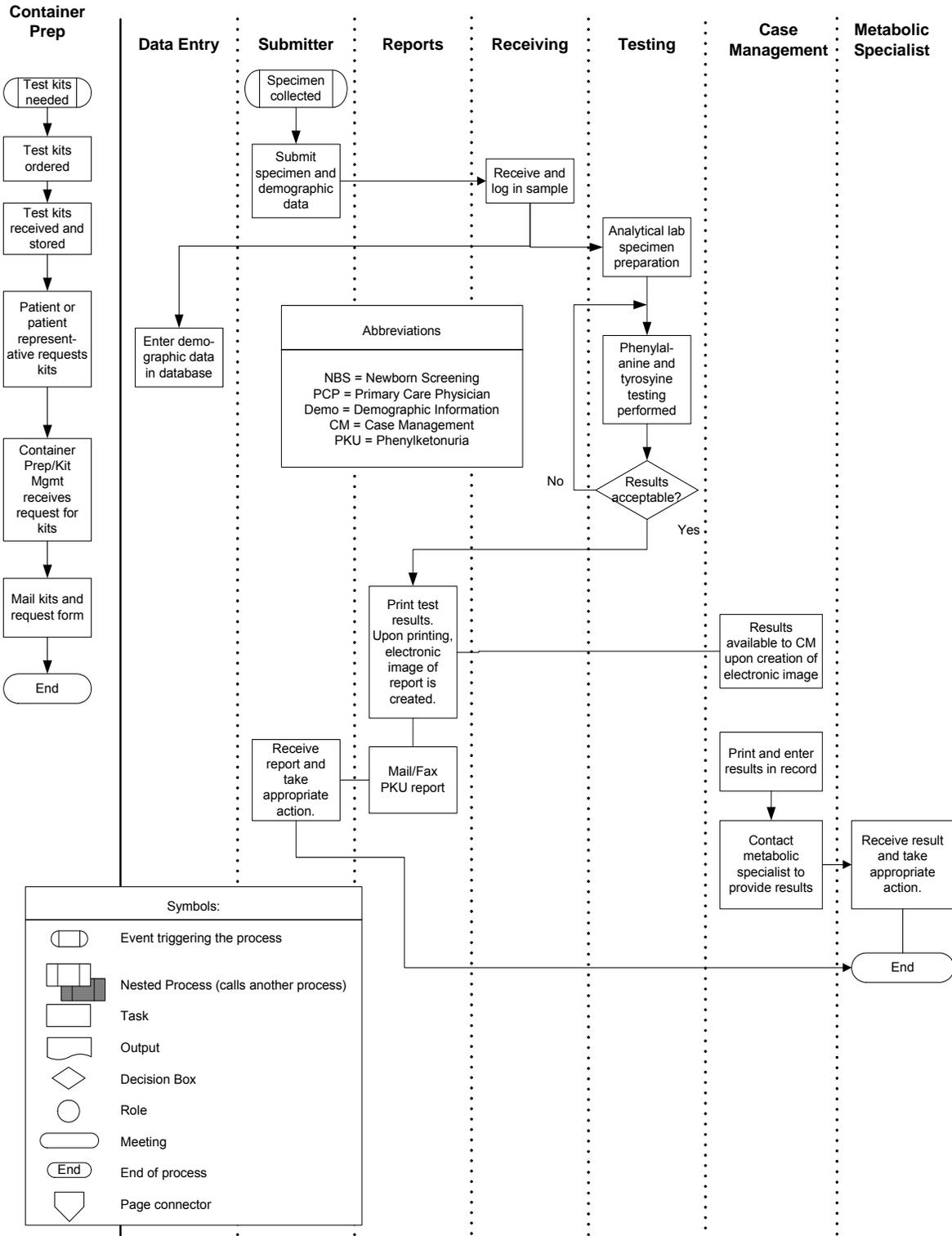
CAH and Hypothyroid run by Endocrine lab area; Galactosemia and BIOT run by GAL/BIOT lab area



NBS Demographic Data Entry in LIMS



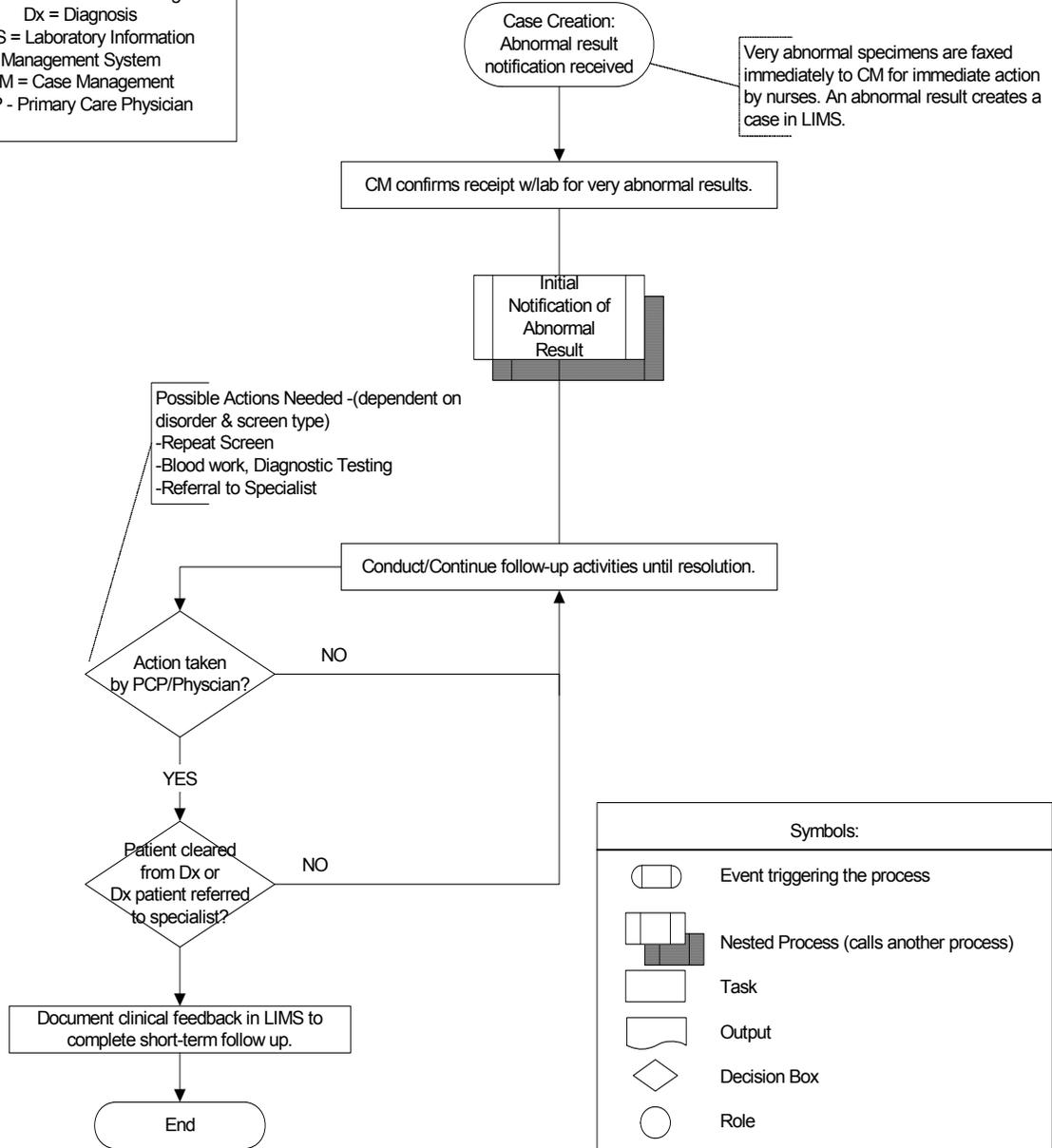
PKU Monitoring



NBS Case Management

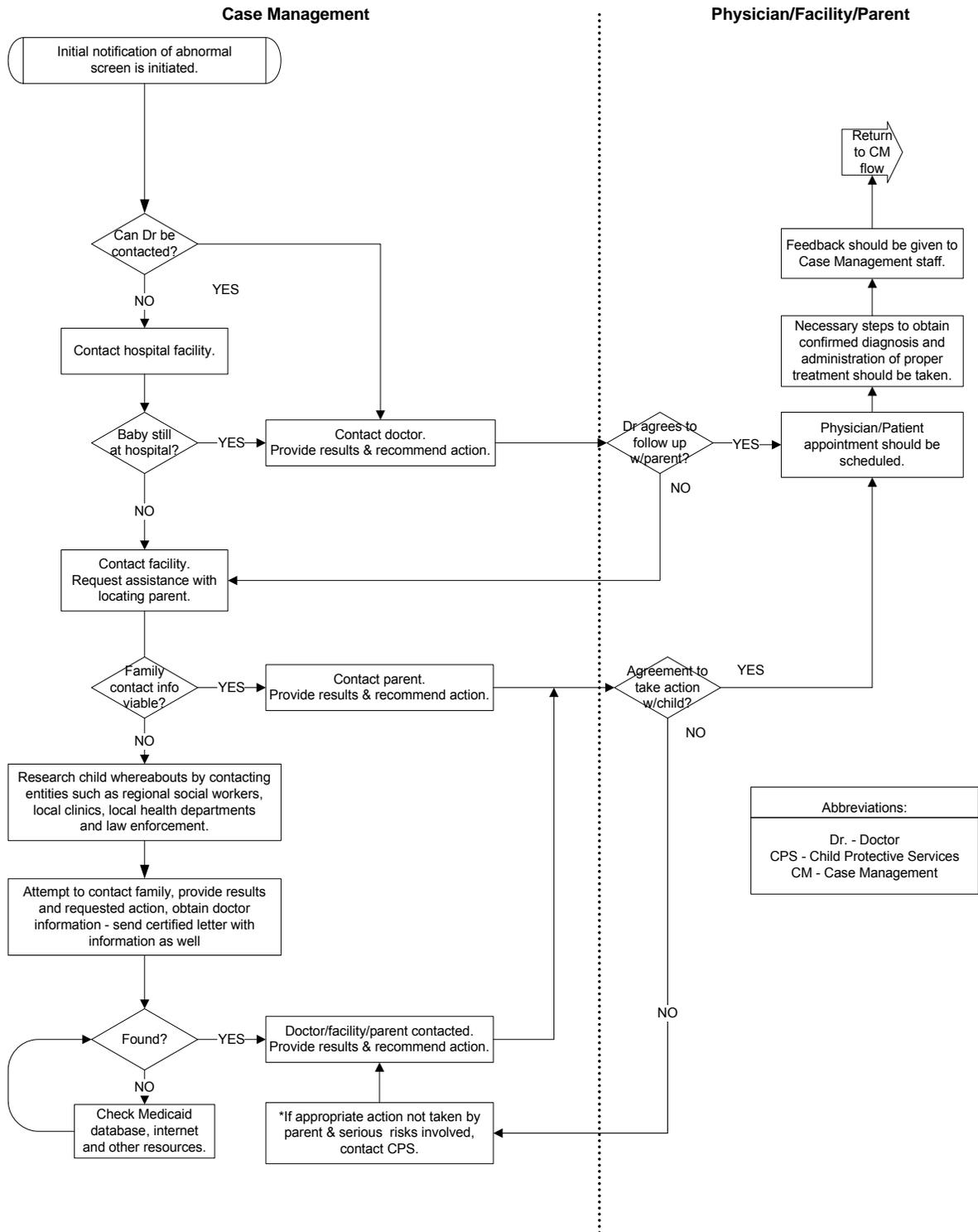
Abbreviations
NBS = Newborn Screening Dx = Diagnosis
LIMS = Laboratory Information Management System
CM = Case Management
PCP - Primary Care Physician

NBS = Newborn Screening
Dx = Diagnosis
LIMS = Laboratory Information Management System
CM = Case Management
PCP - Primary Care Physician

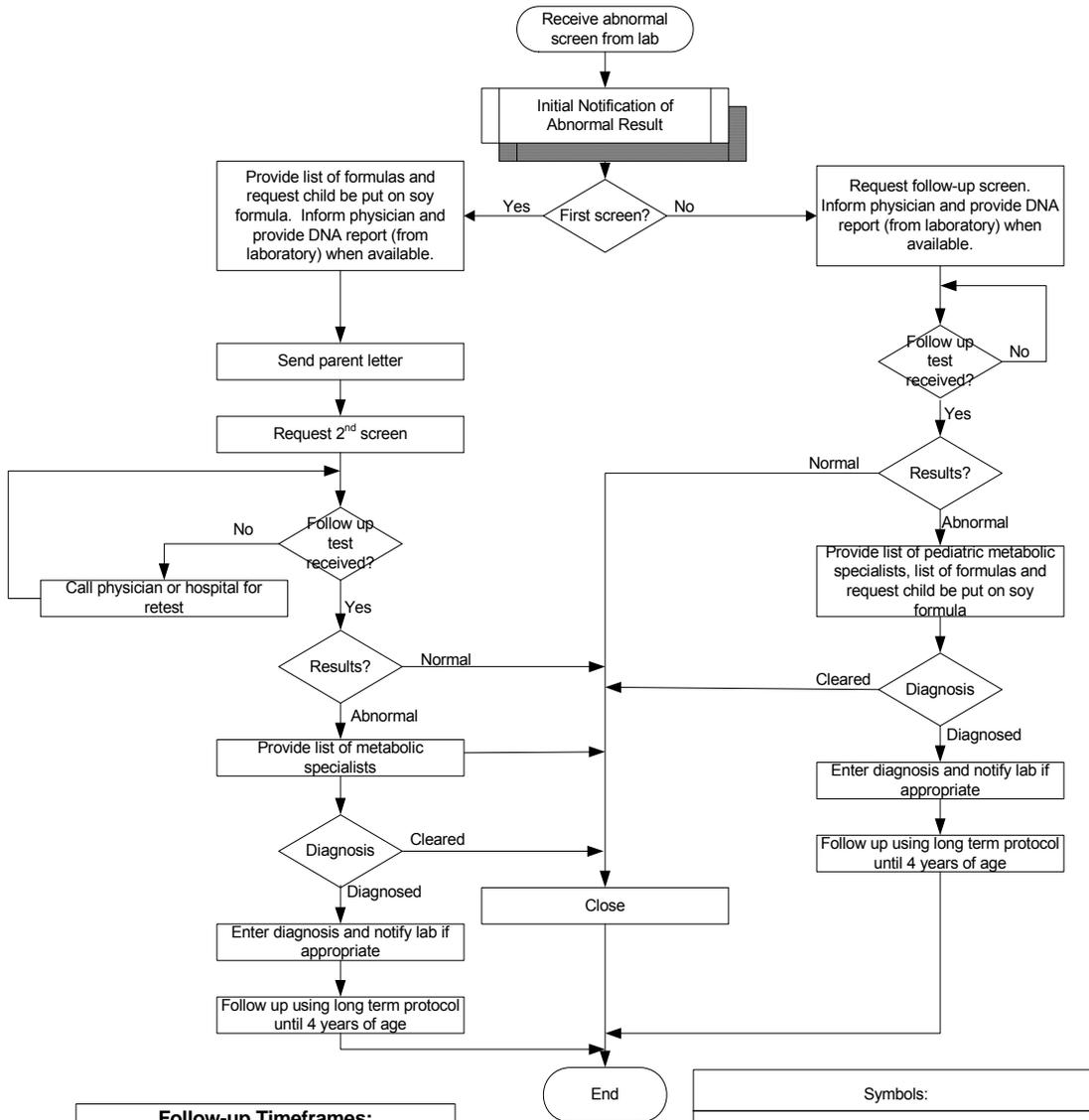


Symbols:	
	Event triggering the process
	Nested Process (calls another process)
	Task
	Output
	Decision Box
	Role
	Meeting
	End of process
	Page connector

Initial Notification of Abnormal Screen



Galactosemia (GAL)



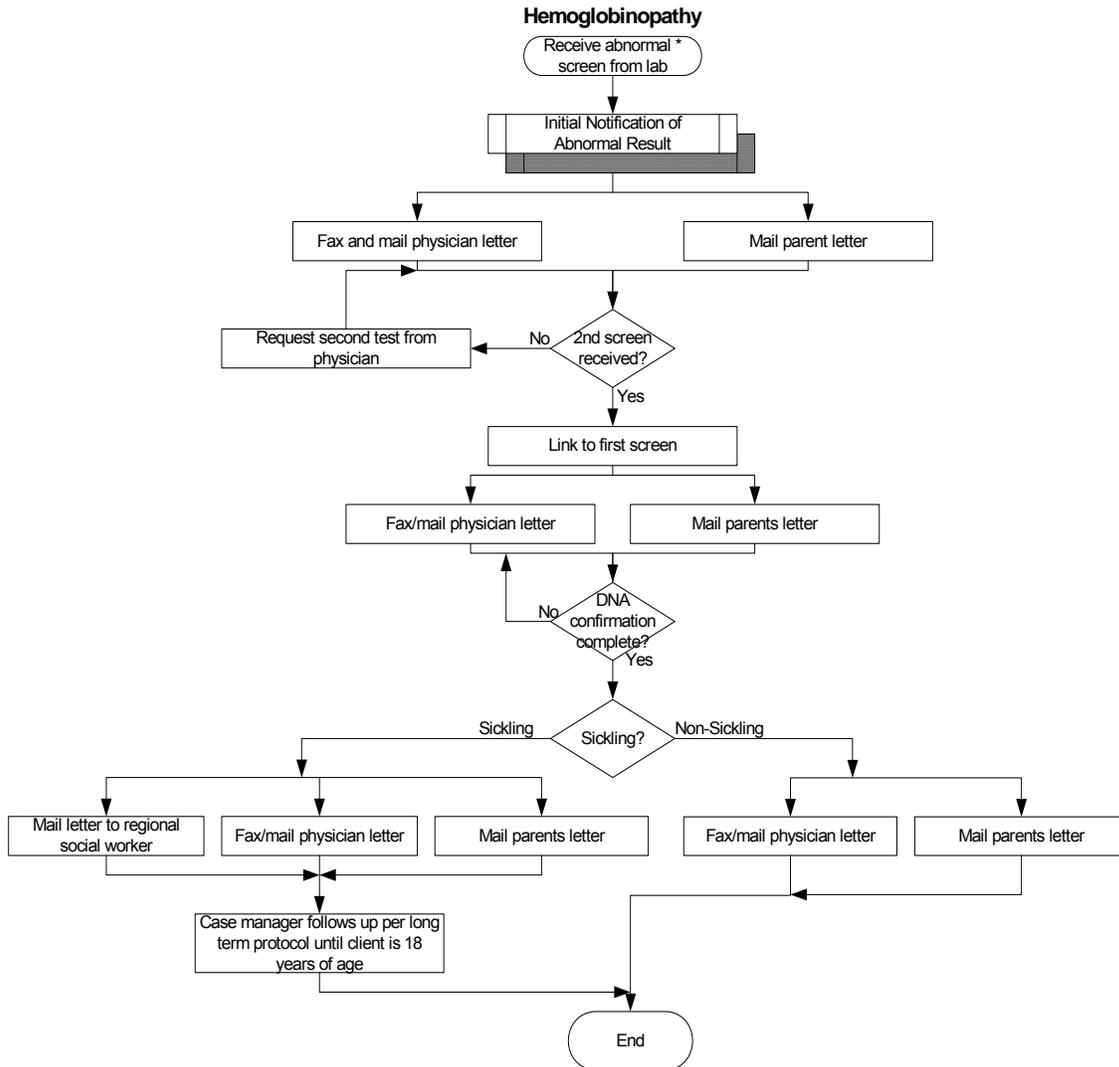
Follow-up Timeframes:

For very elevated – Call back every 24 hours until child is treated

For slightly or moderately elevated – follow up every 2 to 3 days until child is treated

Symbols:

	Event triggering the process
	Nested Process (calls another process)
	Task
	Output
	Decision Box
	Role
	Meeting
	End of process
	Page connector



Follow-up Timeframes:
 Notify primary care physician within 48 hours.

* Most trait conditions are not referred to case management

Symbols:	
	Event triggering the process
	Nested Process (calls another process)
	Task
	Output
	Decision Box
	Role
	Meeting
	End of process
	Page connector

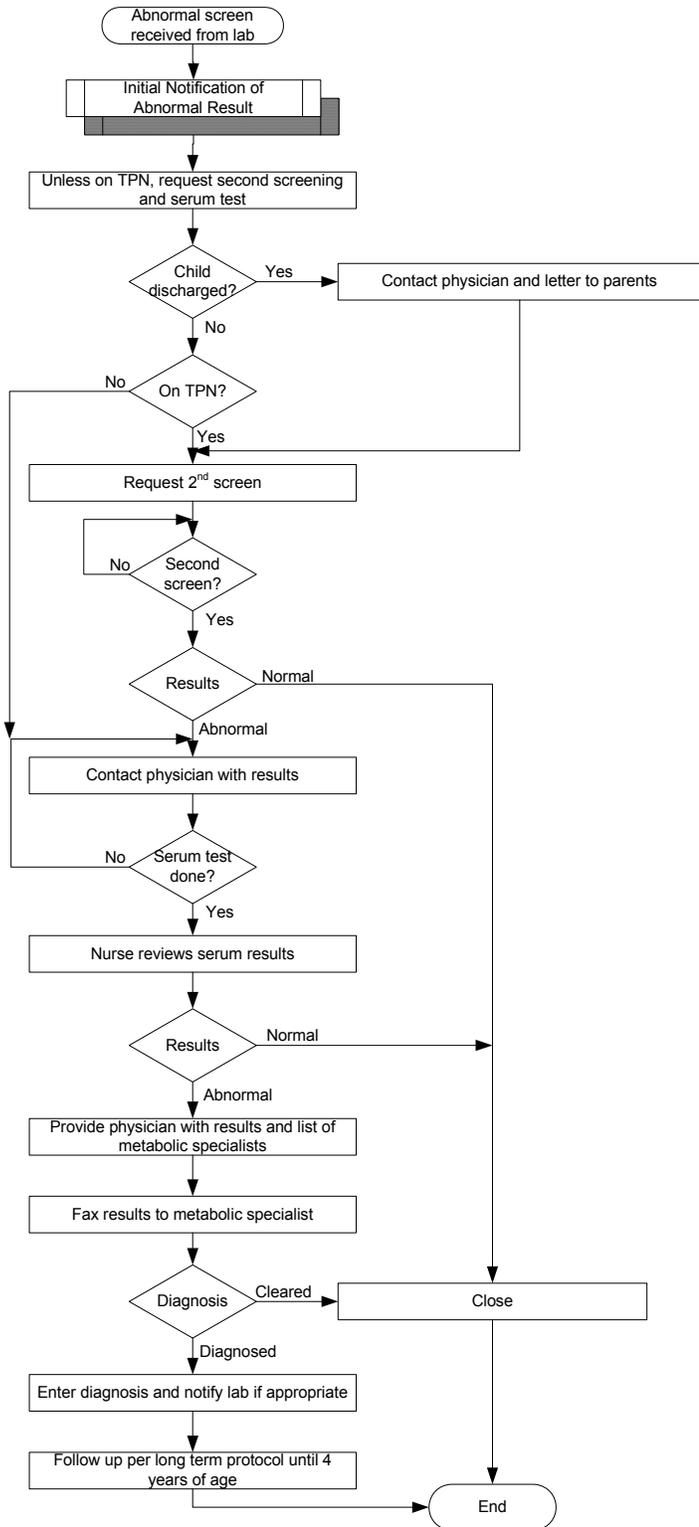
Phenylketonuria (PKU)

Abbreviations
TPN = Total parenteral nutrition

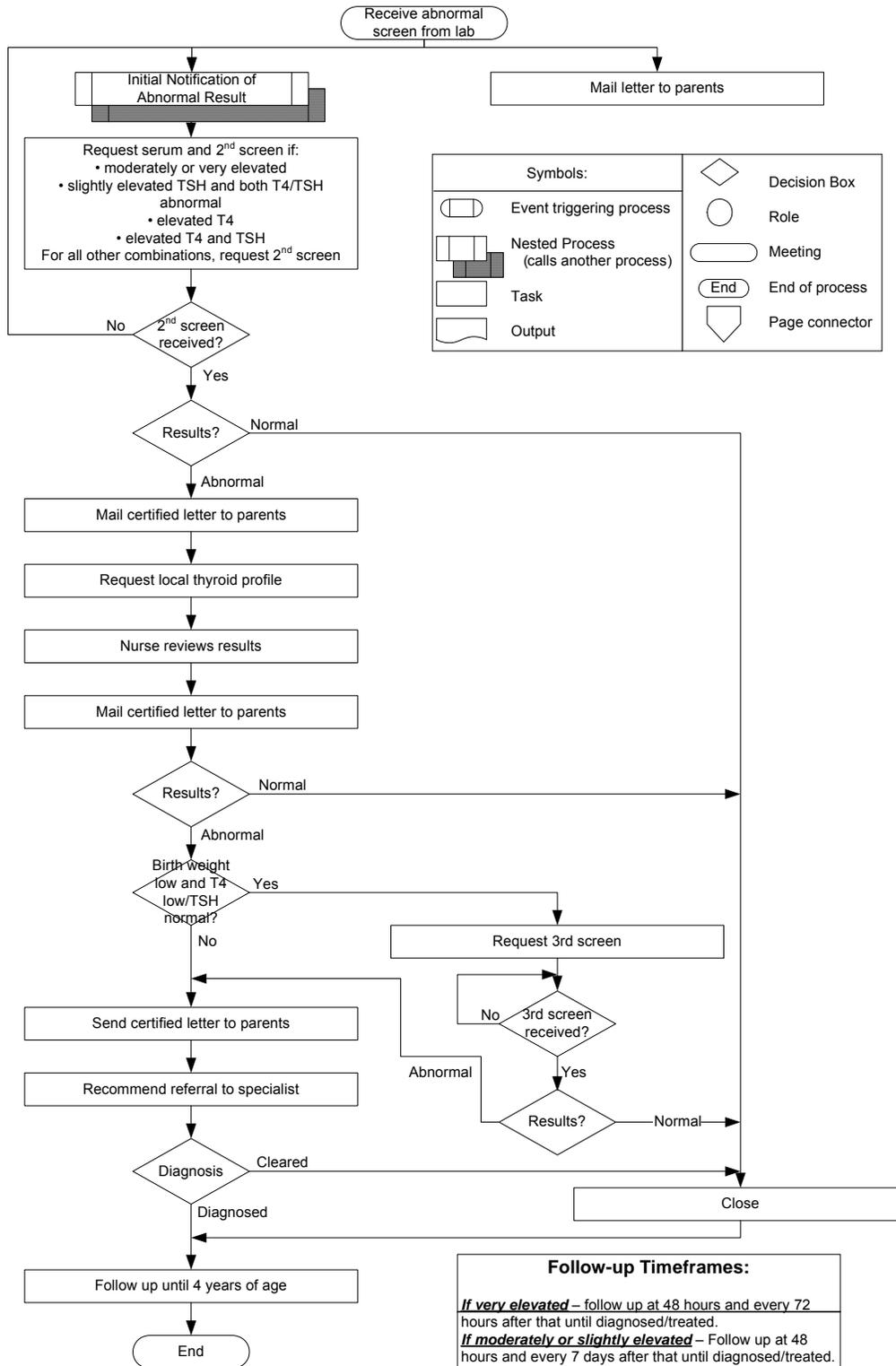
Symbols:	
	Event triggering the process
	Nested Process (calls another process)
	Task
	Output
	Decision Box
	Role
	Meeting
	End of process
	Page connector

Follow-up Timeframes:

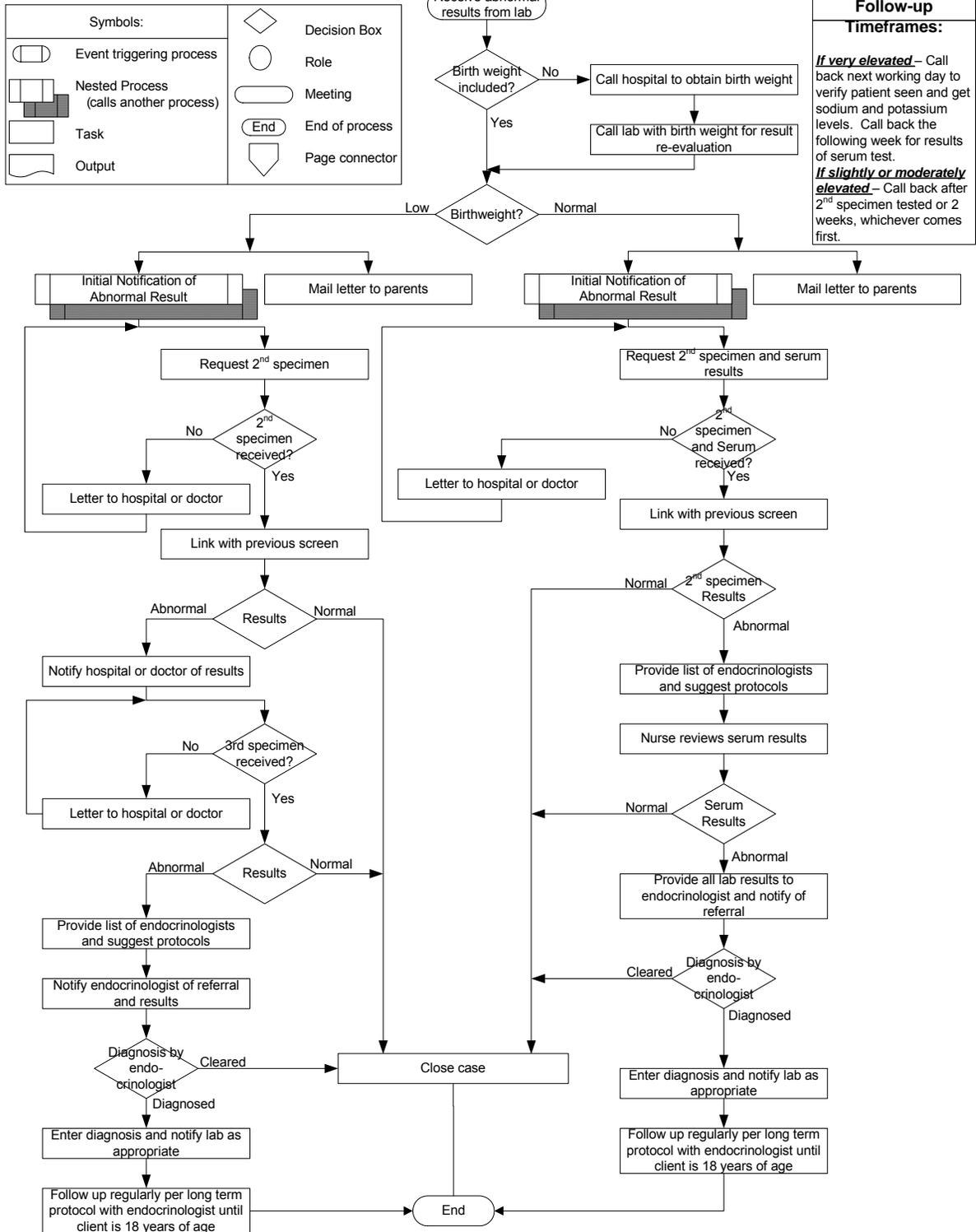
For moderately and very elevated – Next working day follow-up
For slightly elevated – Follow-up of no second screen in 2 weeks.



Hypothyroidism



Congenital Adrenal Hyperplasia (CAH)



14 2007 Newborn Screening Statistics

Below are the newborn screening statistics reported for 2007 (Source: DSHS NBS LIMS).

General Specimen Statistics	
Number of Specimens Reported	791,784
Number of Unsatisfactory Specimens (Not Tested)	14,324
Number of Presumptive Positives	16,596

Below are the 2007 newborn screening disorders and reported cases. Numbers indicated in parenthesis are disorders reported but are not on the core panel for screening.

Biotinidase Deficiency (BIOT) Cases	
Profound BIOT	4
Partial BIOT	8

Congenital Adrenal Hyperplasia (CAH) Cases	
Salt Wasting CAH	14
Simple Virilizing CAH	2
Other	(10)

Congenital Hypothyroidism (CH) Cases	
Primary CH	158

Galactosemia(GAL) Cases	
Classical Gal	5
Other	(71)

Homocystinuria (HCY) Cases	
Homocystinuria	2

Maple Syrup Urine Disease (MSUD) Cases	
MSUD	1

Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency Cases	
MCAD	16

PKU Cases	
Classical PKU	12
Variants	(13)

Sickle Cell Disease (SCD) Cases	
SS	90
SC	40
S/β Thalassemia	10
SD	(1)
Other (Non-Sickling)	(37)

Tyrosinemia (TYR) Cases	
TYR	0
Other	(8)

Other Fatty Acid Oxidation Disorders Cases	
LCHAD	2
VLCAD	6
Other	(1)

Organic Acid Disorders Cases	
3 MCC	7
GA-1	7
IVA	1
MMA	3
MCD	1
PROP	1
Other	(2)

Urea Cycle Disorders (UCD) Cases	
ASA	1
CIT	1