

# ***Division of Consolidated Laboratory Services***



***Building a Framework for Success through Collaboration, Communication, and Commitment***

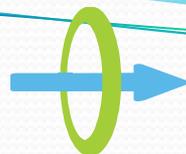
# Who are we and what do we do?

- *The Division of Consolidated Laboratory Services (DCLS) is a Division of the Virginia Department of General Services (DGS) under the Secretary of Administration.*
- *Formed in 1972 and was the first consolidated lab in the nation. DCLS provides laboratory services for a wide variety of local, state and federal law enforcement, emergency response, health and environmental protection programs.*
- *Customers/partners include VDH, DEQ, VDACS, DOC, VDEM, CDC, EPA, FBI, FDA, DHS, police, fire/rescue/HAZMAT, hospitals, physicians and waterworks*
- *Employs 220 full-time scientists and laboratory support staff.*
- *Performs over 6 million tests a year to help ensure the safety and health of Virginia's citizens and the environment.*
- *Accredits environmental laboratories (**over 400 facilities**) throughout the Commonwealth to ensure compliance with Virginia Regulations 1 VAC 30, Chapters 45 and 46 and VA Safe Drinking Water act.*

# Who are we and what do we do?

- One of 4 state public health laboratories initially selected and funded by the federal government as a regional site to test human specimens for evidence of exposure to biological and chemical agents (i.e. anthrax).
- On March 28, 2008 was the first state public health lab in the nation to send Influenza test results to CDC using HL7 2.3.1 and nationally recognized data standards and vocabulary such as LOINC, SNOMED and UCUM.
- Performs testing on every infant born within the Commonwealth for twenty-eight metabolic and genetic disorders. (*Approximately 120,000 infant samples per year*)
- Key player in the Commonwealth's Emergency Preparedness Plan

## PHL Functions



## Health Impacts

- *Performs all-hazards testing and is regional lab backup to CDC*
- *Performs testing to detect emerging public health threats*
- *Provides State-to-State Mutual Assistance*
- *Performs statewide newborn screening testing*
- *Performs environmental analysis*

- *Facilitates the Commonwealth's Emergency preparedness response*
- *Enhances population health management capabilities, provides key data for surveillance, outbreak management, and treatment recommendations*
- *Enhances the Commonwealth's surge and pandemic response capabilities*
- *Reduces infant mortality/ improves child health*
- *Ensures safe drinking water, soil, and air we breathe....*



# The Newborn Screening Story

How One Simple Test Changed  
Lives, Science, and Health in America



## Background on Newborn Screening Testing

- Almost 50 years ago, Robert Guthrie devised a screening test for Phenylketonuria (PKU) using a dried blood spot collected on a filter paper card.
- Nationally over 164 million infants have been screened for various genetic and metabolic disorders since the program started.
- Today approximately 97% of all babies born in the US are initially screened by a State public health lab.
- On average each year, 1 in every 800 babies born in the United States each year, are identified with a condition detected through the newborn screening program.

# Screening begins in the States

**1963**

Massachusetts  
Oregon  
Delaware\*  
Vermont\*



**1964**

Louisiana  
New Jersey  
New York

**1965**

Alabama  
Alaska  
California  
Colorado  
Connecticut  
Florida  
Hawaii  
Idaho  
Illinois  
Indiana  
Iowa  
Kansas  
Maine  
Maryland

Michigan  
Minnesota  
Missouri  
Montana  
New Hampshire  
Ohio  
Oklahoma  
Pennsylvania  
Rhode Island  
South Carolina  
Utah  
West Virginia  
Wisconsin

**1966**

Georgia  
Kentucky  
New Mexico  
Texas  
Virginia

**1967**

Arkansas  
Nebraska  
Nevada  
North Dakota  
Washington

**1968**

Tennessee



**1966**

**1973**

South Dakota

**1979**

Arizona

**1980**

District of Columbia

**1983**

North Carolina  
Wyoming

**1985**

Mississippi



\* Sources vary on dates screening was established.  
Therrell, B., & Adams, J. (2007). Newborn screening in North America.

# NBS – National Outcomes.....

<b>Disorder</b>	<b>Prevalence</b>	
Primary Congenital Hypothyroidism (CAH)	<b>1 in 3,000</b>	
	<i>Untreated</i>	<i>Serious intellectual, development and physical disabilities and slow growth within first month following birth</i>
	<i>Treated</i>	<i>Normal development with daily dose of medicine</i>
<b>Disorder</b>	<b>Prevalence</b>	
Cystic Fibrosis (CF)	<b>1 in 3,700</b>	
	<i>Untreated</i>	<i>Life long health problems, lung damage, and possibly early death</i>
	<i>Treated</i>	<i>Treatment, medications, therapies leading to longer and healthier lives</i>
<b>Disorder</b>	<b>Prevalence</b>	
Galactosemia (GAL)	<b>1 in 53,000</b>	
	<i>Untreated</i>	<i>Serious intellectual disabilities, seizures, sepsis, shock, or death possible within 4 weeks of birth</i>
	<i>Treated</i>	<i>Normal health and development with a special diet</i>
<b>Disorder</b>	<b>Prevalence</b>	
Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)	<b>1 in 15,000</b>	
	<i>Untreated</i>	<i>Metabolic crisis possibly leading to seizure, coma, and death within 3 months of birth</i>
	<i>Treated</i>	<i>Normal health and development with a special diet and monitoring</i>
<b>Disorder</b>	<b>Prevalence</b>	
Severe Combined Immunodeficiency (SCID)	<b>1 in 75,000</b>	
	<i>Untreated</i>	<i>Death within 1 to 2 years following birth</i>
	<i>Treated</i>	<i>With bone marrow transplant within 3 months of birth, normal healthy life</i>
<b>Disorder</b>	<b>Prevalence</b>	
Sickle Cell Anemia (SCA)	<b>1 in 3,700</b>	
	<i>Untreated</i>	<i>Pain, infections, possible death within 1<sup>st</sup> year following birth</i>
	<i>Treated</i>	<i>Antibiotic and other therapies lead to healthier lives with fewer symptoms</i>

A smiling baby is the central focus, surrounded by a pile of wooden alphabet blocks. The blocks are scattered around the baby, with some showing letters like 'C', 'M', 'Z', 'Q', 'G', 'I', 'L', 'K', 'R', 'S', 'T', 'U', 'V', 'W', 'X', 'Y', and 'Z'. The background is white with a blue wave-like border at the top.

*Newborn Screening:*  
Saves or Improves  
the Lives of Over  
**12,000**  
Babies a Year!

**PARENT  
EDUCATION**

Obstetrician  
explains newborn  
screening process to  
expectant parents.

**HOSPITAL  
SCREENING**

Hospital nurse tests  
baby's hearing and  
heart, and collects blood  
from baby's heel.

**LAB  
SCREENING**

State public health lab  
tests baby's blood  
for at least  
29 genetic conditions.

**NORMAL  
RESULTS**

Pediatrician  
reviews test  
results with  
parents at baby's  
first wellness visit.

**POSITIVE  
RESULTS**

Health Department  
staff calls  
pediatrician/parents to  
request re-testing baby.  
Medical specialists  
perform tests and  
make diagnosis.

**FOLLOW-UP**

Medical specialists and  
pediatrician develop a  
treatment plan and  
guide parents in caring  
for baby.

# ***Virginia's Newborn Screening Program***



***Healthy babies lead to healthy citizens***

# Virginia's Newborn Screening Program

- Mission - To prevent mental retardation, permanent disability, or death through early identification and treatment of infants who are affected by certain heritable disorders and genetic disease.
- Overview – The Newborn Screening Program is a coordinated and comprehensive program consisting of education, newborn screening, follow-up, diagnostic confirmation, medical and dietary management, and treatment and referral.
- Partners - The Department of General Services, Division of Consolidated Laboratory Services along with the Genetics and Newborn Screening Division of Family Health Services, at the Virginia Department of Health, partner to co-administer the Commonwealth's Newborn Screening program.

# Virginia's Newborn Screening Program

- **Guidance** – Virginia's newborn screening panel is based on the nationally recommended uniform screening panel, with endorsement by the Virginia Genetics Advisory Committee and approval of the VA Board of Health - and the Governor.
- **Authority** - The Code of Virginia 32.1-65 mandates that every child born in Virginia be screened.
- **Current State** - DCLS currently screens for 28 genetic and metabolic conditions and performs approximately 120,000 dried-blood spot screens per year. On average 12,000 screens are sent to VDH follow-up and 4,000 babies are diagnosed with either a disorder or as a carrier, and receive treatment, long-term care and/or counseling.

# Newborn Screening Program Overview

- Goal is to test every newborn in Virginia within a few days of birth unless a parent or guardian objects on the grounds that the test conflicts with his or her religious practices.
- DCLS performs newborn testing from the dried blood spot card.
- VDH newborn screening staff coordinates follow-up activities until the infant is diagnosed, screened negative, or reaches 6 months of age.
- Diagnosed babies with certain heritable disorders or genetic diseases are referred to the Care Connection for Children network for coordination services.



## *Background on Virginia's Newborn Screening Program*

<b>Year</b>	<b>Disorders Tested</b>	<b>Disorder(s) Added / Condition(s) Screened</b>
1966	1	PKU
1984	5	MSUD, HCU, Hypothyroidism, Galactosemia
1986	6	Biotinidase Deficiency
1989	7	Hemoglobinopathies (Sickle Cell Anemia, etc)
2002	8	Congenital Adrenal Hyperplasia (CAH)
2004	9	Med. Chain CoA Dehydrogenase Def. (MCAD)
2006	28	Cystic Fibrosis-18 Amino, Fatty Acid Oxidation / Organic Acid Disorders
2015	29*	Severe Combined Immunodeficiency (SCID)

*\*Pending regulatory approval*

# Newborn Screening Outcomes

- Education* → *Improved prenatal care, early intervention options, and genetic and dietary counseling.*
- Screening* → *Rapid identification and prevention of disorders that can result in delayed growth and development, reoccurring infections, blindness, mental retardation, and death.*
- Follow-up* → *Consultation and coordination of treatment and continual follow-up – transitioning into adult healthcare.*
- Diagnosis* → *Development of care management plans, dietary care management, referrals to specialist/Care Connection, and targeted treatment.*

# Strengths of the Virginia NBS Program

- Dedication to saving babies!
- Consistent with the uniform screening panel recommended by the HHS Secretary's Advisory Committee on Heritable Disorders in Newborns and Children
- Strong partnership between laboratory and follow-up teams
- Supported by pediatric specialists in all areas (Pulmonology, Metabolics, Endocrinology, Hematology, Geneticists, Neonatologists, etc)
- Supported by an active Genetics Advisory Committee and NBS Subcommittee
- Rapid screening (24 hour turn around for normal results)
- Critical results are reported Monday through Saturday to VDH
- Statewide courier service – next day sample delivery
- Availability of educational products for parents and healthcare providers
- Shared information management system – used by laboratory and follow-up program staff

# Consent and Confidentiality of Data

- All newborns are screened unless their parent or guardian objects on religious grounds.
- A statement of written objection by the parent or guardian is included in the child's medical record.
- The Virginia Board of Health, the State Health Commissioner, and the Commissioner's agents have access to any newborn screening records.
- With approval from the Lab Director, NBS de-identifiable data is released for the purpose of research and statistical analysis.
- Request for NBS results for a specific child, requires written consent from the parent or guardian before data is shared.

# How is Newborn Screening Funded?

- Since 1992, the Virginia Newborn Screening program has operated as a fee-for service program.
- The fee for Newborn Screening services is \$53.00\* per child and is paid by the birthing facility or provider through the purchase of NBS collection kits.
- Virginia's NBS Fee covers –
  - Kit components, assembly and distribution
  - Laboratory services
  - Follow-up services

\* The NBS fee will increase to \$78.00 in 2014

# Storage and Use of Dried Blood Spots

- Upon completion of testing, there are some dried blood spots left on the filter paper card.
- DCLS holds residual blood spots for normal screens 6 months and abnormal screens 10 years. Confirmatory testing may also be performed after the initial screen.
- DCLS policy prohibits the use of residual blood spot samples for any purpose other than newborn screening testing.
- Samples are never released without written parental consent and that consent form must be notarized.

# NBS Data Retention Requirements

- *Records which contain the observations and interpretations at the time of testing and include test results and interpretations, observations of temperatures, QC logs, maintenance logs, etc. are maintained as follows:*
  - *All documentation and/or gels for Adult Sickle Cell: 3 years.*
  - *Records regarding unsatisfactory samples: 10 years*
  - *Records regarding Abnormal samples: 10 years*
  - *Records regarding Normal samples: 10 years*
  - *Proficiency Results/Data: 10 years*
  - *Quality Control Records: 10 years*
- *All electronic records are maintained for 23 years.*

# VA's NBS Diagnosed Cases - In 2012

- Hemoglobin disorders – 93 (Sickle Cell, Sickle C Disease, Sickle Beta Thal, etc)
- Primary Congenital Hypothyroidism – 28
- Cystic Fibrosis – 15
- MCAD (Medium Chain CoA Dehydrogenase Deficiency) – 8
- Congenital Adrenal Hyperplasia – 3
- Galactosemia – 2
- Phenylketonuria – 2
- Biotinidase - 1
- Propionic Acidemia – 1
- Maple Syrup Urine Disease – 1
- 3-MCC (3-methylcrotonyl-CoA Carboxylase Deficiency) – 1

*\* Thousands of infants identified as carriers of Sickle Cell and Cystic Fibrosis\**

# *What's on the horizon for the Virginia's NBS Program?*

- *Severe Combined Immunodeficiency (SCID) is in the final phase of adoption for inclusion in Virginia's NBS screening panel of conditions.*
  - *On target for implementation - January 2015*
- *Pompe Disease, a lysosomal storage disorder, has recently been recommended for inclusion in the nationally recommended uniform screening panel.*
  - *Under review by the U.S. Secretary of Health and Human Services*

# *Overview Laboratory Information Management Systems and Data Integration Engine*



# *Laboratory Information Management System (LIMS)*

- *LIMS Vendor – StarLIMS (Implemented in over 29 PHL's in U.S. for clinical reporting and 3 PHL's for Newborn Screening.)*
- *Application – “Custom COTS” – Built using a combination of coding and configuration.*
- *Development Language – Proprietary to the vendor*
- *Development Tool - Proprietary to the vendor*
- *Platform – Uses Web-based architecture*
- *Database - Oracle 11G – RAC configured for high availability*
- *Barcode Technology – Font Code 3 of 9 (lab standard) and 128*

# *Laboratory Information Management System (LIMS)*

- *Heavy Integration with Laboratory Instrumentation – Bi-directional interface between LIMS and instrument systems such as PerkinElmer’s Specimen Gate and Agilent’s OpenLab systems.*
- *Business Continuity - Onsite data center at DCLS that allows lab to run as island when connectivity to COV CESC / outside network is unavailable.*
- *Standards Adoption*
  - *Current NBS LIMS – None*
  - *New NBS LIMS - LOINC, SNOMED-CT, UCUM, OLIM, UNIPROT, Enzyme and local codes*

# *Data Integration Engine*

- *Vendor – Orion Health Systems*
- *Application – Rhapsody Data Integration Engine v5.4 and Symphonia Mapping tool*
- *Development Tool - Proprietary to the vendor*
- *Development Language – Proprietary to the vendor*
- *Platform - Web-based architecture*
- *HL7 Versions currently supported – v2.3.1 and v2.5.1*
  - *Influenza Surveillance (PHLIP) to CDC – v2.3.1 ORU R01*
  - *All-Hazard (LIMSi) reporting to CDC – v2.5.1 OUL R22*
  - *Electronic Lab Reporting (ELR) to VDH – v2.5.1 ORU R01*

# ORU R01 vs. OUL R22 for PHL Lab Reporting

## ORU R01 - Observation Result Unsolicited

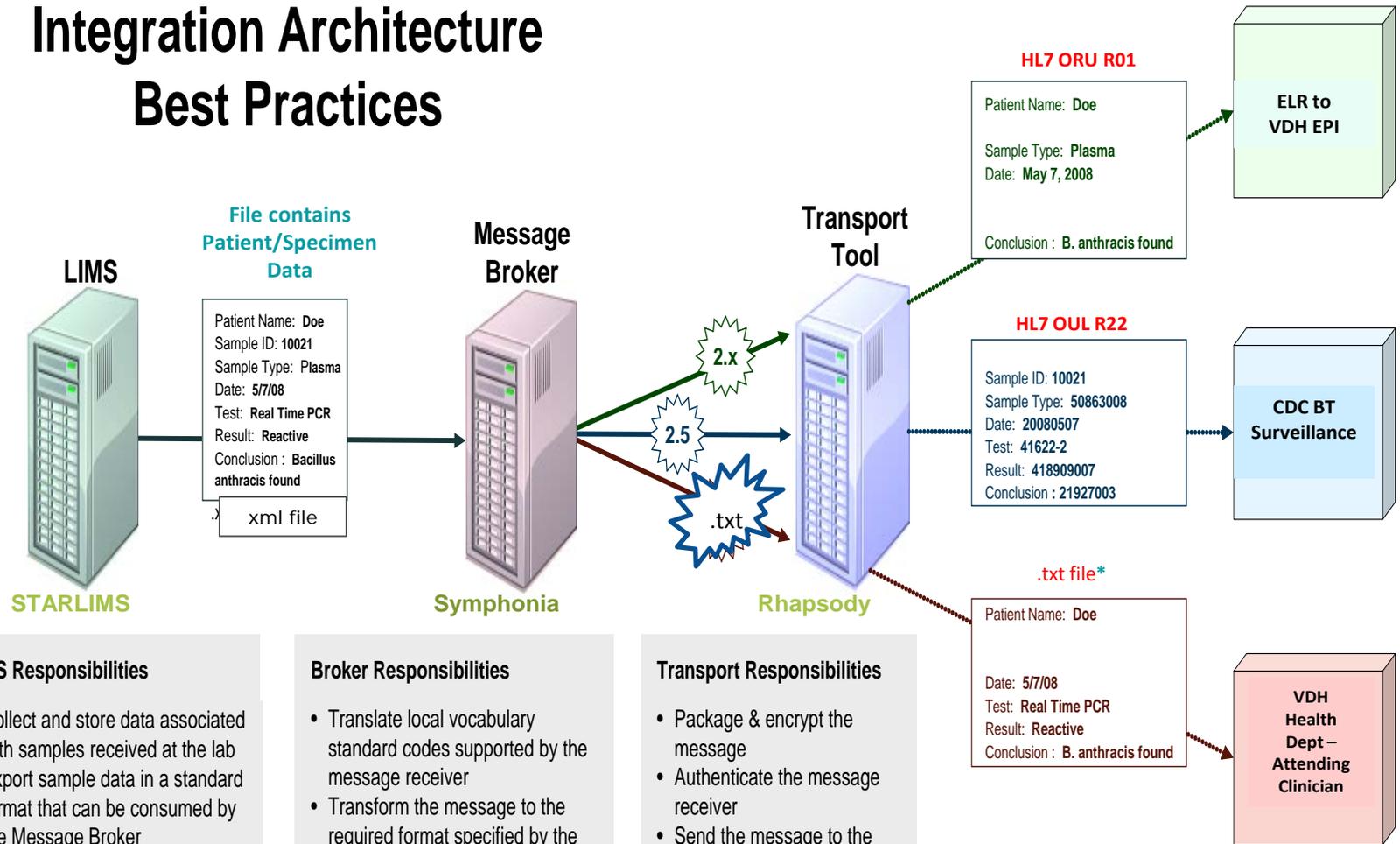
- ✓ Used to convey any type of results, including Lab, Radiology, and Anatomic Pathology.
- ✓ Provides flexibility as results may or may not be related to a specimen.
- ✓ Facilitates reporting of patient-oriented test results to VDH for reportable conditions or to VDH clinics, hospitals and providers for patient results.

## OUL R22 – Unsolicited Laboratory Observation

- ✓ Used for reporting of laboratory results for a specimen.
- ✓ Facilitates reporting of lab results between labs (e.g. isolates from lab to reference lab, or hospital labs to labs or PHL to CDC).

# Current Architecture for Clinical Messaging

## Integration Architecture Best Practices



### LIMS Responsibilities

- Collect and store data associated with samples received at the lab
- Export sample data in a standard format that can be consumed by the Message Broker

### Broker Responsibilities

- Translate local vocabulary standard codes supported by the message receiver
- Transform the message to the required format specified by the message receiver

### Transport Responsibilities

- Package & encrypt the message
- Authenticate the message receiver
- Send the message to the designated receiver

\*Data feed will be converted to ORU R01 by January 2014

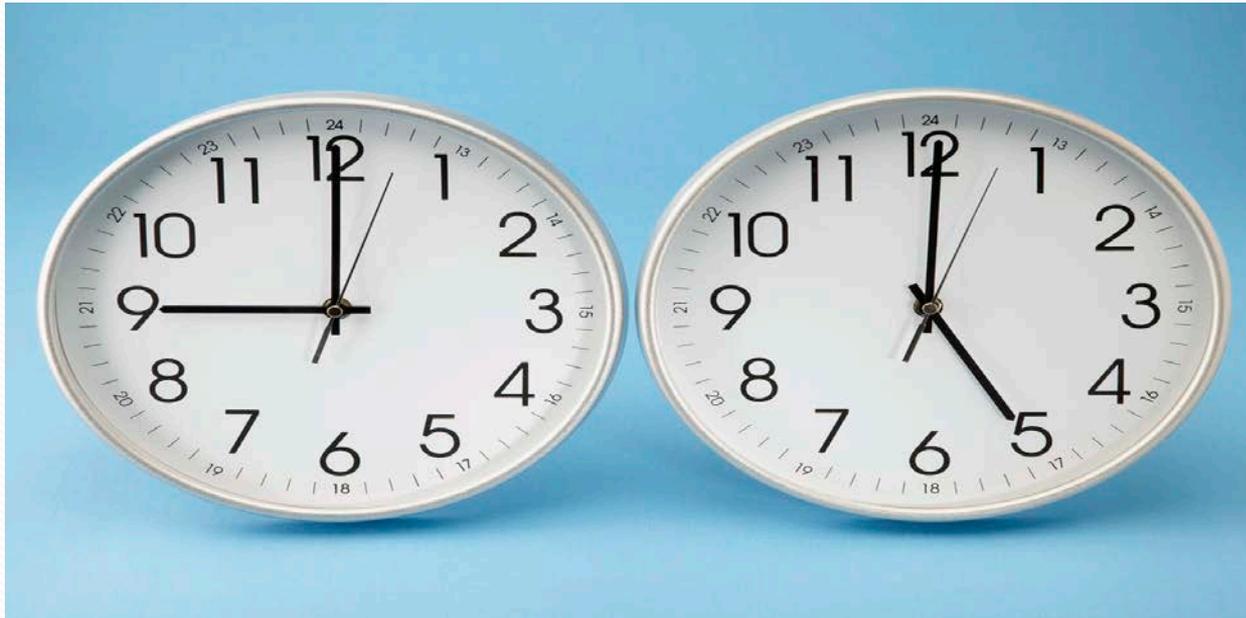
# *NBS Data Collection Process*

*NBS Dried Blood Spot Card*

*NBS LIMS*

*Birth Registry*

*Hearing Screening*



# Newborn Screening Timeline

Dried Blood Spot collected by birthing facility and lab order submitted to DCLS on filter paper card.



All results sent to the birthing facility / Doctor



Abnormal/  
critical  
results to VDH  
Follow-up

Facility Birth Registrar enters information into VDH VVESTS



Facility staff enters hearing screen results into VVESTS



24 - 36 hours  
following birth

24 -48 hours  
following sample  
receipt

Up to 7  
days later

Up to 14  
days later

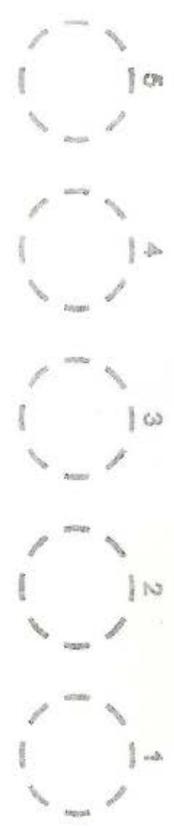


# Dried Blood Spot Card

*Birthing Facility completes within 24-36 hrs of Birth*

NBS Device ID

SN 20368577		SN 20368577		1111720229		FILTER COPY	
BABY'S NAME: LAST		FIRST	MEDICAL RECORD NUMBER	BIRTH DATE	BIRTH TIME (MILITARY)	SEX ( ) MALE ( ) FEMALE	
BIRTH WEIGHT	ETHNICITY/RACE	BIRTH		STATUS		4 TRANSFUSED	
GRAMS / LBS	1 ( ) BLK. 4 ( ) ASIAN 2 ( ) WHT. 5 ( ) AMER. INDIAN 3 ( ) HISP. 6 ( ) OTHER	1 ( ) SINGLE 2 ( ) TWIN A 3 ( ) TWIN B	4 ( ) TRIPLET A 5 ( ) TRIPLET B 6 ( ) TRIPLET C	7 ( ) OTHER	1 ( ) NORMAL 2 ( ) SICK	3 ( ) PREMATURE Check (✓) only if < 37 Weeks	( ) Y ( ) N DATE
FEEDING TYPE:		DATE OF COLLECTION	TIME OF COLLECTION (MILITARY)	AGE AT COLLECTION	SPECIMEN TYPE		
1 ( ) Breast Only	2 ( ) Cow's Formula	3 ( ) TPN		1 ( ) ≤ 24 HRS.	1 ( ) INITIAL SPEC.		
4 ( ) Soy Formula	5 ( ) Breast + Cow Formula	8 ( ) Other		2 ( ) > 24 HRS.	2 ( ) REPEAT SPEC.		
6 ( ) Breast + Soy Formula	7 ( ) Breast + TPN						
MOTHER'S NAME: LAST		FIRST	AGE	MOTHER'S SSN	MOTHER'S TELEPHONE NUMBER		
MOTHER'S ADDRESS		CITY	STATE	ZIP CODE (+4 CODE)	COUNTY OF RESIDENCE		
PLACE OF BIRTH	BIRTH HOSPITAL CODE	BIRTH HOSPITAL NAME	BIRTH HOSPITAL ADDRESS	CITY	STATE	ZIP CODE (+4 CODE)	
1 ( ) HOSPITAL BIRTH 2 ( ) HOME BIRTH 3 ( ) OTHER							
DOCTOR CODE NUMBER	TELEPHONE NUMBER	(SUBMITTER) RESULT TO CODE	TELEPHONE NUMBER	FORM COMPLETED BY		FOR LAB USE:	
						UNSAT CODE	
BABY'S PRIMARY CARE PHYSICIAN		(SUBMITTER) RESULT TO NAME		SPECIMEN COLLECTED BY		DATE / INT.	
DOCTOR'S ADDRESS		SUBMITTER'S ADDRESS		Commonwealth of Virginia Department of General Services Newborn Screening Laboratory 600 N. 5th St. Richmond, VA 23219 Telephone: (866) 378-7730 DGS-22-174 (Rev. 09/08)			
CITY		CITY		Use by 2012-11			
STATE		STATE					
ZIP CODE (+4 CODE)		ZIP CODE (+4 CODE)					



# Newborn LIMS

## DCLS Data Entry completes within 24 -48 hrs of Sample Receipt

VA NBS LIMS v3.8.0 STARSAT (Data base:NBSU\_DATA, Dictionary:NBSU\_DICT\_VTYSON, User:VTYSON, WorkStation:metamp14, IP Address: 159.1...)

File Submitter Name Submitter ID Add Doctor,Submitte Done

Demographic Data Entry

Close Submitter Name Submitter ID Add Doctor,Submitter,Hospital Done

Control ID  NBS Device ID  Birth time

Baby last name  Baby first name  Medical ID  Birth date  Birth Time Type  Sex

Birth weight  Race ID  Multi bith  Baby status  Transfused

Race other  Multi bith other  Premature  Transfusion date

Transfusion Type

First feed date  Feeding ID  Collection date  Collection time  Coll. age

Feed other  Collection Date Type  Collection time type  Specimen type  Transfused Comments

Mother last name  Mother first name  Mother age  Mother SSN  Mother phone

Mother address  Mother city  Mother state  Mother zip  Mother county

Birth Place ID  Hospital code  Hospital name

Birth place other

Doctor ID  Doctor phone  Submitter ID  Submitter phone  Unsat code

Doctor name  Submitter name  Completed By

Doctor address  Submitter address  Collected by

Doctor city  Submitter city

Doctor state  Submitter state

Doctor zip  Submitter zip

Comments

Done

12:32:15 PM IN3 CAPS NBSU SCROLL

Start Citrix Pro... Inbox - ... Capacity... Microsoft... RE: - M... TSUms - ... nbsu - V... VA NBS... 12:32 PM

# VDH - Birth Registry System

*Facility Birth Registrar completes within 7 days of Birth*

## Mother Update

Next >>

- Mother's Additional Races

First Name:	<input type="text" value="TESTPAT"/> **	Middle Name:	<input type="text" value="TRIGGER TEST"/>
Last Name:	<input type="text" value="PATLAST"/> **	Maiden Name:	<input type="text" value="SMITH"/> **
Suffix:	<input type="text" value="IX"/>	Date Of Birth:	<input type="text" value="12/01/1999"/> (mm/dd/yyyy)
Pin Type:	<input type="text" value="ITIN"/> **	Pin:	<input type="text" value="964-65-5455"/> *
Hispanic Origin:	<input type="text" value="UNKNOWN"/> **	Other Hispanic Origin:	<input type="text"/> *
Birth State:	<input type="text" value="South Carolina"/> *	Birth Country(other than US):	<input type="text"/> *
Race:	<input type="text" value="ASIAN INDIAN"/> **	Specify Race 2 (if any):	<input type="text"/>
Specify Race 1 (if any):	<input type="text"/>	Date Created:	<input type="text" value="05/09/2009 15:11:22"/>
Created By:	<input type="text" value="FBC"/>		

**\*\*Sept. 2013** – VDH will add NBS Device ID (from the Dried Blood Spot card) as a new required field in Birth Registry System – First step towards linking NBS screening to Birth Registry.

# VDH - Birth Registry System

## Birthing Facility completes within 14 days of Birth

WESTS 2.7.6 (User: EBC) - Windows Internet Explorer provided by VA IT Infrastructure Partnership

https://viswebdev.vdh.virginia.gov/ebcdev/client\_view\_adv\_child\_info\_view?preg\_id\_in=&cev\_id\_in=111555&p\_mod=VISITS&res\_id\_in=2805127&child\_id\_in=2805168&child\_de

Child's Name : THOMAS, TEST1  
Date of Birth : 11/10/2011  
Child Id : 2805168

Pending Reg. Entry  
None

Report

### Initial Hearing Screening

Screening Date:  \*\* (mm/dd/yyyy)      Screening Setting:  \*\*

Screening Test:  \*\*      Person Administering Screening:  \*\*

Birth Setting:       Date Received:  \* (mm/dd/yyyy)

Right Ear Results:  \*\*

Left Ear Results:  \*\*

Reported By:  \*\*

Next Appointment Date:  \* (mm/dd/yyyy)      Next Appointment Facility:  \*

#### Risk Indicators

S No.	Hearing Risk Indicators	Comments
1.	<input type="checkbox"/> Family history of permanent childhood hearing loss.	<input type="checkbox"/> Mother of child <input type="checkbox"/> Father of child <input type="checkbox"/> Brother of child <input type="checkbox"/> Sister of child <input type="checkbox"/> Grandfather of child <input type="checkbox"/> Grandmother of child <input type="checkbox"/> Aunt of child <input type="checkbox"/> Uncle of child <input type="checkbox"/> 1st cousin of child <input type="checkbox"/> More than one relative of the same parent
2.	<input type="checkbox"/> Stigmata or other findings associated with a syndrome known to include a sensorineural or conductive hearing loss or Eustachian tube dysfunction.	<input type="checkbox"/> Branchio-oto-renal (BOR) <input type="checkbox"/> CHARGE association <input type="checkbox"/> Goldenhar (oculo-auriculo-vertebral or OAV) <input type="checkbox"/> Noonan <input type="checkbox"/> Pierre Robin <input type="checkbox"/> Rubenstein-Taybi <input type="checkbox"/> Stickler <input type="checkbox"/> Trisomy 21, 18, 13, 9, or 8 <input type="checkbox"/> Williams

Save   Undo

- Infant Search
- Pending Children
- Infant**
- Child
- Mother
- Father
- Contact Information
- Special Circumstances
- Provider
- Status Details
- Infant Summary Data

- Screenings**
- Discharge Summary
- Initial Hearing Screening
- Risk Indicators
- VaCARES / Birth Defects
- Part-C EI Referrals
- Activities
- Referrals and Outcomes (Hearing)
- Referrals and Outcomes (VaCARES)
- Summary

- Reports/Letters**
- Hearing Letters
- VDH Hearing Reports

# Newborn Screening Data Collection

	NBS LIMS	VDH - Birth Registry	VDH - Hearing Screening
Type of Interface	Manual - Data collected from Filter Paper Card	Web portal – Birth Registry System	Web portal – Hearing module within Birth Registry System
Data Entered By	Data Entry Staff	Birth Registrar	Birth Registrar and/or Nursery staff
Data Collected	52 pieces of metadata related to sample, collection data/time, infant, mother, submitter, doctor, and program specific data such as “Feeding Type.”	47 pieces of data collected by lab is also collected by Birth Registry System. System also collects other information as recommended by VDH vital records and National Centers for Health Statistics (NCHS).	All required metadata related to baby and mother is in Birth Registry System. Hearing collects and enters 10 additional pieces of data.
Primary Search Key	Sample/Mother	Mother	Mother/Child
Time Collected	24-36 hours after sample receipt	Up to 7 days after birth	Up to 14 days after birth

# *Opportunity for Business Transformation*

## *Proposal*

*Through collaborative efforts, DCLS, VDH, and hospital partners will work to adopt an HL7 standards-based message and the associated coded terminologies that can be used by the birthing facilities to send at least the minimum required data set to DCLS for NBS screening blood spot testing and to VDH for birth registration and hearing screening, within the required/specified processing times.*

# Proposed High-Level Goals

- Agree on a minimum data set that is required for the timely processing of NBS screening, Birth Registry, and Hearing Screening data.
- Adopt a standards-based message that can be leveraged by the birthing facilities to send data NBS, birth registry, and hearing screening data to Lab and/or VDH.
- Define the required reporting triggers to ensure data is sent by the birthing facilities and received by Lab and/or VDH within the established timeframes to enable dried blood spot screening, birth registration, and hearing screening.
- Comply with the Code of Virginia, HITSAC, other state and federal regulatory requirements specific to NBS screening, birth registration, and hearing screening.
- Comply with all state and federal consent, privacy and security requirements.
- Create Child Health Record and leverage MPI to link NBS screening results, birth registration, vital records and hearing data.

*NBS Messaging  
Getting the Stork to deliver....*



# Background on NBS Message Development

- In 2010, the Public Health Informatics Institute entered into a cooperative agreement with the Health Resources and Services Administration (HRSA) and the U. S. National Library of Medicine.
- Established the need to develop a Technical Implementation Guide to standardize the reporting of NBS laboratory results/orders.
- Workgroup members selected HL7 version 2.5.1 as it is universally adopted in large practices, laboratories and birthing facilities.
- Acknowledged that the process for NBS lab order submission was labor intensive and required a high level of manual data entry.
- Changes were needed to realize operational efficiencies, improve data integrity, enhance matching of NBS data to State vital records birth registry, create the infant/child health record, and to promote interoperability between PHL labs and the birthing facility and provider community so NBS data could be securely exchanged in real-time.

# *National NBS Messaging Workgroup*

*The workgroup members included representatives from public health laboratories, hospitals, public health agencies and the U.S. National Library of Medicine:*

- *Iowa State Lab, University of Iowa Hygienic Laboratory*
- *New York State Department of Health*
- *Northside and Riverside Hospitals, Atlanta GA*
- *National Library of Medicine*
  - ✓ *Dr. Clem McDonald*
  - ✓ *Dr. Alan Zuckerman*
- *Virginia's Division of Consolidated Laboratory Services*
  - ✓ *Willie Andrews*
  - ✓ *Vickie Tyson*
- *HLN Consulting, LLC*
- *Public Health Informatics Institute (PHII)*

# Assumptions Made

- Electronic health record and laboratory systems are in place that allow for the electronic messaging of NBS lab orders and results.
- The data required for NBS data exchange is available in the hospital/EHR/provider system.
- The original order, paper or electronic message contains sufficient information for the laboratory to process the lab order and construct the lab result message.
- Messaging partners agree to adopt the recommended standards, terminologies, consent, privacy and security model for NBS data exchange as set forth by federal, state and local jurisdictions.

# Messaging Standards for NBS Reporting



U.S. National Library of Medicine

## Newborn Screening Coding and Terminology Guide

Data Standards for Electronic Reporting

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### Constructing Newborn Screening HL7 Messages

To help promote efficient electronic exchange of standard newborn screening data, the Lister Hill Center at the U.S. National Library of Medicine (NLM) in cooperation with the Newborn Screening Community and HITSP Population Perspective Technical Committee has developed draft guidance about the use of LOINC and SNOMED CT codes to report newborn screening test results in standard Health Level 7 (HL7) version 2.x message format.

### Newborn Dried Blood Spot (NDBS) Screening HL7 Implementation Guides for Laboratory Orders and Results

Developed by the Public Health Informatics Institute under a cooperative agreement with the Health Resources and Services Administration (HRSA), these Newborn Dried Blood Spot (NDBS) Screening implementation guides provide a recommended approach for an [HL7 Version 2.5.1 OML^O21 message to transmit NDBS laboratory orders](#) from the birthing center/hospital to the public health laboratory, and for NDBS laboratories to use an [HL7 Version 2.5.1 ORU^R01 message for sending standardized NDBS laboratory results](#).

- [HL7 Version 2.5.1 OML^O21 message to transmit NDBS laboratory orders \(v 1.0.1\) Oct 12, 2011 -](http://www.phii.org/sites/default/files/resource/pdfs/PHII-NDBS-Lab-Orders-Impl-Guide-OML%5EO21-HL7-251_v1%200%201_2011-10-12_final.pdf)  
[http://www.phii.org/sites/default/files/resource/pdfs/PHII-NDBS-Lab-Orders-Impl-Guide-OML%5EO21-HL7-251\\_v1%200%201\\_2011-10-12\\_final.pdf](http://www.phii.org/sites/default/files/resource/pdfs/PHII-NDBS-Lab-Orders-Impl-Guide-OML%5EO21-HL7-251_v1%200%201_2011-10-12_final.pdf)
- [HL7 Version 2.5.1 ORU^R01 message for sending standardized NDBS laboratory results \(v 1.0.1\) Nov 1, 2011 -](http://www.phii.org/sites/default/files/resource/pdfs/PHII-NDBS-Lab-Results-Impl-Guide-ORU%5ER01-HL7-251_v1%200%201_2011-11-01_final.pdf)  
[http://www.phii.org/sites/default/files/resource/pdfs/PHII-NDBS-Lab-Results-Impl-Guide-ORU%5ER01-HL7-251\\_v1%200%201\\_2011-11-01\\_final.pdf](http://www.phii.org/sites/default/files/resource/pdfs/PHII-NDBS-Lab-Results-Impl-Guide-ORU%5ER01-HL7-251_v1%200%201_2011-11-01_final.pdf)

# Code Standards for NBS Reporting



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### Code Standards

A coding and terminology framework is essential to standardizing laboratory reporting and enabling interoperability of information exchange across Electronic Health Record (EHR) platforms.

› [LOINC](#) › [SNOMED CT](#) › [ICD-9-CM](#) › [ICD-10-CM](#) › [Enzyme Codes](#) › [OMIM](#) › [UniProt](#)

#### LOINC

[Logical Observation Identifiers Names and Codes](#) (LOINC®) is a terminology standard for identifying laboratory tests and other measurements. It specifies universal codes, names, and other attributes for laboratory results as well as clinical reports, physical exam findings, survey instruments and other observations. It was developed to enable the exchange and pooling of results from diverse sources in order to enhance clinical care, outcomes management and research. The LOINC terminology is available free of charge and was developed by the LOINC Committee and Regenstrief Institute, Inc., a non-profit medical research organization associated with Indiana University. The database is maintained by Regenstrief and updated versions are released twice a year. The LOINC web search tool is available at <http://search.loinc.org/>, or you can download the database and a free browser program, the Regenstrief LOINC Mapping Assistant (RELMA®), from <http://loinc.org/downloads>. The LOINC and RELMA Terms of Use are available at <http://loinc.org/terms-of-use>.

#### SNOMED CT

[Systematized Nomenclature of Medicine — Clinical Terms](#) (SNOMED CT®) is a comprehensive, multilingual clinical health care terminology. It is designed for use in electronic health record systems and aims to facilitate communication and interoperability in electronic health data exchange. Originally created by the College of American Pathologists (CAP) in cooperation with the National Health Service in England, SNOMED CT is now owned, maintained, and distributed by the International Health Terminology Standards Development Organisation (IHTSDO), an international not-for-profit association in Denmark, with contract assistance from the CAP. It is available free of charge in IHTSDO member countries, including the U.S., in low-income countries as defined by the World Bank, and for qualified research projects in any country. The National Library of Medicine (NLM) is the U.S. representative to the IHTSDO. Information about obtaining SNOMED CT is available through NLM's Unified Medical Language System (UMLS®) Metathesaurus at <http://www.nlm.nih.gov/research/umls/licensedcontent/snomedctfiles.html>. A free UMLS license (which includes the IHTSDO Affiliate license) is required and can be obtained at the same site.

The Newborn Screening Coding and Terminology Guide uses some codes from the [US Extension to SNOMED CT](#).

# Code Standards for NBS Reporting

## Enzyme Codes

"Enzyme Nomenclature" is a list of enzymes with their recommended names, Enzyme Commission (EC) codes, and classifications. It was created and is maintained by the Nomenclature Committee of the International Union of Biochemistry and Molecular Biology (NC-IUBMB) in consultation with the Joint Commission on Biochemical Nomenclature (JCBN). The JCBN is a collaborative effort of the International Union of Pure and Applied Chemistry (IUPAC) and the IUBMB. The entries are protected by copyright assigned to the IUBMB. The enzyme codes are freely available for use and can be found at <http://www.chem.qmul.ac.uk/iubmb/enzyme/> .

## OMIM

[Online Mendelian Inheritance in Man](#)  (OMIM®) is a comprehensive, authoritative, and timely compendium of human genes and genetic phenotypes. It was originally authored and edited by Dr. Victor McKusick, a pioneer of medical genetics and the author of "Mendelian Inheritance in Man," and is now maintained by Johns Hopkins University. The full-text, referenced overviews in OMIM contain information on over 12,000 genes and on all known Mendelian disorders. OMIM focuses on the relationship between genotype and phenotype. It is updated daily, and the entries contain links to many other genetics resources. OMIM and Online Mendelian Inheritance in Man are registered trademarks of the Johns Hopkins University. Copyright © 1966-2011 Johns Hopkins University.

## UniProt

Universal Protein Resource (UniProt) is a comprehensive, international resource for information about protein sequence and function. The UniProt accession number is a unique number assigned to an individual protein. The UniProt database includes enzymes as well as other proteins such as hemoglobin subunits and immunoglobulin chains. UniProt is the result of collaboration between the European Bioinformatics Institute, the Swiss Institute of Bioinformatics, and the Protein Information Resource, which is maintained by the Georgetown University Medical Center and the University of Delaware. The database is freely available at <http://www.uniprot.org> .

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### [Contact Help Desk](#)

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Site last updated: April 30, 2013

*Standards endorsed by CDC, NLM, HRSA, NIH and HITSAC*

# *What makes NBS messaging unique?*

- *Uses coded health vocabularies specific to NBS reporting.*
- *Accommodates reporting of Mother and Child data within a single message.*
- *Accommodates reporting of information about the father.*
- *In cases of adoption, accommodates reporting of caregiver or guardian.*
- *Includes specific data that is required to administer the Commonwealth's NBS program.*
- *Facilitates reporting of other pertinent data such as genetic information and treatment recommendations*

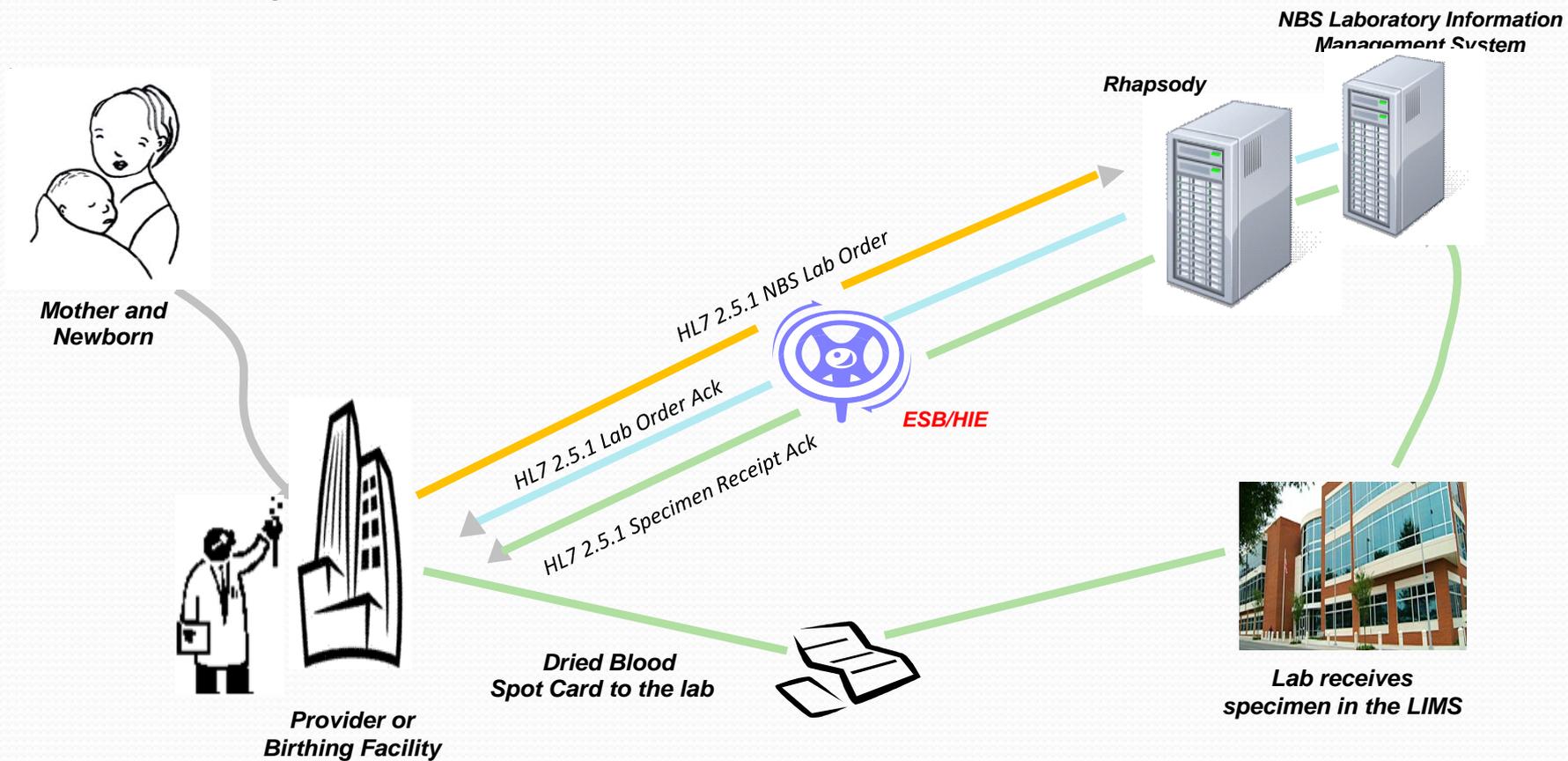
# *NBS Lab Order Submission*



# OML 021 NBS Lab Order

- Does not require the use of the Specimen (SPM) segment. This allows those systems already generating an HL7 version 2.3.1 messaging to create the 2.5.1 NBS laboratory order without complex system/database changes.
- Designed to support the interaction between the birthing facility and healthcare providers that conduct NBS screening and place a laboratory order, and the laboratories that fulfill the order.
- Does not identify, eliminate, or override variations in state or local jurisdiction requirements for data collection, reporting, or protection of privacy and security of patient data.
- Currently does not support other uses cases such as orders for hearing screening.
- Each OML^O21 message contains laboratory order information for a single Newborn Dried Blood Spot card (the specimen).

# Proposed – Lab Order Submission



# *NBS Results Reporting*



# ORU R01 - NBS Results Message

- NBS laboratory results Message was constrained from the full HL7 version v2.5.1 standard to simplify implementation by systems generating v2.3.1.
- Message is used for the reporting of electronic lab results in the current state of business process flow for Newborn Dried Blood Spot Screening.
- Supports the interaction between public health laboratories that conduct NBS results testing and primary care physicians, birth hospitals, public health agencies, health information exchanges (HIEs), and vital records departments.
- Does not support other uses cases such as orders for hearing screening.
- Result message contains patient information about mother and child

# HL7 NBS Result Reporting – Key Segments

- MSH** Defines the message source, purpose and destination.
- ✓ Sending laboratory is identified by a CLIA number
  - ✓ Hospital, birthing facility, provider or practice by an NPI number.
- PID** Contains baby information but also includes data from the mother's admission's record.
- NK1** Includes information about the mother and can also be used to include information about the father or caregiver. In cases where the mother's data is not reported (e.g. adoption), there is only one NK1 segment with the caregiver's information.
- ORC** Includes information that is universal to all orders. Hospitals/Birthing Facilities are generally identified using an NPI number and assigning authority identifier type. Laboratories are identified using a CLIA number and assigning authority. Providers are identified using an NPI number and OID for National Provider Identifiers.

# HL7 NBS Result Reporting – Key Segments

**OBR** Contains information about the test performed.

**OBX** Includes reason for test, specimen quality, overall interpretation, conditions with positive markers, and conditions with equivocal markers.  
Narrative summary segments are optional.

OBX|1|CE|57721-3^ Reason for lab test in Dried blood spot ^LN|1|LA12421-6^ Initial screen  
^LN|||N|||F

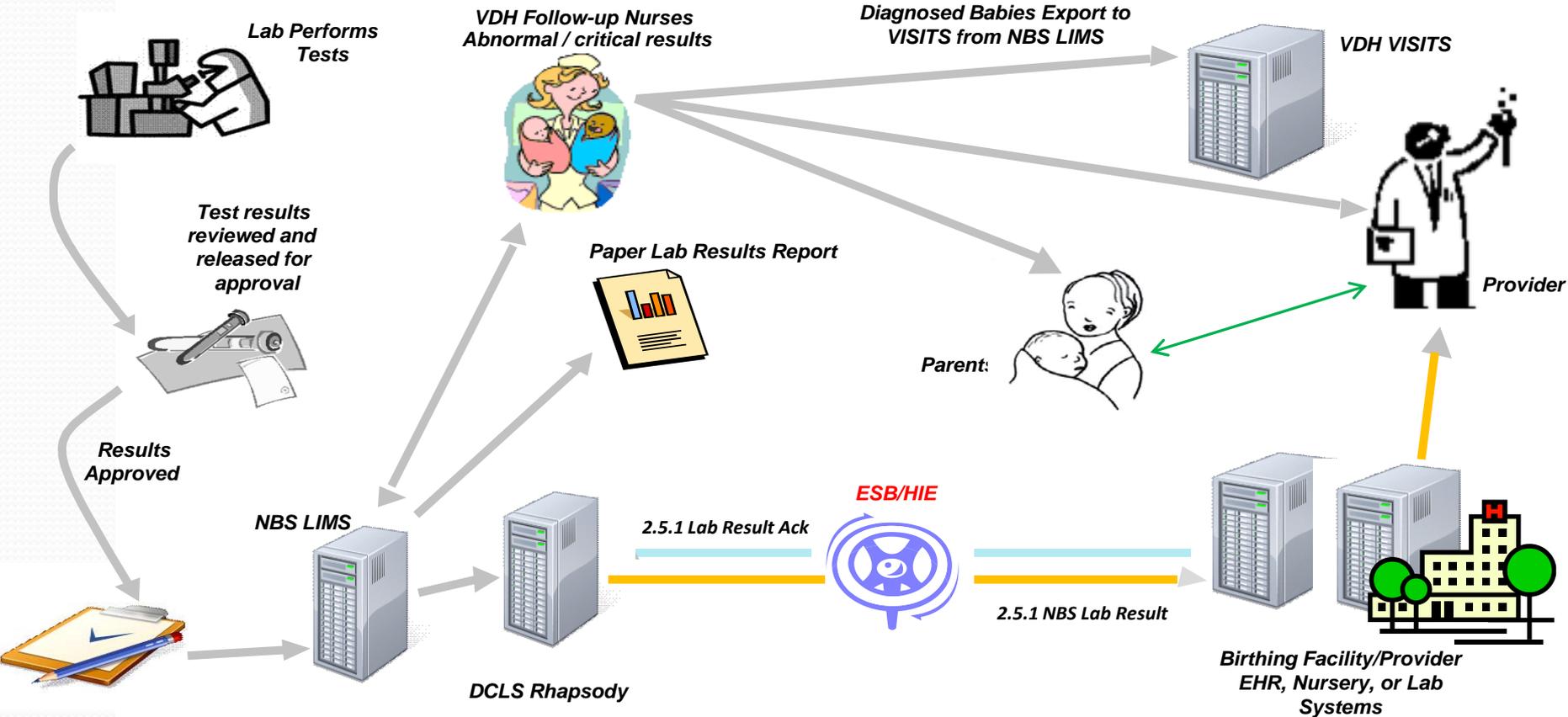
OBX|2|CE|57718-9^ Sample quality of Dried blood spot ^LN|1|LA12432-3^ Acceptable  
^LN|||N|||F

OBX|3|CE|57130-7^ Newborn screening report - overall interpretation ^LN|1|LA12431-5^ Not normal requiring immediate non-filter paper follow-up for at least one condition ^LN|||A|||F

OBX|4|CE|57131-5^ Newborn conditions with positive markers [Identifier] in Dried blood spot  
^LN|1|LA12509-8^MCAD^LN^128596003 ^Medium-chain acyl-coenzyme A dehydrogenase deficiency ^SCT|||A|||F

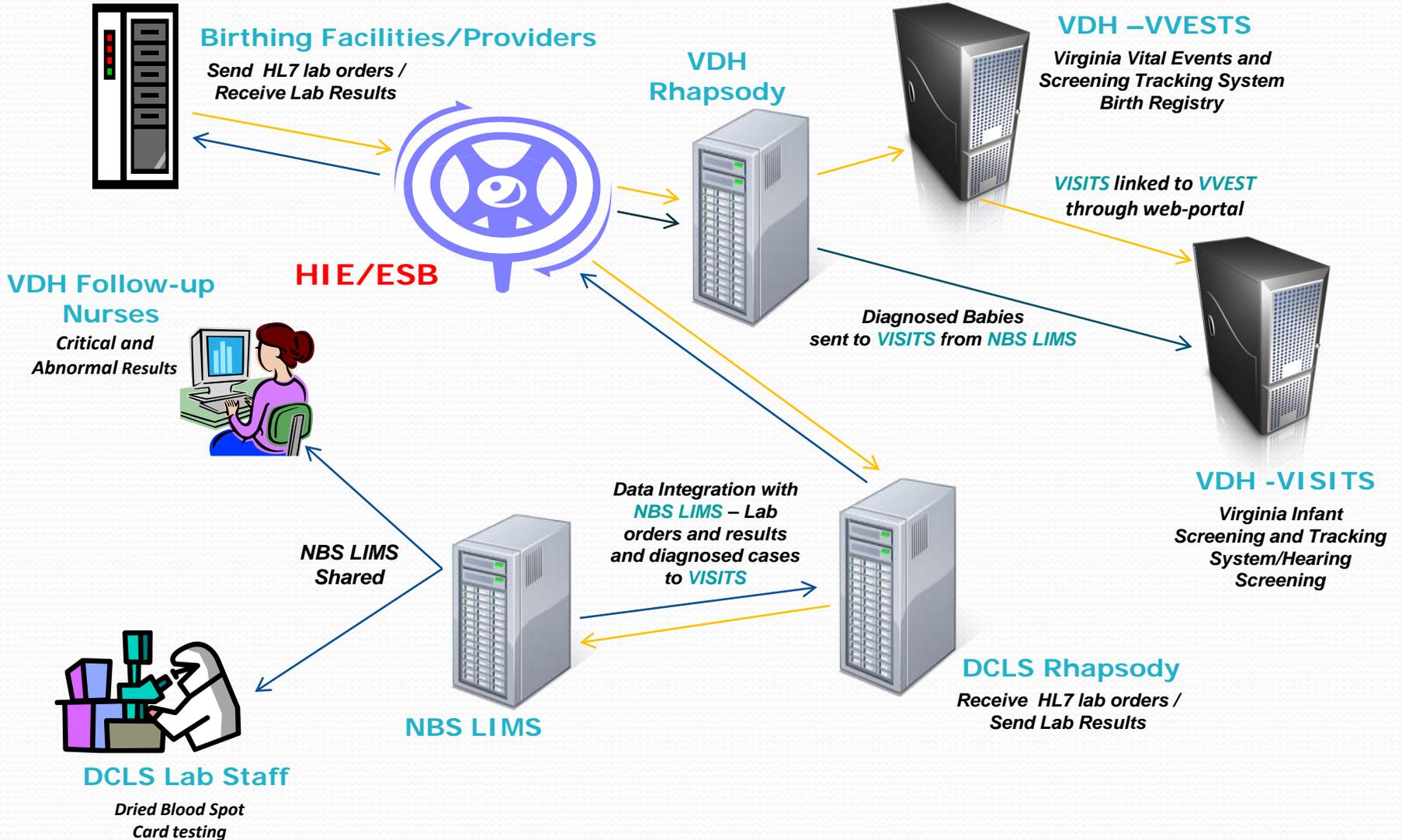
OBX|5|CE|57720-5^ Newborn conditions with equivocal markers [Identifier] in Dried blood spot  
^LN||LA12532-0^BIO^LN^8808004^ Biotinidase deficiency ^SCT|||A|||F

# Lab Result Reporting



# Proposed NBS Integration Architecture

Start Here



# *NBS LIMS - Upgrade*

*August 2013 – December 2015  
(28 months)*



# Implementation Considerations

## *Considerations in current environment?*

- ✓ *Enhance/modify Dried Blood Spot/Filter Paper Card?*
- ✓ *Re-engineer clinical workflows?*
- ✓ *Enhance follow-up and care management capabilities?*
- ✓ *Implement/transform business processes (reduce data entry/printing)?*

## *Considerations for future environment?*

- ✓ *New Reporting requirements – who needs this data?*
- ✓ *Standards adoption – use of coded vocabularies and HL7 messaging?*
- ✓ *Transformation - paper-based to electronic exchange – business impacts?*
- ✓ *New clinical workflows (SCIDS and Pompe disease) – need requirements?*

# Other Challenges and Considerations

- ✓ *Connecting to ESB – level of effort – how will this be funded/sustained?*
- ✓ *Use of Master Person Identifier (MPI) – who will be the assigning authority for NBS – how/when will this identifier be incorporated into the NBS workflow process?*
- ✓ *Use of HIE for data exchange – how will consent be managed – how to handle hospital/providers not connected to the HIE –need for sustainable funding for participation in HIE?*
- ✓ *Hospitals and Providers – timing – buy-in – realizing value added - ability and willingness to participate? (Pilots iNOVA and VCU Medical Center?)*
- ✓ *New legislation/regulations – Any plans to incorporate Meaningful Use objectives related to NBS screening or follow-up care?*

# Challenges

- *Aggressive timeline for implementation*
- *Sustainable funding*
- *Managing change – business transformation*
- *Stakeholder Buy-in – Realizing value-added*
- *Standards adoption and implementation*

# Benefits

- *Newer and supported technologies*
- *Standards based data exchange that promotes interoperability – State and Nation*
- *Business process efficiencies - Reduction in data entry, data errors and omissions*
- *Improved matching with vital records*
- *Enhanced data access/sharing for care management and statistical purposes*
- *Improved quality of care for newborns*
- *Leverage use of Master Person Identifier (MPI) for linking critical health data*
- *Creation of Infant/Child health record*
- *Earlier identification and reporting of NBS results so that timely treatment and intervention can take place, leading to healthier babies.*

# *Any Questions?*

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