Kentucky Newborn Screening

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MT (ASCP)

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Kentucky Public Health
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PHAB
Advancing public health performance

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CABINET FOR HEALTH AND FAMILY SERVICES

Learning Objectives

- Brief overview of the Newborn Screening (NBS) lab in Kentucky
- Incidence Rates
- Newborn Screen Reports Portal

Kentucky Department for Public Health

Mission and Vision in Action

Healthier People,
Healthier Communities.

Our mission is to improve the health and safety of people in Kentucky through prevention, promotion and protection.

Prevention	Promotion	Protection
Diabetes Prevention	Immunizations	Prescription Assistance
Disease Surveillance	KEIS	Public Health and Disaster Preparedness
Environmental Inspections	Mobile Harm Reduction	Smoking Cessation
HANDS	Newborn Screening	WIC

The Newborn Screening Laboratory in KY

- Location: Centralized Laboratory Facility in Frankfort
- Annual Volume: Kentucky's birth rate is approximately 52,000 babies per year; ~180 specimens/day
- Days Performed: Monday-Saturday and all holidays except Thanksgiving Day, Christmas Day, and New Year's Day

Recommended Uniform Screening Panel (RUSP)

- RUSP: Disorders recommended by the Secretary of the Department of Health and Human Services
  - 36 Core disorders
  - 26 Secondary disorders
- It is recommended that every newborn be screened for all disorders on the RUSP
- Kentucky Newborn Screening Statute is written to align with the Recommended Uniform Screening Panel (RUSP)

https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp

How Conditions are Added to the RUSP

- Adding a new condition to the RUSP is a multistep process: First, someone nominates the condition to the RUSP. Any person or group(s) can do this by completing a nomination package.
- The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) meet regularly to discuss proposals from parent advocates, organizations and experts in order to keep newborn screening up to date. In addition, the Secretary of the U.S. Department of Health and Human Services reviews the Committee's recommendations.
- Disorders on the RUSP are chosen based on evidence that supports the potential net benefit of screening, the ability of states to screen for the disorder, and the availability of effective treatments.

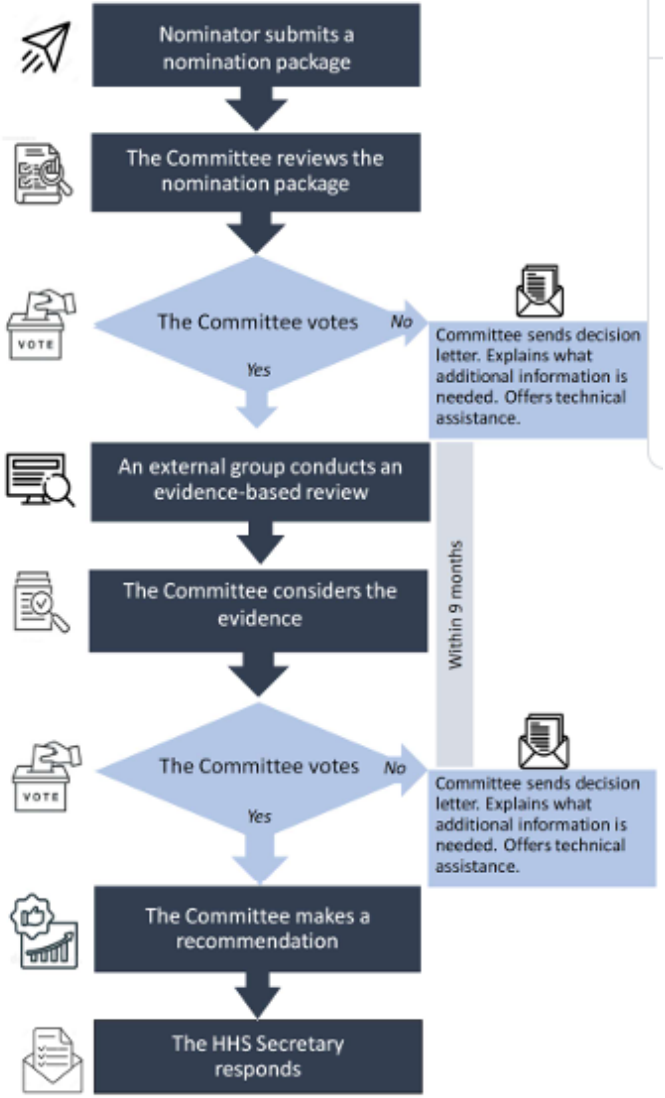
Steps for Adding a Condition to the RUSP

Steps for Adding a Condition to the Recommended Uniform Screening Panel (RUSP)

Step	Description
Nominator submits a nomination package	To start the process, a team of experts and stakeholders complete a nomination package and submit it to the Committee.*
Committee reviews the nomination package	The Committee reviews the nomination package. It considers whether the package met the requirements and addressed key questions about the condition, screening process, and treatment. If the nomination package has met the requirements, a Committee member presents a summary at a full Committee meeting.
The Committee votes:	The Committee reviews and discusses the nomination further. If the nomination requirements are met, then the Committee will vote on whether or not to move the condition forward for more review. If the full Committee review and discussion finds that the nomination package does not meet requirements, the Committee may choose not to vote and ask the nominators to provide missing information and/or provide nominators with technical assistance on developing the nomination package.
No	If the Committee votes *no*, the condition does not move forward. The Committee will explain what else is needed, and provide technical assistance to the nominators
Yes	If the Committee votes *yes*, the condition moves forward for an evidence-based review.
The ERG conducts an Evidence-Based Review	In an evidence-based review, an external Evidence-Based Review Group (ERG) gathers detailed data on how screening and treatment for the condition affect newborns, the population, and the public health system. The ERG prepares a final report and presents it to the Committee.
The Committee considers the evidence	The Committee reviews the ERG report, then discusses and rates the data on three main points: screening benefits, screening feasibility, and state readiness to begin screening.
The Committee votes:	The Committee votes on whether to recommend adding the nominated condition to the RUSP.
No	If the Committee votes *no*, it does not recommend adding the condition. The Committee will explain about what else is needed, and provide technical assistance to the nominators.
Yes	If the Committee votes *yes*, it recommends adding the condition to the RUSP by sending a letter to the HHS Secretary.
HHS Secretary responds	The HHS Secretary reviews the Committee recommendation and decides whether or not to add the condition to the RUSP.

**The full name of the Committee involved is the "Advisory Committee on Heritable Disorders in Newborns and Children." The Consumer Guide uses the term "Committee" to refer to this group.*

Diagram of Steps for Adding a Condition to the RUSP



NBS Panel in Kentucky

- **Fatty Acid Oxidation Disorders:**
  - **Carnitine Uptake Defect (CUD)**
  - **Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCADD)**
  - **Long-chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHADD)**
  - **Trifunctional Protein Deficiency (TFP)**
  - **Very Long-chain Acyl-CoA Dehydrogenase Deficiency (VLCADD)**
  - Short-chain Acyl-CoA Dehydrogenase Deficiency (SCADD)
  - Carnitine Acylcarnitine Translocase Deficiency (CACT)
  - Carnitine Palmitoyl Transferase 1 Deficiency (CPT-1)
  - Carnitine Palmitoyl Transferase 2 Deficiency (CPT-2)
  - Ethylmalonic Encephalopathy (EE)
  - Glutaric Acidemia Type 2 (GA-2)
  - 2,4 Dienoyl-CoA Reductase Deficiency (DE RED)

- **Organic Acid Disorders:**
  - **3-Methlycrotonyl-CoA Carboxylase Deficiency (3MCC)**
  - **Glutaric Acidemia Type 1 (GA-1)**
  - **Beta-kethothiolase Deficiency (BKT)**
  - **Hydroxymethylglutaric Aciduria (HMG)**
  - **Isovaleric Acidemia (IVA)**
  - **Multiple Carboxylase Deficiency (MCD)**
  - **Methylmalonic Acidemia (MMA Cbl A form and MMA Cbl B form)**
  - **Methylmalonyl-CoA Mutase Deficiency (MUT)**
  - **Propionic Acidemia (PA)**
  - 2-Methyl-3-Hydroxybutyric Aciduria (2M3HBA)
  - 2-Methylbutyryl-CoA Dehydrogenase Deficiency (2MBDH)
  - 3-Methylglutaconic Aciduria (3MGA)
  - Methylmalonic Acidemia (MMA Cbl C form and MMA Cbl D form)
  - Malonic Acidemia (MAL)
  - Isobutyryl-CoA Dehydrogenase Deficiency (IBG)

- **Amino Acid Disorders:**
  - **Argininosuccinic Aciduria (ASA)**
  - **Citrullinemia (CIT-1)**
  - **Homocystinuria (HCY)**
  - **Maple Syrup Urine Disease (MSUD)**
  - **Phenylketonuria (PKU)**
  - **Tyrosinemia Type 1 (TYR-1)**
  - Citrullinemia Type 2 (CIT-2)
  - Hypermethioninemia (MET)
  - Hyperphenylalaninemia (H-PHE)Tyrosinemia Type 2 (TYR-2)
  - Tyrosinemia Type 3 (TYR-3)
  - Arginase Deficiency (ARG)
  - Non-Ketotic Hyperglycinemia (NKHG)

- **Endocrine Disorders:**
  - **Congenital Adrenal Hyperplasia (CAH)**
  - **Congenital Hypothyroidism (TSH and T4)**

- **Hemoglobin Disorders:**
  - **Hb S/Beta-thalassemia (HbS/Th)**
  - **Hb S/C Disease (Hb S/C)**
  - **Sickle Cell Anemia (Hb S/S)**
  - Various Hemoglobinopathies (includes Hb E)

- **Lysosomal and Peroxisomal Disorders:**
  - **Mucopolysaccharidosis (MPS-1, Hurler's Disease)**
  - **Pompe**
  - Krabbe
  - **X-linked adrenoleukodystrophy (X-ALD)**

- **Other Disorders:**
  - **Cystic Fibrosis (CF)**
  - **Galactosemia (GAL)**
  - **Biotinidase Deficiency (BIOT)**
  - **Severe Combined Immunodeficiency (SCID)**
  - **Spinal Muscular Atrophy (SMA)**

- **Point of Care Testing:**
  - **Critical Congenital Heart Defects**
  - **Hearing**

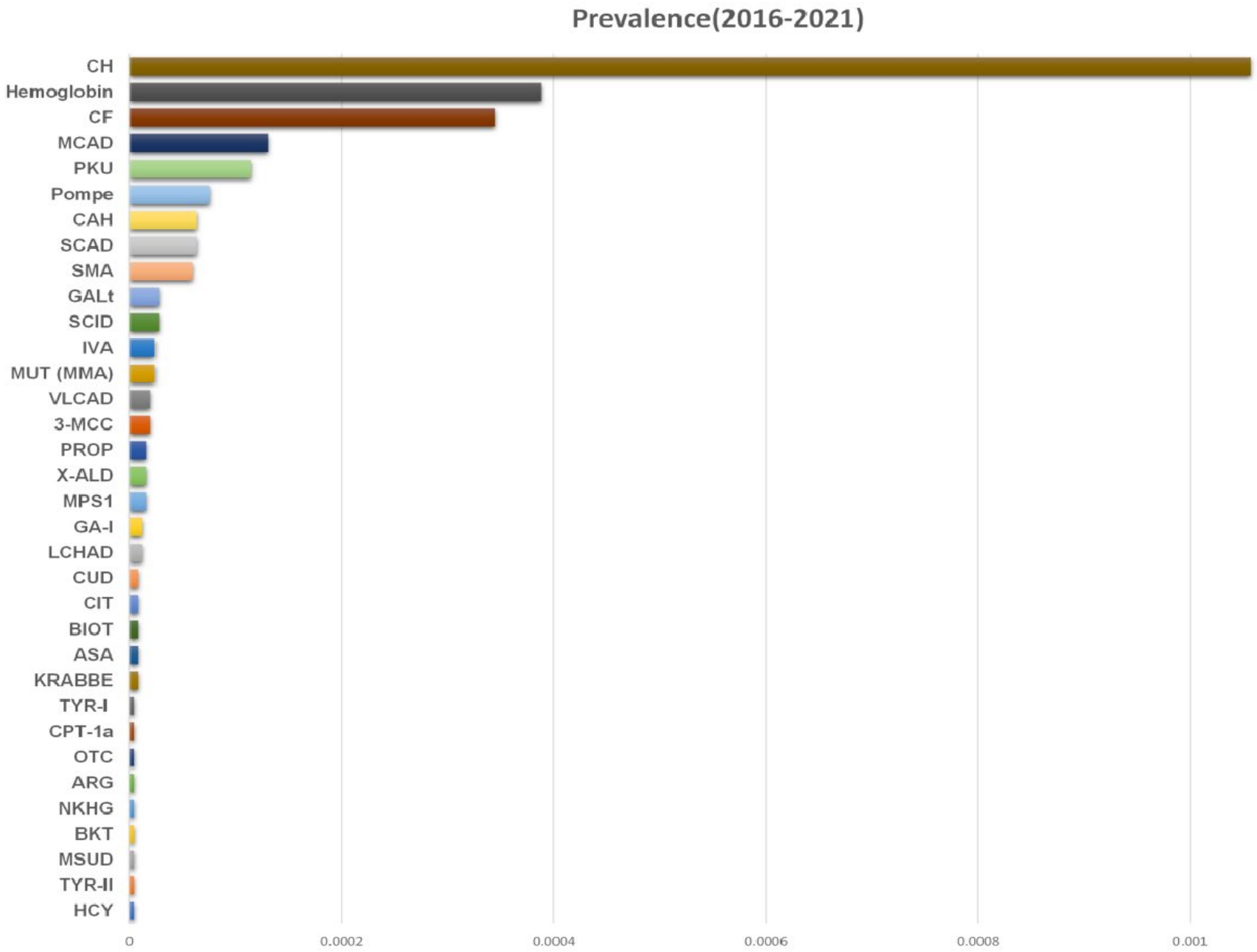
Core Conditions in Bold

55 disorders in total

Conditions	2016 True positive	2017 true positive	2018 true positive	2019 true positive	2020 true positive	2021 true positive	Prevalence rate	Incidence Rate
3-MCC	1	2	0	2	5/252,508	1/50,502		
ARG	1	0	0	0	1/252,508	1/252,508		
ASA	0	1	1	0	2/252,508	1/126,254		
BIOT	0	0	1	0	2/252,508	1/126,254		
BKT	1	0	0	0	1/252,508	1/252,508		
CAH	4	1	4	1	16/252,508	1/15,782		
Cbl A,B	0	0	0	0	0			
CF	12	27	18	15	87/252,508	1/2,902		
CH	45	54	66	53	258/252,508	1/979		
CIT	0	0	0	1	2/252,508	1/126,254		
CPT-1a	0	0	0	0	1/252,508	1/252,508		
CPT-II	0	0	0	0	0			
CUD	0	0	0	2	0	2/252,508	1/126.254	
EE	0	0	0	0	0			
GA-I	1	0	2	0	0	3/252,508	1/84,169	
GA-II	0	0	0	0	0			
GALt	2	2	2	0	1	7/252,508	1/36,073	
Hemoglobin	15	31	12	25	16	99/252,508	1/2,577	
HCY	1	0	0	0	0	1/252,508	1/252,508	
IVA	0	1	1	3	1	6/252,508	1/42,085	
KRABBE	1	0	0	0	0	1	2/298,084	1/126,254
LCHAD	0	0	0	1	1	3/252,508	1/84,176	
MCAD	3	10	8	4	8	33/252,508	1/7,652	
MCD	0	0	0	0	0			
MPS1	1	1	1	1	1	0	4/298,084	1/74,521
MSUD	0	0	1	0	0	1/252,508	1/252,508	
MUT (MMA)	2	1	2	1	0	6/252,508	1/42,085	
NKHG	0	1	0	0	0	1/252,508	1/252,508	
OTC	0	1	0	0	0	1/252,508	1/252,508	
PHE	0	0	0	0	0			
PKU	4	9	3	2	11	29/252,508	1/8,707	
Pompe	3	6	1	7	2	20/298,084	1/14,904	
PROP	1	2	0	1	0	4/252,508	1/63,127	
SCAD	6	2	5	1	2	16/252,508	1/15,782	
SCHAD	0	0	0	0	0			
SCID	1	0	1	2	3	7/252,508	1/36,073	
SMA	N/A	N/A	4	4	7	15/114,154	1/7,610	
TYR-I	0	1	0	0	0	1/252,508	1/252,508	
TYR-II	0	0	0	1	0	1/252,508	1/252,508	
VLCAD	1	1	2	0	1	5/252,508	1/50,502	
X-ALD	N/A	1	2	0	1	4/174,568	1/43,642	
Total True Positives				137	125	121		
Average True Positives out of annual specimens				1/400	1/400	1/400		

2017: 52, 012 initial specimens
2018: 51,400 initial specimens
2019: 50, 520 initial specimens
2020: 49,056 initial specimens
2021: 49,520 initial specimens

initial specimens


Prevalence(2016-2021)
CH
Hemoglobin
CF
MCAD
PKU
Pompe
CAH
SCAD
SMA
GALt
SCID
IVA
MUT (MMA)
VLCAD
3-MCC
PROP
X-ALD
MPS1
GA-I
LCHAD
CUD
CIT
BIOT
ASA
KRABBE
TYR-I
CPT-1a
OTC
ARG
NKHG
BKT
MSUD
TYR-II
HCY
0
0.0002
0.0004
0.0006
0.0008
0.001

Division of Laboratory Services

Kentucky Hospitals, Health Departments, and Birthing Facilities

Kentucky Public Health
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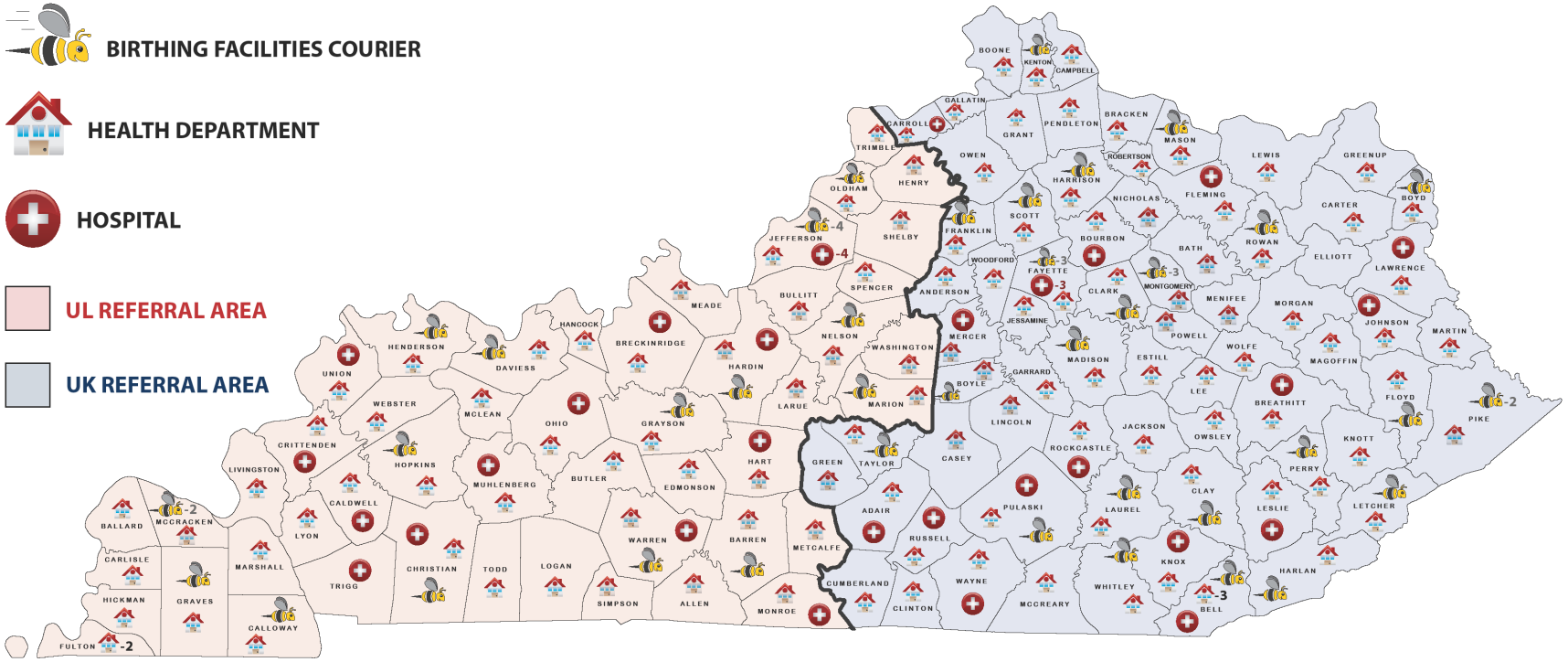
BIRTHING FACILITIES COURIER

HEALTH DEPARTMENT

HOSPITAL

UL REFERRAL AREA

UK REFERRAL AREA



BIRTHING FACILITIES - CITY - COUNTY

Baptist Health Corbin	Corbin	Whitley -Knox	St. Claire Regional Medical Center	Morehead	Rowan
Baptist Health LaGrange	LaGrange	Oldham	St Elizabeth Health Care	Edgewood	Kenton
Baptist Health Lexington	Lexington	Fayette	Women's Hospital at St Joseph East	Lexington	Fayette
Baptist Health Louisville	Louisville	Jefferson	St Joseph London	London	Laurel
Baptist Health Madisonville	Madisonville	Hopkins	St Joseph Mount Sterling	Mount Sterling	Montgomery
Baptist Health Paducah	Paducah	McCracken	St Joseph Mount Sterling	Mount Sterling	Montgomery
Baptist Health Richmond	Richmond	Madison	St Joseph Mount Sterling	Mount Sterling	Montgomery
Clark Regional Medical Center	Winchester	Clark	Taylor Regional Hospital	Campbellsville	Taylor
Ephraim McDowell Fort Logan	Stanford	Lincoln	The Medical Center at Bowling Green	Bowling Green	Warren
Ephraim McDowell Regional	Danville	Boyle	TJ Sampson Community	Glasgow	Barren
Flaget Memorial	Bardstown	Nelson	Twin Lakes Regional Medical Center	Leitchfield	Grayson
Frankfort Regional Medical Center	Frankfort	Franklin	UK Hospital	Lexington	Fayette
Georgetown Community	Georgetown	Scott	UofL Hospital	Louisville	Jefferson
Baptist Health Hardin	Elizabethtown	Hardin	Whitesburg ARH	Whitesburg	Letcher
Harlan ARH Hospital	Harlan	Harlan	Williamson ARH/ Tug Valley	South Williamson	Pike
Harrison Memorial Hospital	Cynthiana	Harrison			
Hazard ARH Regional	Hazard	Perry			
Highlands ARH Regional	Prestonsburg	Floyd			
Jackson Purchase Medical Center	Mayfield	Graves			
Jennie Stuart Medical Center	Hopkinsville	Christian			
King's Daughter Medical Center	Ashland	Boyd			
Lake Cumberland Regional	Somerset	Pulaski			
Lourdes	Paducah	McCracken			
Advent Health Manchester	Manchester	Clay			
Meadowview Regional	Maysville	Mason			
Deaconess Henderson	Henderson	Henderson			
Middlesboro ARH Hospital	Middlesboro	Bell			
Murray Calloway County	Murray	Calloway			
Norton Hospital	Louisville	Jefferson			
Norton Childrens Hospital	Louisville	Jefferson			
Norton Women and Childrens Hospital	Louisville	Jefferson			
Owensboro Medical Health System	Owensboro	Daviess			
Pikeville Medical Center	Pikeville	Pike			
Spring View Hospital	Lebanon	Marion			

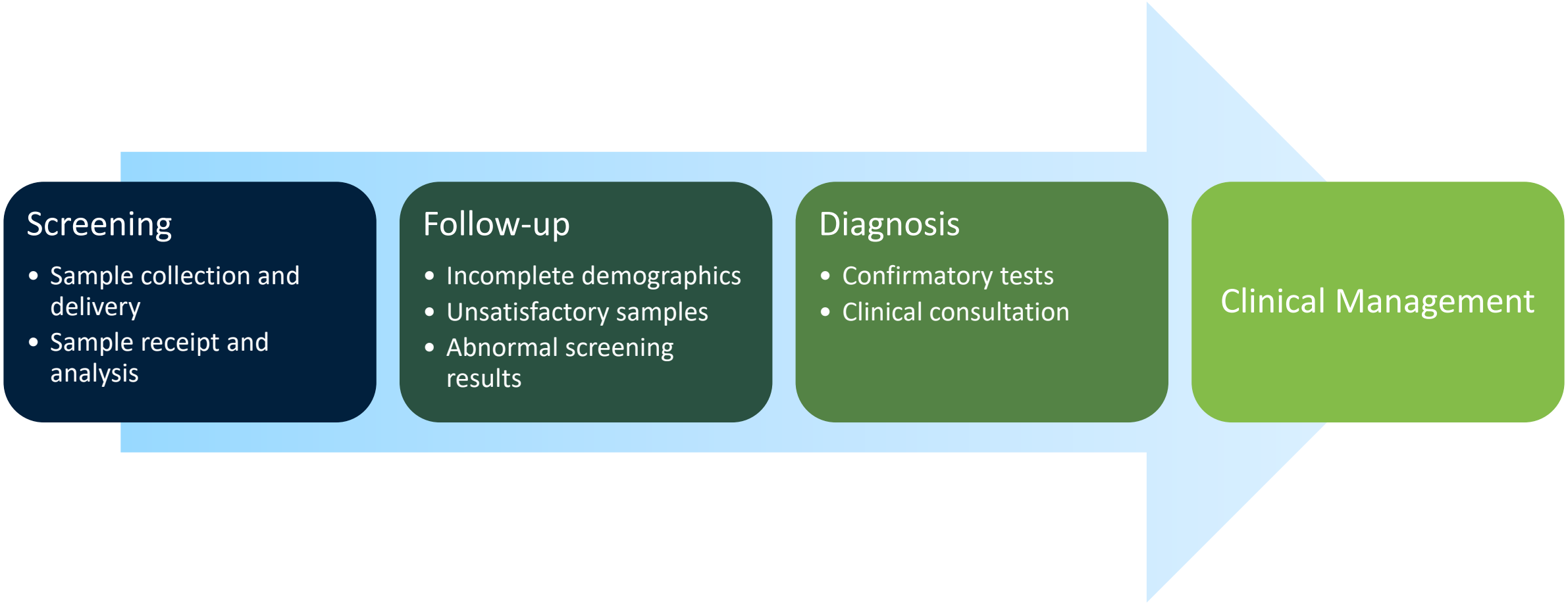
HEALTH DEPARTMENT - CITY - COUNTY

Adair County	Columbia	Adair	Estill	Irvine	Estill	Lewis	Vanceburg	Lewis	Powell
Allen County	Scottsville	Allen	Fleming	Flemingsburg	Fleming	Lexington Fayette	Lexington	Fayette	Pulaski
Anderson County	Lawrenceburg	Anderson	Floyd	Prestonsburg	Floyd	Lincoln	Stanford	Lincoln	Robertson
Ashland-Boyd	Ashland	Ashland-Boyd	Franklin	Frankfort	Franklin	Livingston	Smithland	Livingston	Rockcastle
Ballard	LaCenter	Ballard	Fulton	Hickman	Fulton	Logan	Russellville	Logan	Rowan
Barren	Glasgow	Barren	Fulton East	Fulton	Fulton	Louisville Metro Health	Louisville	Jefferson	Russel
Bath	Owingsville	Bath	Gallatin	Warsaw	Gallatin	Lyon	Eddyville	Lyon	Scott
Bell	Pineville	Bell	Garrard	Lancaster	Garrard	Magoffin	Salyersville	Magoffin	Shelby
Bell - Middlesboro	Middlesboro	Bell	Grant	Williamstown	Grant	Madison	Richmond	Madison	Simpson
Bourbon	Paris	Bourbon	Graves	Mayfield	Graves	Marion	Lebanon	Marion	Spencer
Boyle	Danville	Boyle	Grayson	Leitchfield	Grayson	Marshall	Benton	Marshall	Warren
Boone	Florence	Boone	Green	Greensburg	Green	Martin	Inez	Martin	Whitley
Bracken	Brooksville	Bracken	Greenup	Greenup	Greenup	Mason	Maysville	Mason	Woodford
Breathitt	Jackson	Breathitt	Hancock	Hawesville	Hancock	McCreary	Whitley City	McCreary	Taylor
Breckinridge	Hardinsburg	Breckinridge	Hardin	Elizabethtown	Hardin	McLean	Calhoun	McLean	Todd
Bullitt	Shepardsville	Bullitt	Harlan	Harlan	Harlan	Meade	Brandenburg	Meade	Trimble
Butler	Morgantown	Butler	Harrison	Cynthiana	Harrison	Menifee	Frenchburg	Menifee	Union
Caldwell	Princeton	Caldwell	Hart	Munfordville	Hart	Mercer	Harrodsburg	Mercer	Washington
Calloway	Murray	Calloway	Henderson	Henderson	Henderson	Metcalfe	Edmonton	Metcalfe	Wayne
Campbell	Newport	Campbell	Henry	New Castle	Henry	Monroe	Tompkinsville	Monroe	Webster
Carlisle	Bardwell	Carlisle	Hickman	Clinton	Hickman	Montgomery	Mt. Sterling	Montgomery	Wolfe
Carroll	Carrollton	Carroll	Hopkins	Madisonville	Hopkins	Morgan	West Liberty	Morgan	
Carter	Grayson	Carter	Jackson	McKee	Jackson	Muhlenburg	Central City	Muhlenburg	
Casey	Liberty	Casey	Jessamine	Nicholasville	Jessamine	Nelson	Bardstown	Nelson	
Christian	Hopkinsville	Christian	Johnson	Paintsville	Johnson	Nicholas	Carlisle	Nicholas	
Clark	Winchester	Clark	Kenton	Covington	Kenton	Ohio	Hartford	Ohio	
Clay	Manchester	Clay	Knott	Hindman	Knott	Oldham	LaGrange	Oldham	
Clinton	Albany	Clinton	Knox	Barbourville	Knox	Owen	Owenton	Owen	
Crittenden	Marion	Crittenden	Laurel	London	Laurel	Owsley	Booneville	Owsley	
Cumberland	Burkesville	Cumberland	Larue	Hodgenville	Larue	Paducah McCracken	Paducah	McCracken	
Cumberland Valley Environ	Middlesboro	Bell	Lawerence	Louisa	Lawrence	Pendleton	Falmouth	Pendleton	
Daviess	Owensboro	Daviess	Lee	Beattyville	Lee	Pennyrile Environmental	Eddyville	Lyon	
Edmonson	Brownsville	Edmonson	Leslie	Hyden	Leslie	Perry	Hazard	Perry	
Elliott	Sandy Hook	Elliott	Letcher	Whitesburg	Letcher	Pike	Pikeville	Pike	

HOSPITAL - CITY - COUNTY

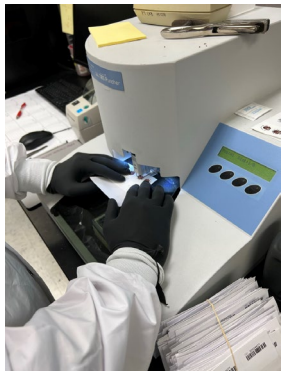
Barbourville ARH	Bourbourville	Knox	Lewis	Lexington Fayette	Vanceburg
Blanchfield Community Hospital	Fort Campbell	Christian	Lexington	Fayette	Lewis
Bourbon Community Hospital	Paris	Bourbon	Stanford	Lincoln	Lincoln
Breckinridge Memorial Hospital	Hardinsburg	Breckinridge	Smithland	Livingston	Livingston
Caldwell Memorial Hospital	Princeton	Caldwell	Russellville	Logan	Logan
Carroll County Hospital	Carrollton	Carroll	Louisville	Jefferson	Louisville
Crittenden Health System	Marion	Crittenden	Eddyville	Lyon	Eddyville
Cumberland Medical Laboratory	Somerset	Pulaski	Salyersville	Magoffin	Salyersville
Fleming County Hospital	Flemingsburg	Fleming	Richmond	Madison	Richmond
Greenview Regional Hospital	Bowling Green	Warren	Marion	Marion	Marion
Ireland Army Hospital	Fort Knox	Hardin	Marshall	Marshall	Marshall
James B Haggin	Harrodsburg	Mercer	Martin	Martin	Martin
Jewish Hospital	Louisville	Jefferson	Mason	Mason	Mason
Kindred Hospital	Louisville	Jefferson	McCreary	McCreary	McCreary
KY River Medical Center	Jackson	Breathitt	McLean	McLean	McLean
Mary Breckinridge Hospital	Hyden	Leslie	Meade	Meade	Meade
Methodist Hospital Union	Morganfield	Union	Menifee	Menifee	Menifee
Monroe Co. Medical Center	Tompkinsville	Monroe	Mercer	Mercer	Mercer
Ohio County Hospital	Hartford	Ohio	Metcalfe	Metcalfe	Metcalfe
Owensboro Medical Health System Muhlenburg	Greenville	Muhlenburg	Monroe	Monroe	Monroe
Paul B Hall Regional Medical Center	Paintsville	Johnson	Montgomery	Montgomery	Montgomery
Pineville Medical Center	Pineville	Bell	Morgan	Morgan	Morgan
Rockcastle Hospital	Mt. Vernon	Rockcastle	Muhlenburg	Muhlenburg	Muhlenburg
Roederer Correctional	LaGrange	Oldham	Nelson	Nelson	Nelson
Russell County Hospital	Russell Springs	Russell	Nicholas	Nicholas	Nicholas
Samaritan	Lexington	Fayette	Ohio	Ohio	Ohio
St. Joseph Main	Lexington	Fayette	Oldham	Oldham	Oldham
St. Mary and Elizabeth	Louisville	Jefferson	Owen	Owen	Owen
The Medical Center at Caverna	Horse Cave	Hart	Owsley	Owsley	Owsley
TJ Samson Columbia	Columbia	Adair	Paducah McCracken	Paducah	McCracken
Three Rivers Medical Center	Louisa	Lawrence	Pendleton	Falmouth	Pendleton
Trigg County Hospital	Cadiz	Trigg	Pennyrile Environmental	Eddyville	Lyon
VA Medical Center	Louisville	Jefferson	Perry	Hazard	Perry
Wayne County Hospital	Monticello	Wayne	Pike	Pikeville	Pike

NBS Program in KY

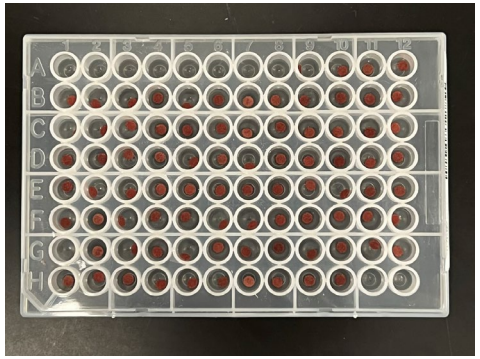


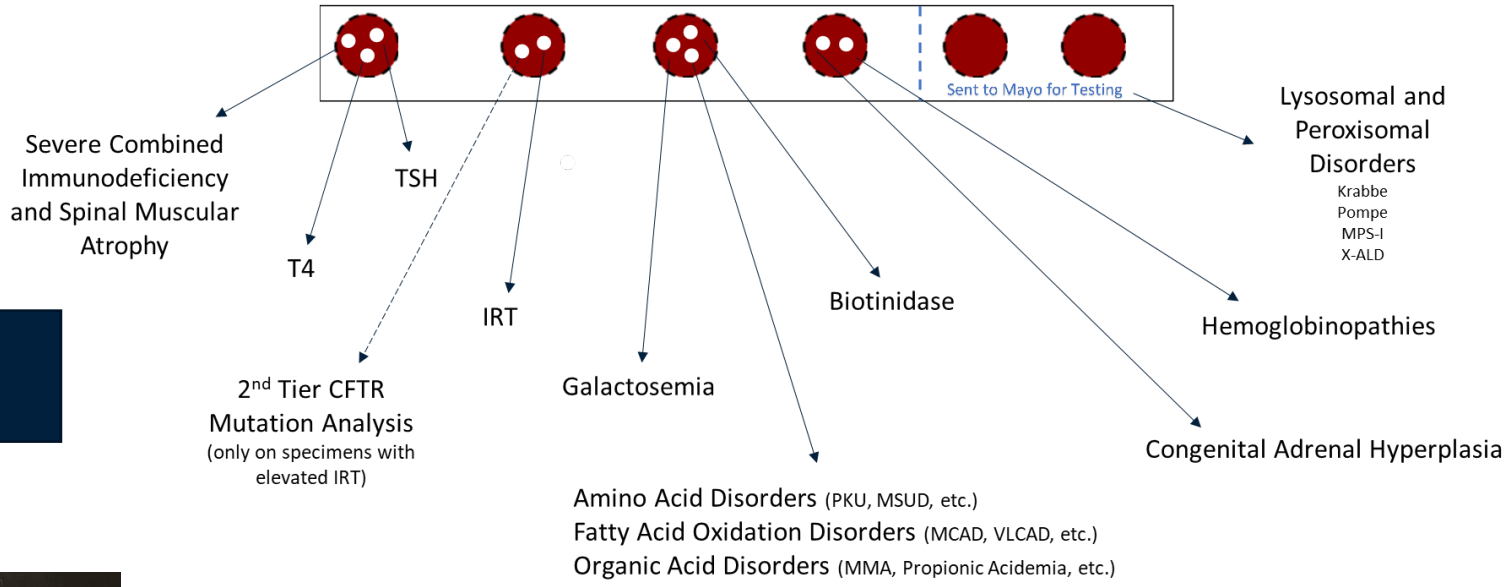
NBS at KY DLS: Workflow

Specimen Receipt:
Within 24-48hrs of age



Specimen Processing





For approximately 10% of specimens, additional punches are necessary to retest abnormal results.

NBS in KY: Challenges from Lab Perspective

- National recommendations call for all critical results to be reported within 5 days and all results to be reported within 7 days
- Data from our lab:
  - 6% of specimens are delayed in transit
  - 25% of specimens are collected >30hrs (Must be 24-48hrs to meet recommendation and we prefer collection closer to 24 hours)
  - Looking at June-December 2022 we had 102 critical specimens and 24 specimens were >5 days due to collection or transit

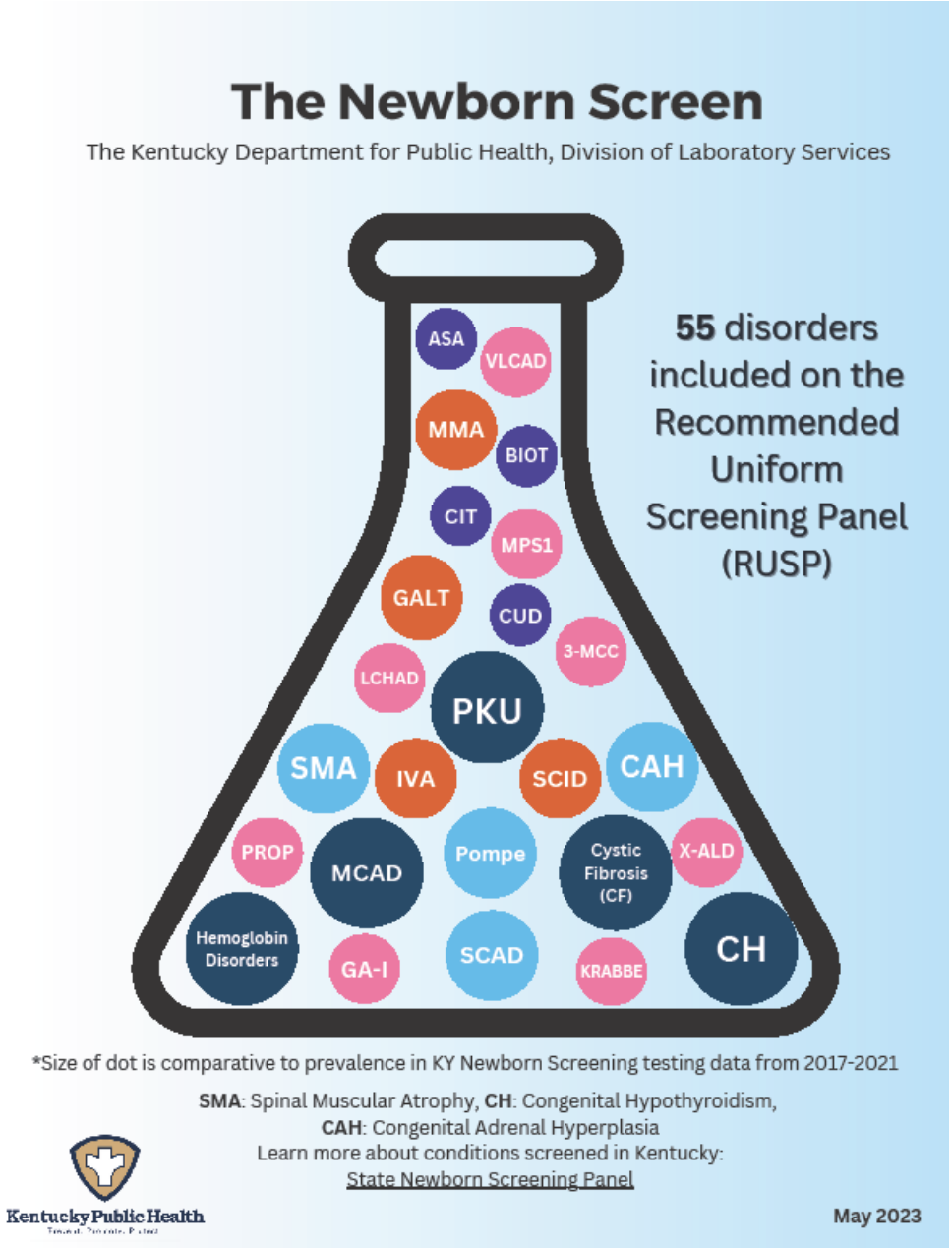
<u>Good quality NBS requires teamwork !</u>

Help us educate new parents:

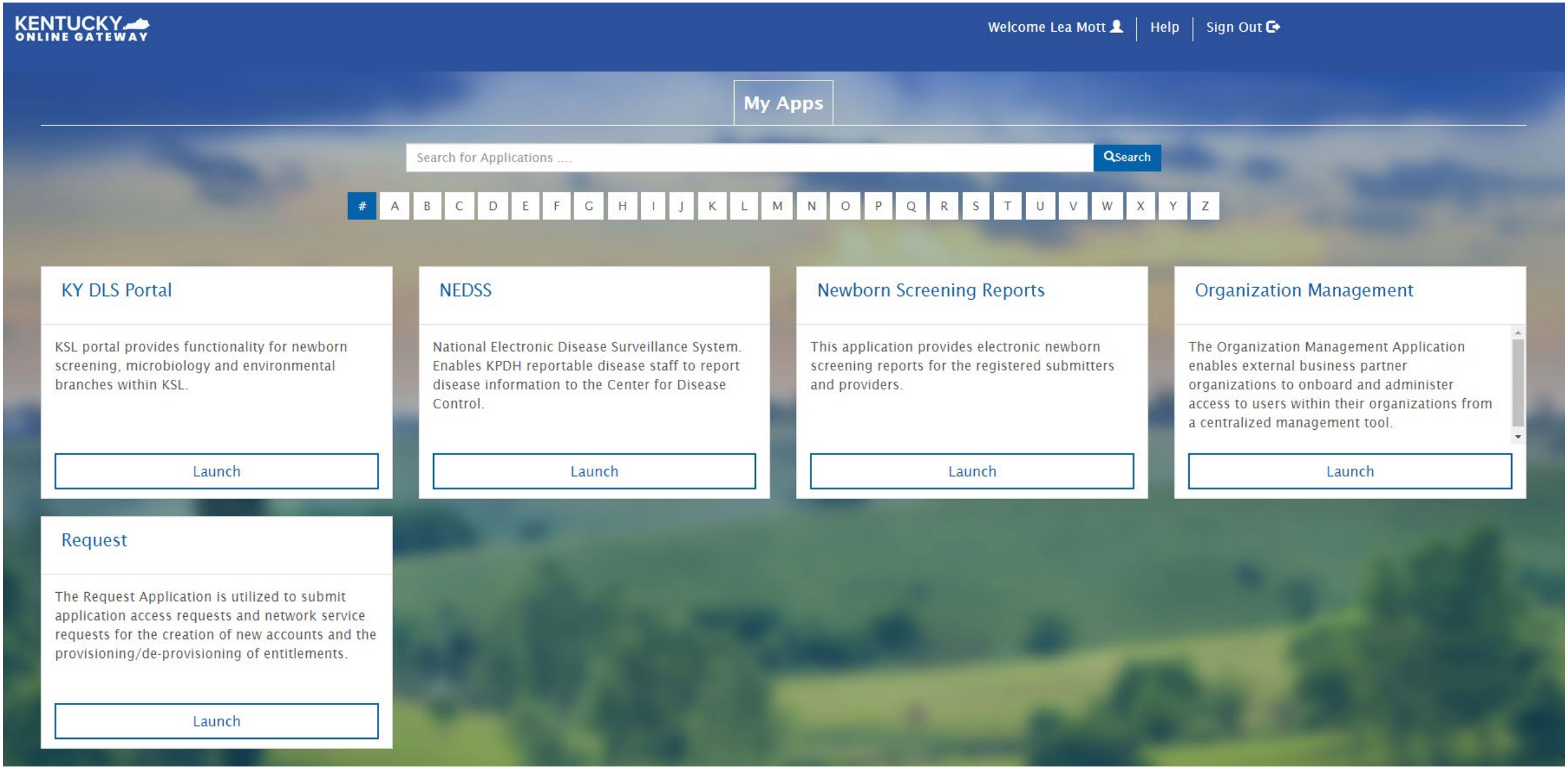
- Encourage them to have a pediatrician chosen prior to delivery
- Remind them to check that the pediatrician is accepting new patients
- Tell them to make sure the pediatrician takes their insurance

Newborn screening is more than just PKU

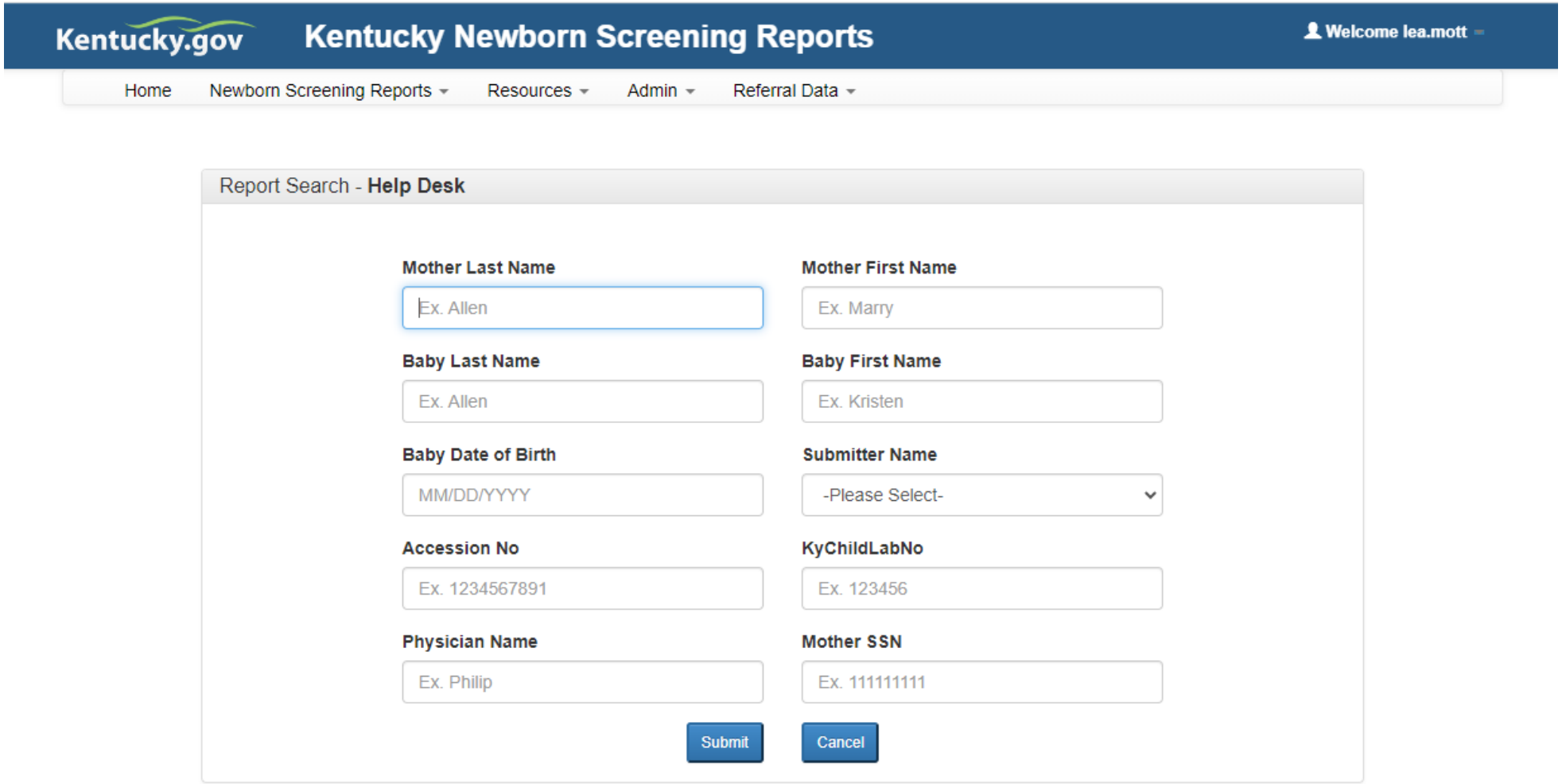
- Help us change the culture of referring to the newborn screen as “the PKU”
- Using one disorder as a shortcut for a panel that includes 54 other disorders leads to confusion



Newborn Screen Reports Portal


KENTUCKY ONLINE GATEWAY
Welcome Lea Mott
Help
Sign Out
My Apps
Search for Applications ....
Search
A B C D E F G H I J K L M N O P Q R S T U V W X Y Z
KY DLS Portal
KSL portal provides functionality for newborn screening, microbiology and environmental branches within KSL.
Launch
NEDSS
National Electronic Disease Surveillance System. Enables KPDH reportable disease staff to report disease information to the Center for Disease Control.
Launch
Newborn Screening Reports
This application provides electronic newborn screening reports for the registered submitters and providers.
Launch
Organization Management
The Organization Management Application enables external business partner organizations to onboard and administer access to users within their organizations from a centralized management tool.
Launch
Request
The Request Application is utilized to submit application access requests and network service requests for the creation of new accounts and the provisioning/de-provisioning of entitlements.
Launch


Kentucky.gov
Kentucky Newborn Screening Reports
Welcome lea.mott
Home
Newborn Screening Reports
Resources
Admin
Referral Data
Welcome to Kentucky Division of Laboratory Services - Newborn Screening Reports


Kentucky.gov
Kentucky Newborn Screening Reports
Welcome lea.mott
Home
Newborn Screening Reports
Resources
Admin
Referral Data
Report Search - Help Desk
Mother Last Name
Ex. Allen
Mother First Name
Ex. Marry
Baby Last Name
Ex. Allen
Baby First Name
Ex. Kristen
Baby Date of Birth
MM/DD/YYYY
Submitter Name
-Please Select-
Accession No
Ex. 1234567891
KyChildLabNo
Ex. 123456
Physician Name
Ex. Philip
Mother SSN
Ex. 111111111
Submit
Cancel

NBS Reports

Search:

		Accession No	Mother Last Name	Mother First Name	Baby Last Name	Baby First Name	Baby DOB	Submitter Name	Physician Name
Screening under process	Card	1234567890	TEST	TEST	TEST	BABY	05/27/2016	HOSPITAL	TEST,PHYSICIAN

Showing 1 to 1 of 1 entries

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Report Search - **Help Desk**

Mother Last Name
Ex. Allen

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NBS Reports

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KyChildLabNo
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Physician Name
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Mother SSN
Ex. 111111111

Submit Cancel

Control ID:
111111111
Submitter Ref. No.
111111

KY Cabinet for Health and Family Services
Department for Public Health - Division of Laboratory Services (KY State Lab)
100 Sower Blvd Suite 204, Frankfort, KY 40601-8272

Newborn Screening Tests

Mother's Name (L,F): TEST, TEST
Mother's Address: 100 MAIN ST
City, State, Zip: CITY, KY 40004 502 XXX
Mother's Phone: XXXX
Mother's SSN: XXX-XX-XXXX

Baby's Name (L,F): TEST, BABY
Date of Birth: 5/27/16 12:54 pm
Baby's Sex: F Baby's Weight: 3,515 g
Gestation Age: Greater than or equal to 37 Weeks
Physician: TEST, PHYSICIAN

Submitter: HOSPITAL
Submitter's Address: 100 N AVENUE
City, State, Zip: CITY, KY 42701
Date Reported: Date Collected: 5/29/16

Physician's Phone: 1234567890
Physician's Address: 100 MAIN ST
City, State, Zip: CITY, KY 42701
Test Type: Initial

Disorder:	Analyte:	Results:	Normal Reference Range:	Comments:
Biotinidase Deficiency	Biotinidase	Full Enzyme Activity Detected	Full Enzyme Activity	
Congenital Adrenal Hyperplasia	17-OHP	Normal	Within Normal Limits*	
Congenital Hypothyroidism	T4,TSH	Normal	Within Normal Limits**	
Cystic Fibrosis (CF)	IRT	Normal	Within Normal Limits***	
Galactosemia	Galactose-1-Phosphate Uridyltransferase	Full Enzyme Activity Detected	Full Enzyme Activity	
Hemoglobinopathies	Hemoglobin	Not Requested	Not Tested	
Fatty Acid Oxidation Disorders	Fatty Acids	Not Requested	Not Tested	
Amino Acid Disorders	Amino Acids	Not Requested	Not Tested	
Organic Acid Disorders	Organic Acids	Not Requested	Not Tested	
SCID	TREC	Within Normal Limits	TREC Within Normal Limits	
Lysosomal Storage Disorders	Lysosomal Enzymes	Full Enzyme Activity Detected	Full Enzyme Activity****	

*Congenital Adrenal Hyperplasia-17OHP normal weight-based limits for all initial specimens and repeat specimens on infants less than one week old: <1500g, <70 ng/mL; 1500g-2500g, <40 ng/mL; >2500g, <25 ng/mL. Normal limit for repeat specimens on infants greater than one week old of any weight is <25 ng/mL. Treatment of the mother or the child with steroids may result in false negative results.
**T4- Normal for specimens from infants < 4 weeks of age is 5-27 µg/dL. Normal T4 for specimens from infants > or = 4 weeks of age is 5-19 µg/dL. Normal TSH is <20µU/mL. TSH values below 2.91, the lower limit of linearity for this method, will be reported as <2.91. Rare hypothalamic and pituitary disorders may be causes of fatal hypothyroidism with low TSH and low T4. Neonates born to women with Graves' disease may have fatal hyperthyroidism with low TSH and normal/elevated T4. Recommend clinical correlation and follow up as indicated.
***IRT - Normal for initial specimens from infants < 4 weeks of age is <58 ng/mL. IRT - Normal for initial specimens from infants > or = 4 weeks of age is <50 ng/mL. IRT - Normal for repeat specimens (regardless of age) is <50 ng/mL. Meconium Ileus may result in false negative results.
****Enzyme activities of galactocerebrosidase, acid alpha-glucosidase, and alpha-L-iduronidase. Testing performed at Mayo Medical Laboratories, 3050 Superior Drive NW, Rochester, MN 55901.

-Specimen source: dried blood spots.
-This report contains patient information that must be protected in accordance with the Health Insurance Portability and Accountability Act.
-Analyte Specific Reagent: The Severe Combined Immunodeficiency assay was developed and the performance characteristics determined by Kentucky Division of Laboratory Sevices. It has not been cleared or approved by the U.S. Food And Drug Adminsitration.

Jeremy Hart MD, FCAP
Director - (502) 564-4446

Vaneet Arora MD MPH
Associate Director - (502) 564-4446

TEST HOSPITAL ATTENTION:
NURSERY
100 N AVENUE
CITY, KY 42701

TESTS CONDUCTED:

Enzyme Immunoassay: Congenital Adrenal Hyperplasia (CAH), Congenital Hypothyroidism (CH), Cystic Fibrosis (CF), Galactosemia (GALT), Biotinidase (BIO)

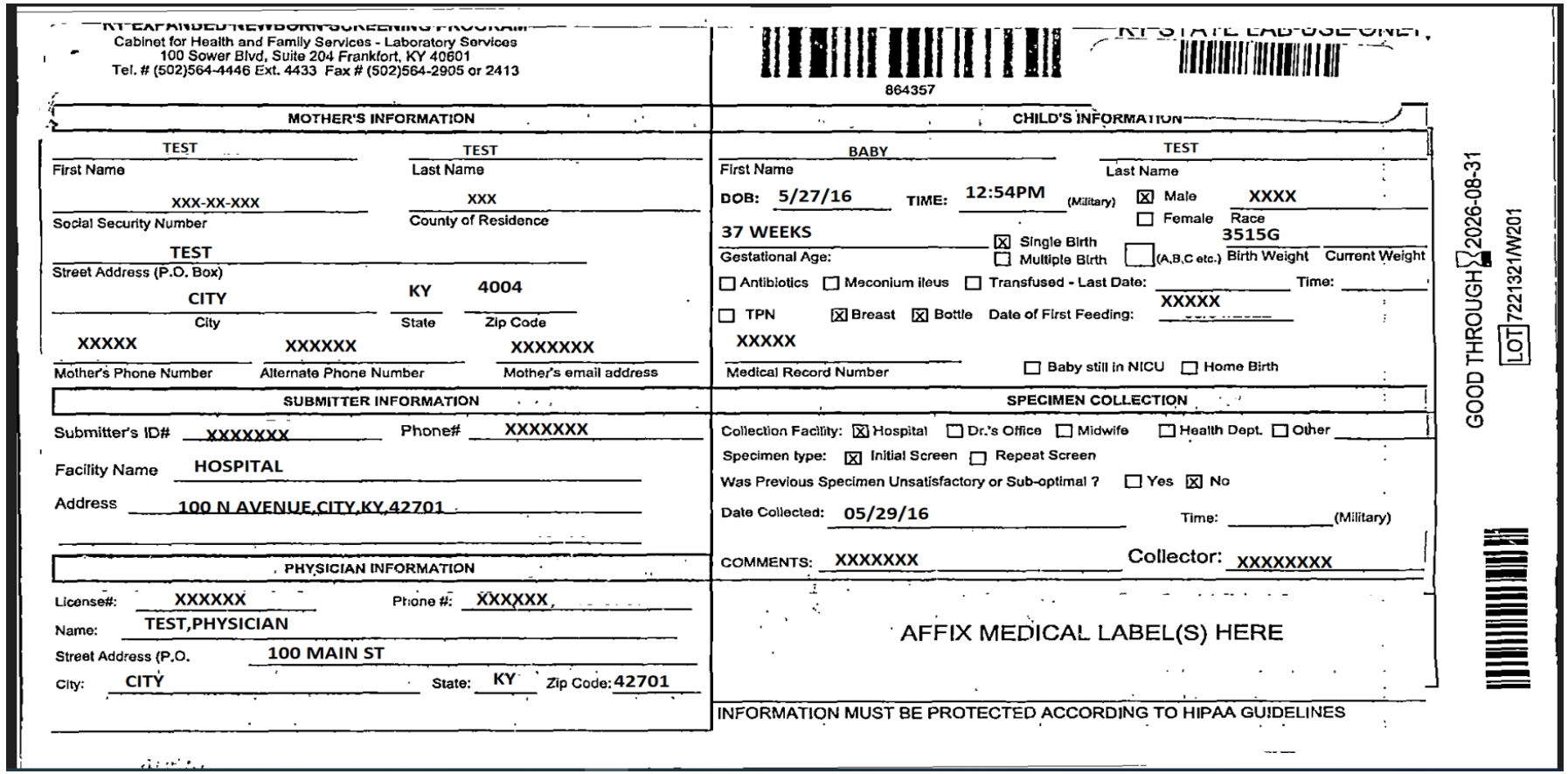
High Performance Liquid Chromatography (HPLC): Hemoglobinopathies

Tandem Mass Spectrometry (MS/MS):
Fatty Acid Oxidation Disorders: Medium-chain acyl-CoA dehydrogenase deficiency (MCADD), Very long-chain acyl-CoA dehydrogenase deficiency (VLCADD), Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD), Trifunctional protein deficiency (TFP), Carnitine uptake defect (CUD), Carnitine acylcarnitine translocase deficiency (CACT), Carnitine palmitoyl transferase I deficiency (CPT-I), Carnitine palmitoyl transferase II deficiency (CPT-II), Glutaric acidemia type II (GA-II), Short-chain acyl-CoA dehydrogenase deficiency (SCADD)
Amino Acid Disorders: Argininosuccinic acidemia (ASA), Citrullinemia Type I (CIT-I), Tyrosinemia Type I (TYR-I), Maple syrup urine disease (MSUD), Homocystinuria (HCY), Phenylketonuria (PKU), Argininemia (arginase deficiency) (ARG), Citrullinemia Type II (CIT-II), Hyperphenylalaninemia (H-PHE), Hypermethioninemia (MET), Tyrosinemia Type II (TYR-II), Tyrosinemia Type III (TYR-III), Nonketotic Hyperglycinemia (NKHG)
Organic Acid Disorders: Beta-ketothiolase deficiency (BKT), Isovaleric acidemia (IVA), Glutaric acidemia Type I (GA-I), 3-Hydroxy-3-methylglutaric aciduria (HMG), Multiple carboxylase deficiency (MCD), 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC), Methylmalonic acidemia (MMA Cbl A, B, C, D), Methylmalonyl-CoA mutase deficiency (MUT), Propionic acidemia (PA), 2-Methyl-3-Hydroxybutyric aciduria (2M3HBA), 3-Methylglutaconic aciduria (3MGA), Isobutyryl-CoA dehydrogenase deficiency (IBD), Malonic acidemia (MAL), Ethylmalonic encephalopathy (EE), 2-Methylbutyryl-CoA dehydrogenase deficiency(2MBDH)

Real-Time Polymerase Chain Reaction (PCR): T-Cell Receptor Excision Circles (TREC) for Severe Combined Immunodeficiency

Flow Injection Analysis-Tandem Mass Spectrometry:
Lysosomal Storage Disorders: Krabbe Disease, Pompe Disease, Mucopolysaccharidosis Type I (MPS I) [Testing performed at Mayo Medical Laboratories in Rochester, MN]

The laboratory values in this report represent screening test results and are intended to identify infants at risk for selected disorders and in need of more definitive testing. The above results should be correlated clinically with consideration of age at the time of collection, nutrition, birth weight, prematurity, health status, and treatments. It is very important for physicians to be aware that a negative screening result does not indicate with certainty the absence of the above listed disorders. The physician should be alert to the clinical symptoms of these conditions, so that diagnosis and treatment can take place as early as possible in infants who are not identified through the newborn screening program.



KY EXPANDED NEWBORN SCREENING PROGRAM
Cabinet for Health and Family Services - Laboratory Services
100 Sower Blvd, Suite 204 Frankfort, KY 40601
Tel. # (502)564-4446 Ext. 4433 Fax # (502)564-2905 or 2413

KY STATE LAB USE ONLY

864357

MOTHER'S INFORMATION

TEST — First Name
TEST — Last Name
XXX-XX-XXX — Social Security Number
XXX — County of Residence
TEST — Street Address (P.O. Box)
CITY — City
KY — State
4004 — Zip Code
XXXXX — Mother's Phone Number
XXXXXX — Alternate Phone Number
XXXXXXX — Mother's email address

SUBMITTER INFORMATION

Submitter's ID# XXXXXXX Phone# XXXXXXX
Facility Name HOSPITAL
Address 100 N AVENUE,CITY,KY,42701

PHYSICIAN INFORMATION

License#: XXXXXX Phone #: XXXXXX
Name: TEST,PHYSICIAN
Street Address (P.O. 100 MAIN ST
City: CITY State: KY Zip Code: 42701

CHILD'S INFORMATION

BABY — First Name
TEST — Last Name
DOB: 5/27/16 TIME: 12:54PM (Military) ☒ Male ☐ Female XXXX — Race
37 WEEKS — Gestational Age: ☒ Single Birth ☐ Multiple Birth ☐ (A,B,C etc.) 3515G — Birth Weight — Current Weight
☐ Antibiotics ☐ Meconium ileus ☐ Transfused - Last Date: Time:
☐ TPN ☒ Breast ☒ Bottle Date of First Feeding: XXXXX
XXXXX — Medical Record Number ☐ Baby still in NICU ☐ Home Birth

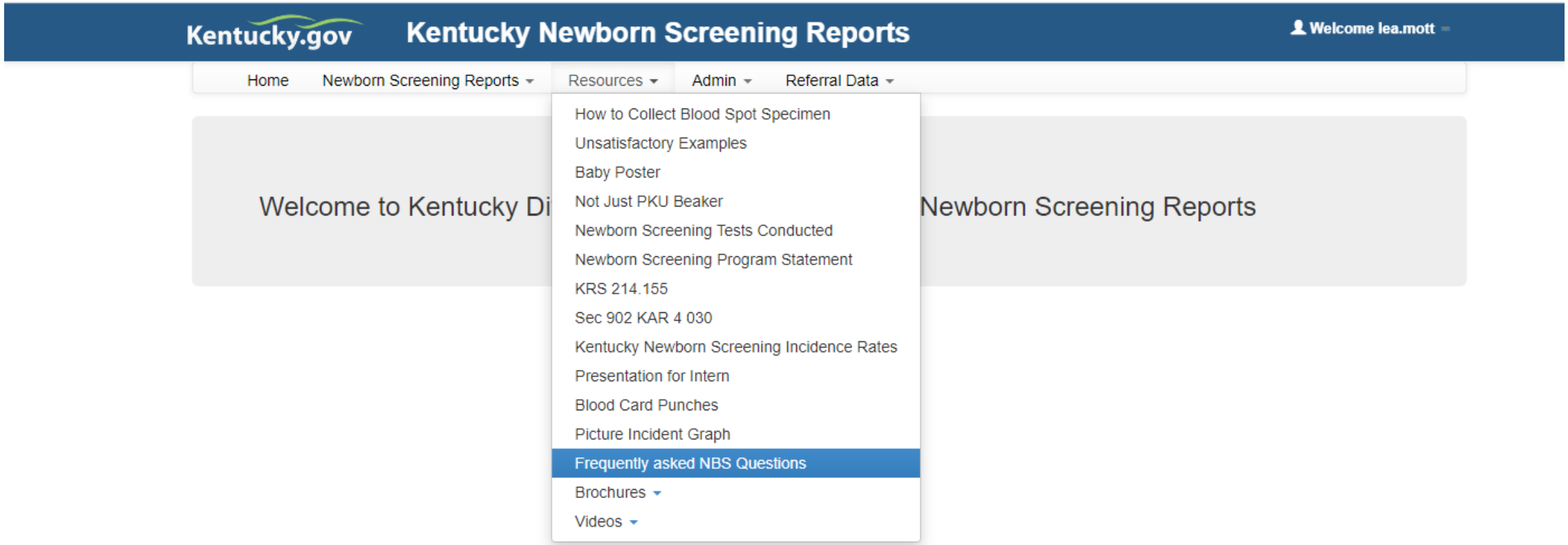
SPECIMEN COLLECTION

Collection Facility: ☒ Hospital ☐ Dr.'s Office ☐ Midwife ☐ Health Dept. ☐ Other
Specimen type: ☒ Initial Screen ☐ Repeat Screen
Was Previous Specimen Unsatisfactory or Sub-optimal ? ☐ Yes ☒ No
Date Collected: 05/29/16 Time: (Military)
COMMENTS: XXXXXXX Collector: XXXXXXXX

AFFIX MEDICAL LABEL(S) HERE

INFORMATION MUST BE PROTECTED ACCORDING TO HIPAA GUIDELINES

GOOD THROUGH 2026-08-31
LOT 7221321/W201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How to Collect Blood Spot Specimen
Unsatisfactory Examples
Baby Poster
Not Just PKU Beaker
Newborn Screening Tests Conducted
Newborn Screening Program Statement
KRS 214.155
Sec 902 KAR 4 030
Kentucky Newborn Screening Incidence Rates
Presentation for Intern
Blood Card Punches
Picture Incident Graph
Frequently asked NBS Questions
Brochures
Videos
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Thank You!
Questions & Comments

Lea Mott,
Newborn Screening
Lea.mott@ky.gov