



Attachment 1

Newborn Screening Program Description

The core mission of Texas' Newborn Screening Program is to save children's lives through the early detection of life-threatening disorders.

Newborn Screening in Texas

- DSHS performs all the Newborn Screening (NBS) tests for Texas
- NBS program screens for congenital and heritable disorders
- NBS consists of specimen collection, testing, reporting results, case management, diagnosis and treatment
- NBS in Texas began in 1963 as a pilot program for the screening of phenylketonuria (PKU)

Newborn Screening in Texas

Texas currently screens for all 29 core panel disorders recommended by the American College of Medical Genetics (ACMG)

- The program expanded from 7 to 27 disorders with passage of HB 790 in 2005
 - Required expansion of NBS program using the ACMG recommended panel as funds allowed
 - Expanded screening began in December 2006
- Babies born in hospitals or birthing centers receive a hearing screen (or are referred for screening)
 - ACMG Core Panel includes hearing screen
- Screening for Cystic Fibrosis was funded by the 81st Legislature
 - Screening began December 1, 2009

Newborn Screening in Texas

- For each of the core diseases, an accurate test is currently available
- Early detection and treatment can lead to improved growth and development, increased life expectancy, and reduced medication, hospitalizations and mortality in children with these disorders

Newborn Screening in Texas

- Each baby born in Texas is required to be screened twice
 - 24 – 48 hours of age or before leaving hospital, in order to detect some disorders at the earliest possible opportunity
 - The second screen at 1 – 2 weeks of age is necessary because some cases may only be detected on the second screen
- There are approximately 400,000 births in Texas each year
- Two screens for each child means ~ 800,000 specimens collected each year

Newborn Screening in Texas

- The NBS program follows-up on approximately 27,000 abnormal screens a year
- ~700 disorders diagnosed each year
- Because every baby is tested soon after birth, any child who may have a screened disorder is identified early and can get early care

Newborn Screening in Texas

- Most children born with these disorders appear healthy at birth
- Serious problems can be prevented if the disorders are caught early
- If treated, infants may live relatively normal lives
- If left untreated, disorders may result in:
 - Poor growth, problems gaining weight
 - Developmental delays such as mental retardation or serious learning disabilities
 - Behavioral/emotional problems such as hitting, biting, hyperactivity
 - Deafness or blindness
 - Seizures
 - Coma,
 - Death

Examples of Screened Disorders

- **Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)**
 - MCAD is a type of fatty acid oxidation disorder. People with MCAD have problems breaking down fat into energy for the body.
 - MCAD can cause bouts of illness called Metabolic Crises. If a Metabolic Crisis is not treated, a child with MCAD can develop breathing problems, seizures, mental retardation, cerebral palsy and coma, sometimes leading to death
 - Children with MCAD should eat often to avoid a metabolic crisis; some benefit from a low-fat, high carbohydrate diet; some are prescribed L-carnitine to help body cells make energy and get rid of harmful wastes
 - 15 cases identified by NBS in 2009

Examples of Screened Disorders

- **Phenylketonuria (PKU)**

- PKU is a type of amino acid disorder. People with PKU have problems breaking down an amino acid called phenylalanine (Phe) from protein in food they eat.
- Babies with PKU seem perfectly normal at birth. The first symptoms are usually seen around 6 months of age. Untreated PKU results in mental retardation, seizures, behavior issues, as well as other problems.
- Children with PKU who start treatment soon after birth usually have normal growth and intelligence.
- Treatment for PKU includes a medical formula with low Phe and a low-Phe food plan.
- 30 cases identified by NBS in 2009

Examples of Screened Disorders

- **Congenital Hypothyroidism (CH)**
 - CH occurs when the thyroid gland doesn't make any or enough hormones. This mainly occurs because the thyroid gland didn't develop, or because it developed in a different place in the body.
 - Synthetic thyroid hormone is prescribed to replace the child's missing thyroid hormone.
 - The effect of hypothyroidism on a baby's mental development is difficult to predict. Starting a hypothyroid baby on treatment as soon as possible after birth is aimed at preventing permanent brain damage and mental retardation.
 - Babies should have normal growth and development when they get their thyroid medicine regularly.
 - 176 cases identified by NBS in 2009

Newborn Screening Collection Form

- The Texas Newborn Screening Collection form, or “kit,” has several tear-off pages
- The version of the Screening Collection forms pictured in this presentation will be available to providers in Summer 2010
- The Disclosure/Destruction request form will be a part of the NBS collection form

NBS Collection Form

The Collection kit contains:

1. Form for parent to take to doctor for second screen when infant is 1-2 weeks old
2. Use and Storage of Newborn Screening Blood Spot Cards Disclosure/Destruction Request form for parents (in English and Spanish)
3. Form for collection of demographic information regarding infant, infant's mother and primary care physician
4. Blood collection filter paper
5. Instructions for provider regarding how to perform newborn screening specimen collection

NBS Collection Form

Parent Copy



TX 10-2000001 X

TX 10-2000001 X

Infant's Last Name Infant's First Name Infant's Date of Birth MM/DD/YYYY

Serial Number TX 10-2000001

Texas Newborn Screening – IMPORTANT – Parent Take to Doctor

Parent,
Take your baby and this form to your baby's doctor when your baby is 1-2 weeks of age. The State of Texas requires that every baby be screened at 1-2 days and again at 1-2 weeks of age. The screen is done on blood taken from a heel stick and tests for a number of rare disorders that can cause mental retardation or death. It is important that you take this form to the baby's doctor so that your infant's first and second screen can be linked. For more information on the Texas Newborn Screen refer to the website listed below. For information on the use and storage of newborn screening specimens see <http://www.dshs.state.tx.us/lab/newbornscreening.shtm>.

Estimados padres:
Lleve a su bebé junto con este formulario al médico de su bebé cuando el bebé tenga de 1 a 2 semanas de edad. El estado de Texas requiere que todos los bebés tengan una revisión médica al 1 ó 2 días de nacidos y otra vez a la edad de 1 a 2 semanas. Se revisa la sangre tomada con un piquete en el talón y se hacen pruebas para detectar varias enfermedades raras que podrían causar retraso mental o la muerte. Es importante que lleve este formulario al médico de su bebé para que relacionen la primera revisión de su niño con la segunda. Si desea más información sobre la Revisión de recién nacidos de Texas consulte el siguiente "website". Para informarse sobre el uso y el almacenamiento de las muestras para las pruebas de detección temprana a los recién nacidos consulte <http://www.dshs.state.tx.us/lab/newbornscreening.shtm> (contenido en inglés).

Texas Department of State Health Services – Newborn Screening Program
P.O. Box 149341, Austin, Texas 78714 - 9341
<http://www.dshs.state.tx.us/lab/newbornscreening.shtm> (800) - 252-8023

Expires 04/02/2013

PARENT COPY

Provider/Submitter Instructions:

Complete Infant's Last Name, First Name and Date of Birth.

For 1st screen:

Remove this PARENT COPY and give to the parent. Inform the parent that they MUST take this form to their infant's doctor at the baby's 1-2 week check-up.

For 2nd screen:

Write the serial number from the 1st screen PARENT COPY (brought to you by the parent) in the box labeled "Previous Specimen Serial Number" in the Newborn Information area of the demographic form for the specimen being submitted.

Retain the PARENT COPY from the 2nd screen in the patient chart. If an additional screen is requested, use this serial number on the additional screen form.

For ALL screens:

Remove page 2 titled "Use and Storage of Newborn Screening Blood Spot Cards" of this kit and give to the parent. Then check the box on the Demographic Information (DSHS copy) page to indicate that page 2 was distributed.

PAID / INSURANCE

NBS Collection Form

Disclosure/Directive to Destroy (English)



X

TX 10-2000001



Use and Storage of Newborn Screening Blood Spot Cards

PARENT / MANAGING CONSERVATOR / LEGAL GUARDIAN
PLEASE READ CAREFULLY

What is newborn screening? The Texas Newborn Screening Program checks Texas babies for a list of serious medical conditions. These conditions can cause death or severe disability. Finding a medical problem during newborn screening can help prevent problems and may save your baby's life.

How does your baby get screened? A small amount of your baby's blood is placed on a special blood spot card. The blood spot card is sent to the state laboratory and tested.

What happens after the blood is tested? After testing, blood spot cards are safely stored by the Texas Department of State Health Services (DSHS) because they still have important public health uses. The main uses are: 1) quality assurance/quality control, such as making sure that testing equipment continues to produce accurate newborn screening test results for Texas babies, and 2) medical research (see Texas Health and Safety Code Sec. 33.017(b)-(c) for a complete list of uses allowed by law). Specific information that could identify your child and connect him/her to a particular blood spot card is not allowed outside of DSHS without permission from the child's parent, managing conservator, or legal guardian unless otherwise provided by law.

You can have your baby's blood sample destroyed if you do not want it to be used after the newborn screening tests are completed. If you are okay with the sample being stored and used as described above, then there are no further steps for you to take.

If you want your baby's blood sample to be destroyed, YOU must fill out ALL of the information on this form and send it back to DSHS at the address given below (DSHS will also accept the form from your healthcare provider, once you have completed and signed the form). If the newborn screening blood spot card is destroyed, the blood sample will not be available for any future needs you may have for the sample.

To Request to have your child's blood spot(s) destroyed:

1. Fill out the entire attached form. Do not leave any fields blank.
2. Mail original to: Texas Department of State Health Services
Newborn Screening Laboratory, MC 1947
PO Box 149347
Austin, Texas 78714-9347
3. For additional information, call 1 (888) 963-7111 ext. 7333 or visit web site:
<http://www.dshs.state.tx.us/lab/newbornscreening.shtm>

DSHS Laboratory Services Section NBS Form F14-13230 - February 2010



Directive to Destroy Newborn Screening Blood Spot Card Following Testing

Fill out this form ONLY if you WANT your baby's blood sample destroyed after newborn testing is complete

I, _____ (please print full name) hereby certify that I am the (check one) parent, managing conservator, or legal guardian of the child named below, and I further certify that there is no court order in effect which restricts my legal ability to make this request.

As parent, managing conservator, or legal guardian, I am telling DSHS to destroy my child's blood spot card(s) after the newborn screen testing is finished.

Full Name of Child: _____

Child's Date of Birth: _____

Full Name of Mother: _____

Contact Information: _____
(Telephone number, e-mail address)

(Mailing address)

(City, State, Zip)

Check here to instruct DSHS to destroy NBS specimens from the child named above.

(Signature)

(Date)

I hereby certify under penalty of law that all the information I have provided herein is true and accurate. I understand that providing false information on this form constitutes a crime in Texas under Penal Code Sec. 37.09.

Provider: Detach this Page and Give to Parent for Review

NBS Collection Form

Demographics Form

INSURANCE
TEXAS DEPARTMENT OF STATE HEALTH SERVICES Laboratory Services Section CLIA#45D0860644
FORM NBS 4 Rev 03/10 Expires 04/02/2013. Telephone # (800) 252-6023 ext. 7318

Newborn Screening

Please read the instructions on the back of this form before starting. USE BLACK INK. PRINT INFORMATION COMPLETELY, ACCURATELY, & LEGIBLY IN BLOCK CAPITAL LETTERS.

DSHS Lab No. For Texas DSHS Use Only

MOTHER INFORMATION

Mother's Last Name _____ Mother's First Name _____
 Maiden Name _____ Social Security # _____
 Mother's Birth Date MMDDYY
 Street Address _____ APT No. _____
 City _____ Zip Code _____ State _____
 Best Phone Number to Reach Mother _____ Newborn Father's Last Name _____

BABY'S PRIMARY CARE PHYSICIAN INFORMATION

Physician Name (Last, First) _____ NPI No. _____
 Street Address _____ Apt. No. _____
 City _____ Zip Code _____ State _____
 Phone No. _____ Fax No. _____

NEWBORN INFORMATION

Newborn's Last Name _____ First Name/Twin A or B _____
 Medical Record No. _____ Birth Order (1-9), if Multiple _____
 Birthweight (grams) _____ Previous Specimen Serial Number _____
 Birth - Date MMDDYY Military Time _____
 Collection - Date MMDDYY Military Time _____

Sex	Ethnicity	Status	Baby's Age at Time of Collection / Test
1. Male <input type="checkbox"/>	1. White <input type="checkbox"/>	0. Normal	1. Less than 7 days old <input type="checkbox"/> 2. 7 days or older 3. Previous Abnormal: Enter Texas DSHS Laboratory No. _____
2. Female <input type="checkbox"/>	2. Af. Amer. <input type="checkbox"/>	1. Sick/Premature	
	3. Hispanic <input type="checkbox"/>	2. On Medications	
1. Breastmilk only <input type="checkbox"/>	4. Asian <input type="checkbox"/>	3. Transfused <input type="checkbox"/>	
2. Formula only <input type="checkbox"/>	5. Am. Indian <input type="checkbox"/>	4. Both 1 & 2 <input type="checkbox"/>	
3. TFN ± Milk <input type="checkbox"/>	6. Other <input type="checkbox"/>	5. Both 1 & 3 <input type="checkbox"/>	
4. Breastmilk & Formula <input type="checkbox"/>		6. Both 2 & 3 <input type="checkbox"/>	
		7. All 1-3 <input type="checkbox"/>	

SUBMITTER INFORMATION

NBS ID No. _____ / NPI No. _____
 Name _____
 Address _____
 City _____ TX Zip Code _____

Check to verify disclosure, and destruction option, distributed.

TX10-2000001 X P
DSHS Copy

10-2000001

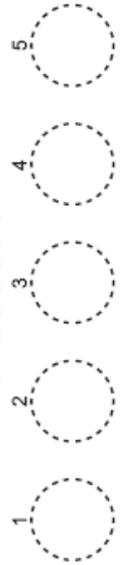
NBS Collection Form

Filter Paper for Blood Collection

SN TX10-2000001 X



EXPIRES 04/02/2013
TX10-2000001
AVOID HANDLING
COLLECTION AREA



TEXAS NEWBORN SCREENING INSTRUCTIONS

A first specimen is required for all newborns at 24-48 hours of age, or just prior to discharge. A second specimen is also required at 1-2 weeks of age.

Invalid results may occur with:

- Infants having received a transfusion.
- Specimens accompanied by improper or incomplete paper work.
- Infants on TPN

DO NOT use expired form.

DO NOT use chlorhexidine gluconate, such as Chlorascrub Swabs

DO NOT use devices that contain EDTA or citrate anticoagulant.

DO NOT use capillary tubes.

DO NOT expose card to heat, moisture, or direct sunlight.

DO NOT stack or mail wet specimens.

DO NOT hold specimens for bulk mailing. Send within 24 hours of collection.

Completion of Form:

1. Legibly and accurately print ALL information in spaces provided. USE ONLY BLACK INK AND BLOCK CAPITAL LETTERS. Press hard for good copies.

2. Enter complete and accurate information.

3. Complete information on top 'PARENT COPY' form, remove and give copy to parent explaining that the form must be taken to the baby's doctor for the baby's 1-2 week check-up.

4. Detach page 2 "Use and Storage of Newborn Screening Blood Spot Cards" and give to parent.

5. Check box on demographic information sheet (DSHS copy) that indicates

"Check to verify disclosure, and destruction option, distributed."

6. For 2nd screens: write serial number on baby's 1st screen 'PARENT COPY' (given to parent at baby's birth) on demographic information sheet in the designated location.

7. Fill in correct number to indicate infant's 'Sex', 'Feed', 'Ethnicity', 'Status' and 'Baby's Age at Time of Collection / Test'.

8. If the specimen is a repeat for a previous abnormal, enter Texas DSHS laboratory number in appropriate spaces.

9. Enter complete submitter's address for location where results are to be sent. Labels for this purpose may be ordered from Texas DSHS, Laboratory Service Section.

10. Remove and keep the submitter 'yellow' copy of the demographic information.

Collection Procedure: Cord blood is not acceptable

1. Place infant/child's limb in dependent position.

2. Cleanse skin with alcohol, DRY, and puncture with disposable lancet.

3. Wipe off first drop of blood.

4. Allow a drop of blood to form and apply DIRECTLY to filter paper. Apply to one side only while viewing from the other side to ensure COMPLETE SATURATION OF THE ENTIRE CIRCLE (see below).

5. Complete one circle at a time and FILL ALL FIVE (5) CIRCLES.

6. Allow card to dry thoroughly in a horizontal position (min 3-4 hours). Do NOT allow specimen to touch any surface.

7. Cover dried specimen with attached flap.

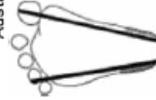
8. Must ship dried specimen WITHIN 24 HOURS in provided envelope to

Texas Department of State Health Services

Laboratory Services Section

P.O. Box 149341

Austin, TX 78714 - 9341



Acceptable

Circle filled and completely saturated



Unacceptable

Caked, clotted, or layered



Insufficient, multiple applications

Collect sample from shaded area



Serum rings present



To order more Newborn Screening Forms, contact

Texas Department of State Health Services

Laboratory Services Section

1-888-963-7111 ext. 7661

Fax: 1-512-458-7672

PAID / INSURANCE

- This form is for the collection of a newborn screening specimen and
- complete the form according to the instructions on the back of this form.
- SPECIMEN REJECTED if NO Date of Collection or NO Newborn's Last Name is provided.
- Do not touch the blood collection area of the form.
- DO NOT remove fold over flap. Cover DRIED blood spots with the flap before mailing.



Blood Spot Collection Process

To do the test, a health care provider takes a few drops of blood from the baby's heel

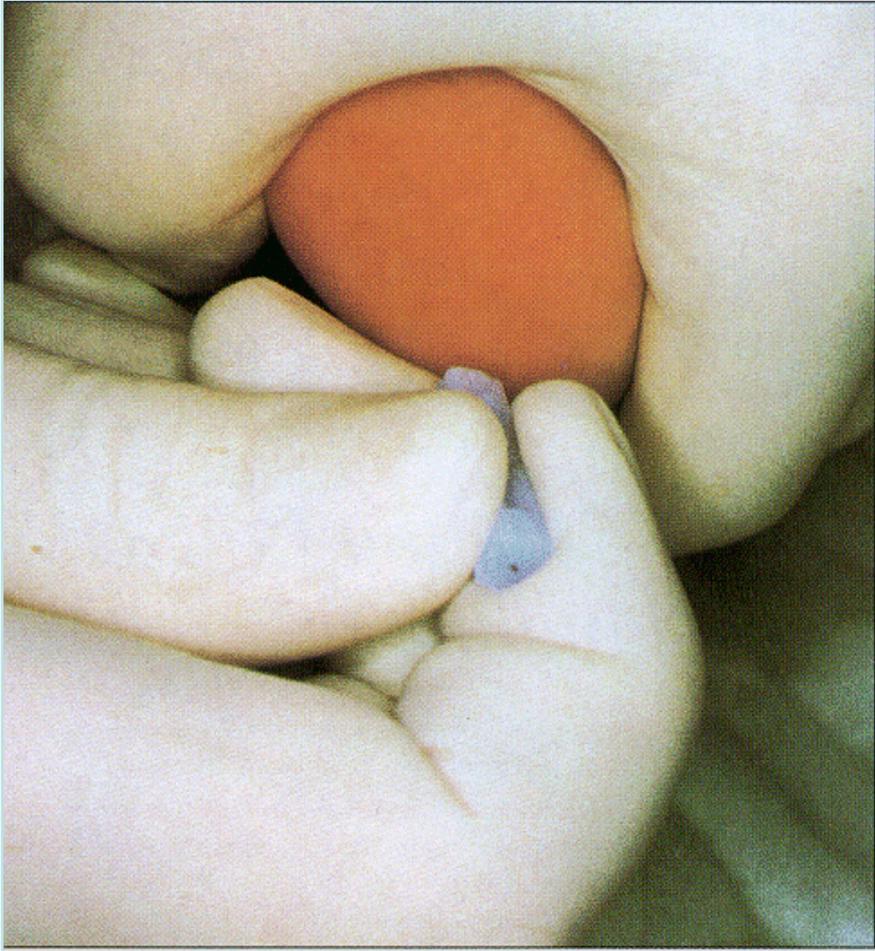


Equipment Used to Collect Blood Sample

- sterile lancet with tip
- sterile alcohol prep
- sterile gauze pads
- soft cloth
- blood collection form
- gloves



Collecting a Blood Sample



The baby's heel is cleansed with a sterile alcohol pad, allowed to dry, then punctured with a sterile lancet

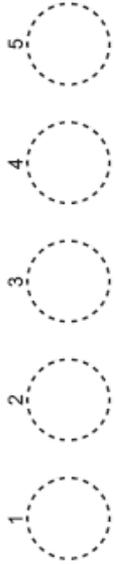
Collecting a Blood Sample

The blood sample is put on a piece of absorbent filter paper, which is part of the NBS blood collection kit

SN TX10-2000001 X

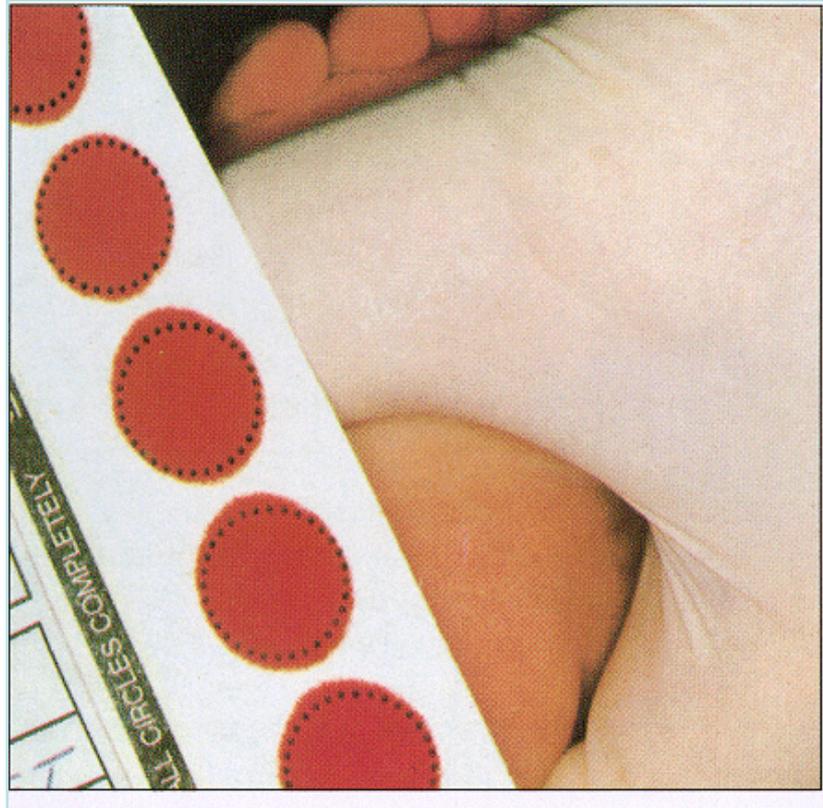


EXPIRES 04/02/2013
TX10-2000001
AVOID HANDLING
COLLECTION AREA



Collecting a Blood Sample

- Blood is allowed to soak through and completely fill all five circles on the paper
- The blood specimens are then allowed to air-dry for at least 4 hours



Laboratory Receives Screening Forms

- Receives and opens NBS envelopes
- Evaluates specimens against acceptance criteria
 - Assigns date of receipt
 - Assigns laboratory identification number
- Processes specimens by type:
 - Newborn: Collected < 7 days after birth
 - Follow-up: Collected ≥ 7 days after birth
 - Requested Repeats: Repeat of Previous Abnormal
 - Unsatisfactory for Testing
- Blood collection cards sent to testing area
- Demographic form sent to data entry area

Lab Specimen Receiving Area



Laboratory Testing Areas

- Technicians punch circular blood samples from blood spot card into plates for specific disorder analysis
- Plates distributed to 6 different lab areas for testing
- Different disorders require different methods for testing
- Specimens are prepared, analyzed and reported

Laser Punching of Specimens



Hemoglobinopathy Screening



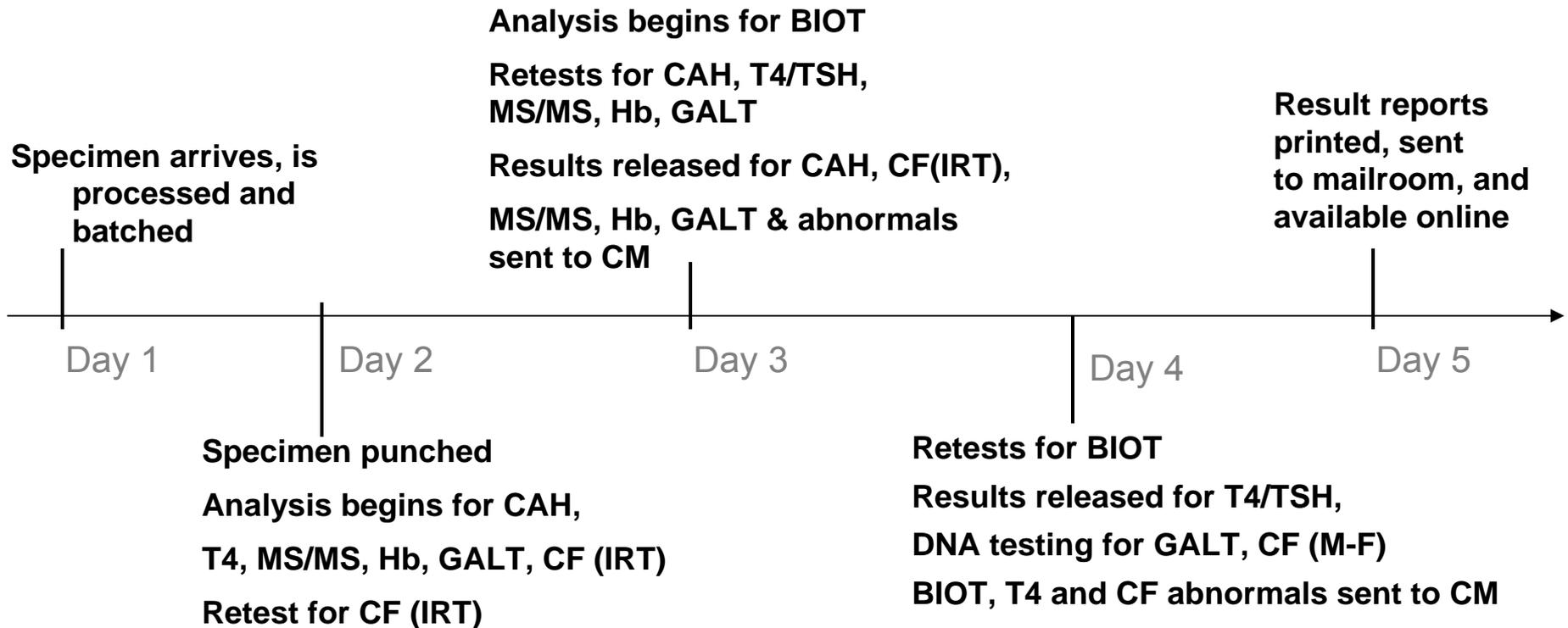
AutoDELFIA Instrument for Hypothyroidism, CAH and Cystic Fibrosis Analysis



Tandem Mass Spectrometer Tests for 20 Disorders



Timeline of a Specimen in the Lab



Normal Screen Results from Lab

- Results are sent to submitter/provider when all tests are final
- Provider notifies family
- The next page shows an example of a report from the Laboratory indicating a **Normal Screen Result**

Report Indicating Normal Screen Results



Texas Department of State Health Services

LABORATORY SERVICES SECTION
 CLIA #45DD650544

MAILING ADDRESS
 PO BOX 149347
 AUSTIN, TEXAS 78714-9347
 1-888-963-7111

CONFIDENTIAL LABORATORY REPORT

SUBMITTER NAME – SUBMITTER ID#
 STREET ADDRESS
 CITY, ST ZIP CODE

Overall Status

NEWBORN SCREENING REPORT

Patient's Name: SMITH TEXAN
 Mother's Name:
 Date of Birth: 04/29/2010
 Medical Record:
 Birth Weight: 2,800 grams
 Race/Ethnicity:
 Sex: Birth Order:
 Feed: BOTTLE
 Status: NORMAL

Laboratory Number: 2010 122 4568
 Form Serial No: 10-4477696
 Date Collected: 04/30/2010
 Date Received: 05/02/2010
 Date Reported:
 Test:
 Mother's SSN:
 Mother's Address: 1100 WEST 49TH
 AUSTIN, TX
 Mother's Telephone:
 Physician's Name:
 Physician's Telephone:

NORMAL SCREEN

Disorder	Screening Result
Amino Acid Disorders	Normal
Fatty Acid Disorders	Normal
Organic Acid Disorders	Normal
Galactosemia	Normal
Biotinidase Deficiency	Normal
Hypothyroidism	Normal
CAH	Normal
Hemoglobinopathies	Normal
Cystic Fibrosis	Normal

Result Table: Results in the table are listed by category of the disorder.

Message clarifying the scope of newborn screening

List of Disorders: Complete listing of disorders screened in each category appearing in the result table.

--The newborn screen identifies newborns at increased risk for specific disorders. The recommended collection period and the test reference ranges have been designed to minimize the number of false negative and false positive results in newborns and young infants. When the newborn screen specimen is collected before 24 hours of age or on older children, the test may not identify some of these conditions. If there is clinical concern, diagnostic testing should be initiated.

*Disorders Screened: AMINO ACID DISORDERS: Argininosuccinic Aciduria (ASA), Citrullinemia (CIT), Homocystinuria (HIC), Maple Syrup Urine Disease (MSUD), Phenylketonuria (PKU), Tyrosinemia type I (TYRI), FATTY ACID DISORDERS: Medium-Chain Acyl-CoA Dehydrogenase Def. (MCAD), Very Long Chain Acyl-CoA Dehydrogenase Def. (VLCAD), Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD), Trifunctional Protein Def. (TFP), Carnitine Uptake Def. (CUD), ORGANIC ACID DISORDERS: Glucose Acidemia I (GA-I), 3-OH 3-Methyl Glucose Acidemia (HMG), Inorganic Acidemia (IVA), Multiple Carboxylase Def. (MCD), 3 Methyl Crotonyl-CoA Carboxylase Def. (3-MCC), Methylmalonic Acidemia (MMA), Propionic Acidemia (PA), Beta-Ketothiolase Def. (BKT), GALACTOSEMIA, BIOTINIDASE DEFICIENCY, CONGENITAL HYPOTHYROIDISM (CH), CONGENITAL ADRENAL HYPERPLASIA (CAH), HEMOGLOBINOPATHIES: including Hb S/S, Hb S/C, Hb S-Beta thalassemia, CYSTIC FIBROSIS (CF).

Abnormal Screening Results from Lab

- Abnormal screen results are reported to the Case Management unit of the Newborn Screening program as soon as available
- The next page shows an example of a report from the Laboratory indicating an **Abnormal Screen Result**

Report Indicating Abnormal Screen Results



Texas Department of State Health Services

LABORATORY SERVICES SECTION
CLIA #45D0660644

CONFIDENTIAL LABORATORY REPORT

MAILING ADDRESS
PO BOX 149347
AUSTIN, TEXAS 78714-9347
1-888-963-7111

SUBMITTER NAME – SUBMITTER ID#
STREET ADDRESS
CITY, ST ZIP CODE

Patient's Name: SMITH TEXAN
Mother's Name:
Date of Birth: 01/10/2010
Medical Record:
Birth Weight: 2,800 grams
Race/Ethnicity:
Sex:
Feed: BOTTLE
Status: NORMAL

NEWBORN SCREENING REPORT
Laboratory Number: 2010 023 4568
Form Serial No: 10-4277896
Date Collected: 01/11/2010
Date Received: 01/23/2010
Date Reported:
Test:
Mother's SSN:
Mother's Address: 1100 WEST 49TH
AUSTIN, TX

Mother's Telephone:
Physician's Name:
Physician's Telephone:

ABNORMAL SCREEN

Disorder	Screening Result	Analyte	Analyte Result
Amino Acid Disorders	Normal		
Fatty Acid Disorders	Normal		
Organic Acid Disorders	Normal		
Galactosemia	Normal		
Biotinidase Deficiency	Abnormal: See Note 1	Biotinidase	Abnormal
Hypothyroidism	Abnormal: See Note 2	T4/TSH	T4 Low, TSH Slightly Elevated
CAH	Normal		
Hemoglobinopathies	Normal		
Cystic Fibrosis	Abnormal: See Note 3	Immunoreactive Trypsinogen CFTR 40 Mutation Panel	Elevated 2 Mutations Detected

Screening Result Notes:

1. Possible Biotinidase Deficiency. Recommend enzyme assay for biotinidase. Refer to a metabolic specialist.
2. Possible Hypothyroidism. Please repeat the newborn screen.

The List of Disorders and Scope of NBS Testing will print on all pages.

The Cystic Fibrosis molecular testing panel consists of 40 mutations and 2 variants in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene and is performed by polymerase chain reaction and detection. The mutation detection rate is estimated to be 30.7-39.12% depending on the patient's ethnicity and the residual risk of carrying a CFTR mutation not included on the panel is approximately 0.4-0.9%. This test was developed and its performance characteristics determined by the Laboratory Services Section at DSHS. The test has not been approved or cleared by the US Food and Drug Administration (FDA). Test results should not be used to diagnose but should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in the interpretation of results may occur if information given to the laboratory is inaccurate or incomplete.

The newborn screen identifies newborns at increased risk for specific disorders. The recommended collection period and the test reference ranges have been designed to minimize the number of false negative and false positive results in newborns and young infants. When the newborn screen specimen is collected before 24 hours of age or on older children, the test may not identify some of these conditions. If there is clinical concern, diagnostic testing should be initiated.

*Disorder Screened: AMINO ACID DISORDERS: Argininosuccinyl Aspartate (ASA), Citrullinemia (CT), Homocystinuria (HCU), Maple Syrup Urine Disease (MSUD), Phenylketonuria (PKU), Tyrosinemia type I (TYR); FATTY ACID DISORDERS: Medium-Chain Acyl-CoA Dehydrogenase Def. (MCAD), Very Long Chain Acyl-CoA Dehydrogenase Def. (VLCAD), Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD), Trifunctional Protein Def. (TFP), Carnitine Uptake Def. (CUD); ORGANIC ACID DISORDERS: Glutamic Acidemia I (GA-I), 3-OH Isovaleric Aciduria (HVA), Isoleucine Acidemia (IVA), Multiple Carboxylase Def. (MCC), Methylcronyl-CoA Carboxylase Def. (MCC), Methylmalonic Acidemia (MMA), Propionic Acidemia (PA), Isovaleric Acidemia (IVA), GALACTOSEMIA, BIOTINIDASE DEFICIENCY, CONGENITAL HYPOTHYROIDISM (CH), CONGENITAL ADRENAL HYPERPLASIA (CAH), HEMOGLOBINOPATHIES: including β S β , β S β , β 0- β thalassaemia, CYSTIC FIBROSIS (CF).

For more information, please refer to <http://www.dshs.state.tx.us/lab/newbornscreening.shtml>

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Texas Department of State Health Services

LABORATORY SERVICES SECTION
CLIA #45D0660644

CONFIDENTIAL LABORATORY REPORT

MAILING ADDRESS
PO BOX 149347
AUSTIN, TEXAS 78714-9347
1-888-963-7111

SUBMITTER NAME – SUBMITTER ID#
STREET ADDRESS
CITY, ST ZIP CODE

Patient's Name: SMITH TEXAN
Date of Birth: 01/10/2010
Medical Record:
Birth Weight: 2,800 grams
Date of Birth: 01/10/2010
Medical Record:
Birth Weight: 2,800 grams

NEWBORN SCREENING REPORT

Laboratory Number: 2010 023 4568
Form Serial No: 10-4277896
Date Collected: 01/11/2010
Date Received: 01/23/2010
Date Reported:

Screening Result Notes Continued:

3. A homozygous Cystic Fibrosis-causing mutation, delta F508, in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were identified. Recommend referral for confirmatory sweat testing and consider genetic counseling.

The Screening Result column indicates if the disorder category tested is Normal, Abnormal, non-specific, Possible TPN, Indeterminate, Inconclusive, or Unsatisfactory.

The Result Table includes an "Analyte" and "Analyte Result" column for Abnormal Screens.

The Analyte column lists which analyte's results were used to determine a non-Normal Screening result.

The Screening Result Notes provide additional information on possible disorders, recommendations for follow-up testing and reasons for unsatisfactory specimens. Notes may continue on Page 2.

The Cystic Fibrosis molecular testing panel consists of 40 mutations and 2 variants in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene and is performed by polymerase chain reaction and detection. The mutation detection rate is estimated to be 30.7-39.12% depending on the patient's ethnicity and the residual risk of carrying a CFTR mutation not included on the panel is approximately 0.4-0.9%. This test was developed and its performance characteristics determined by the Laboratory Services Section at DSHS. The test has not been approved or cleared by the US Food and Drug Administration (FDA). Test results should not be used to diagnose but should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in the interpretation of results may occur if information given to the laboratory is inaccurate or incomplete.

The newborn screen identifies newborns at increased risk for specific disorders. The recommended collection period and the test reference ranges have been designed to minimize the number of false negative and false positive results in newborns and young infants. When the newborn screen specimen is collected before 24 hours of age or on older children, the test may not identify some of these conditions. If there is clinical concern, diagnostic testing should be initiated.

*Disorder Screened: AMINO ACID DISORDERS: Argininosuccinyl Aspartate (ASA), Citrullinemia (CT), Homocystinuria (HCU), Maple Syrup Urine Disease (MSUD), Phenylketonuria (PKU), Tyrosinemia type I (TYR); FATTY ACID DISORDERS: Medium-Chain Acyl-CoA Dehydrogenase Def. (MCAD), Very Long Chain Acyl-CoA Dehydrogenase Def. (VLCAD), Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD), Trifunctional Protein Def. (TFP), Carnitine Uptake Def. (CUD); ORGANIC ACID DISORDERS: Glutamic Acidemia I (GA-I), 3-OH Isovaleric Aciduria (HVA), Isoleucine Acidemia (IVA), Multiple Carboxylase Def. (MCC), Methylcronyl-CoA Carboxylase Def. (MCC), Methylmalonic Acidemia (MMA), Propionic Acidemia (PA), Isovaleric Acidemia (IVA), GALACTOSEMIA, BIOTINIDASE DEFICIENCY, CONGENITAL HYPOTHYROIDISM (CH), CONGENITAL ADRENAL HYPERPLASIA (CAH), HEMOGLOBINOPATHIES: including β S β , β S β , β 0- β thalassaemia, CYSTIC FIBROSIS (CF).

For more information, please refer to <http://www.dshs.state.tx.us/lab/newbornscreening.shtml>

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Case Management

- The primary functions of Newborn Screening case management are:
 1. Notify health care providers and families of abnormal screens;
 2. Facilitate and track linkages between providers and families for timely follow-up care, including confirmation of diagnosis and treatment;
 3. Educate health care providers and affected families about disorders; and
 4. Provide a safety-net benefit package to some families (includes dietary supplements, medications, vitamins, confirmatory testing, and follow-up care).

Case Management

- In cases of very abnormal results, immediate action is taken by CM nurses to contact the doctor and locate the child and family in order to ensure a rapid response for retesting and confirmation of diagnosis
- Case Management sends to provider:
 - Lab results for that disorder
 - “ACT” sheet specific to that disorder to provider
 - ACT sheets contain information such as the diagnosis, condition description, and recommended actions or immediate actions the provider needs to take
 - “FACT” sheet about the disorder for families
 - FACT sheet describes the disorder and treatment
 - List of Specialists for families
- Case Management continues to follow up with provider after diagnosis made to ensure child continues to receive treatment