Texas Department of State Health Services

Biochemistry & Genetics Branch Update

Susan M. Tanksley, PhD

April 18, 2007

TEXAS Department of State Health Services

Biochemistry & Genetics Branch Update

- Clinical Chemistry
  - LIMS implementation
  - Changes to specimen acceptability criteria
    - US Postal System Transit Study
- Newborn Screening
  - Expansion Timeline/Statistics
  - New report format
- Report Cards for Submitters

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Clinical LIMS

- Orchard - Harvest and Copia
  - South Texas Laboratory
  - Women's Health Laboratory
- Reporting in new system began August 2006
- Very near completion
- New feature – web-based data entry and results reporting (Copia)

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Clinical Chemistry Updates

- Stricter acceptance criteria for specimens
  - Hemoglobin Types, Leads & Total Hemoglobin
  - Working with manufacturer to lengthen acceptable timeframe for specimens
  - Potential upcoming changes:
    - Change Hb Types specimen type – filter paper
    - Decrease from 21 days to 7 or 14 days depending on input from manufacturer & review of stability data
  - US Postal System Study

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US Postal System Study

- Goal: To determine transit time using USPS from different areas around Texas to Austin.
- Over 80 healthcare providers throughout Texas agreed to participate.
- Participants to ship a specimen canister containing a short survey for 12 consecutive working days.
- Date/time of arrival in laboratory will be recorded.
- Data analysis
- Problem areas will be investigated

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CLINICAL CHEMISTRY SHIPPING SURVEY - 2007
SUBMITTER / PROVIDER SECTION

Submitter ID/TPI:		DATE:	TIME:
Submitter Name:		How was survey sent?	
		In-house mail drop	
Submitter Address:		Post Office drop box:	
City, State Zip Code		Taken to Post Office	
Person who completed form:		Zip Code of Post Office Used:	

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CLINICAL CHEMISTRY SHIPPING SURVEY - 2007

DSHS USE ONLY ---------- CHECK-IN SURVEY

DATE RECEIVED:		DATE OPENED:	
TIME RECEIVED:		TIME OPENED:	
Business Reply Label:	Old New Other	Was it sent by a courier?	Yes No
Size of Can Used:	Small Medium Large	Was a specimen submitted:	Yes No
Check-In Staff:		What test was requested?	

Newborn Screening Expansion in Texas

- Mandated by House Bill 790, 79th Legislature
- ACMG recommended core panel of 29 disorders as funds allowed
  - Expansion Timeline & Statistics
  - Specimen collection form
  - New report format

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Newborn Screening Expansion: Timeline

- May 23, 2006: Cost-effectiveness Study concluded DSHS Laboratory to continue NBS for Texas
- December 6, 2006: 1st abnormal MS/MS results reported (19 new disorders)
- January 8, 2007: Biotinidase deficiency testing was added
- February 8, 2007: New report implemented to provide results on all 27 disorders

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NBS Expansion: Statistics

- MS/MS (Dec 6 to Mar 31):
  - 126,464 1st screens
  - 123,296 2nd screens
- BIOT (Jan 8 to Mar 31):
  - 93,924 1st screens
  - 91,020 2nd screens

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Confirmed Diagnoses for Expansion Panel

Disorder	Diagnosed cases
Medium Chain Acyl CoA Dehydrogenase Def	5
Tyrosinemia, transient	3
Methylmalonic Acidemia	1
Homocystinuria	2
Glutaric Acidemia Type 1	1
Long Chain Hydroxyacyl-CoA Dehydrogenase Def	1
Maternal 3-Methyl Crotonyl-CoA Carboxylase Def	2
Biotinidase Deficiency	5

Last updated 3/20/07

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New
Report Format

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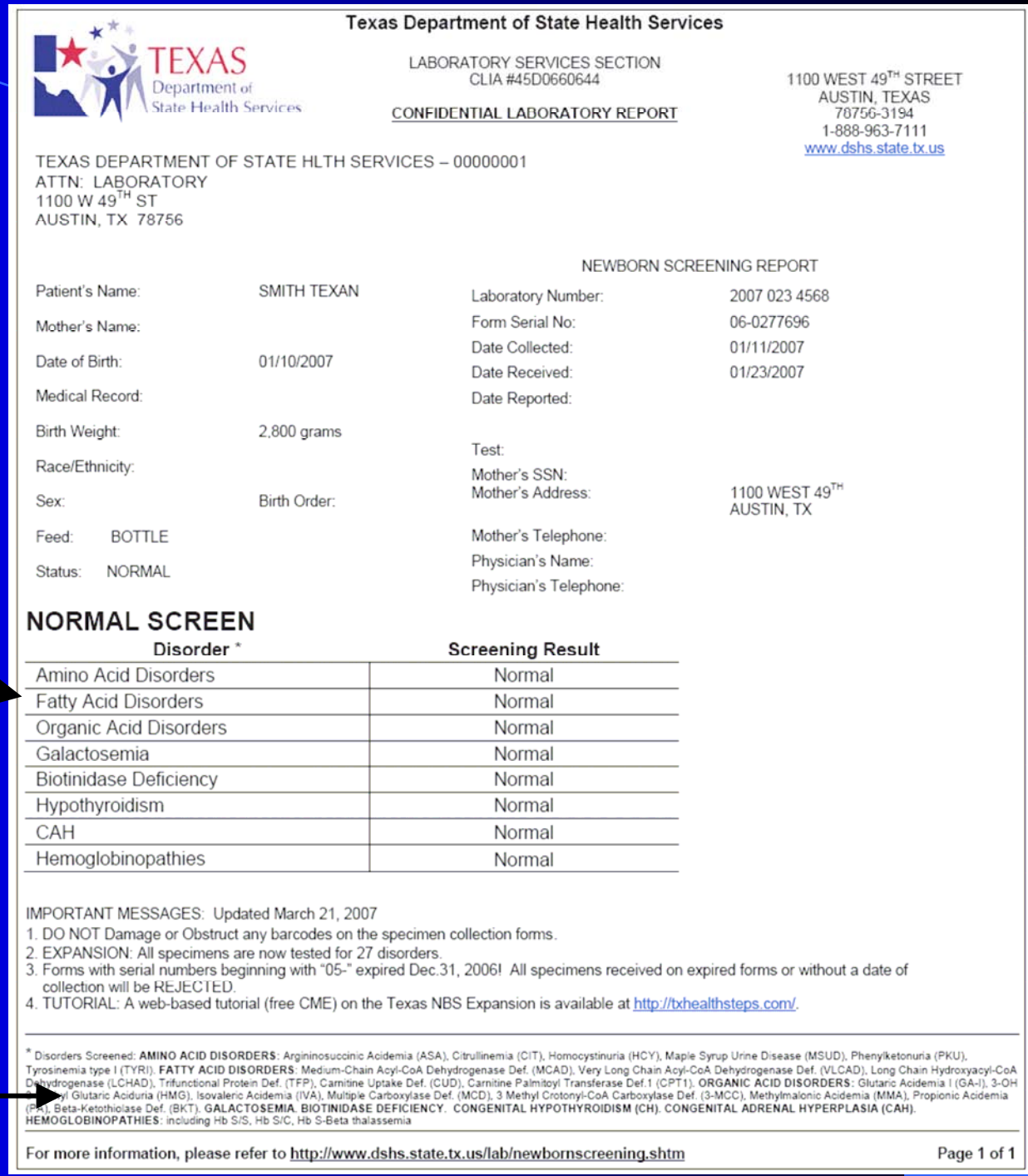
Normal Screen

Groups of disorders listed.

PKU not listed separately.

List of all disorders, arranged by group.



Texas Department of State Health Services

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LABORATORY SERVICES SECTION
CLIA #45D0660644

CONFIDENTIAL LABORATORY REPORT

1100 WEST 49TH STREET
AUSTIN, TEXAS
78756-3194
1-888-963-7111
www.dshs.state.tx.us

TEXAS DEPARTMENT OF STATE HLTH SERVICES – 00000001
ATTN: LABORATORY
1100 W 49TH ST
AUSTIN, TX 78756

NEWBORN SCREENING REPORT

Patient's Name:	SMITH TEXAN	Laboratory Number:	2007 023 4568
Mother's Name:		Form Serial No:	06-0277696
Date of Birth:	01/10/2007	Date Collected:	01/11/2007
Medical Record:		Date Received:	01/23/2007
Birth Weight:	2,800 grams	Date Reported:	
Race/Ethnicity:		Test:	
Sex:	Birth Order:	Mother's SSN:	
Feed: BOTTLE		Mother's Address:	1100 WEST 49TH AUSTIN, TX
Status: NORMAL		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

IMPORTANT MESSAGES: Updated March 21, 2007
1. DO NOT Damage or Obstruct any barcodes on the specimen collection forms.
2. EXPANSION: All specimens are now tested for 27 disorders.
3. Forms with serial numbers beginning with "05-" expired Dec.31, 2006! All specimens received on expired forms or without a date of collection will be REJECTED.
4. TUTORIAL: A web-based tutorial (free CME) on the Texas NBS Expansion is available at http://txhealthsteps.com/.

* Disorders Screened: **AMINO ACID DISORDERS**: Argininosuccinic Acidemia (ASA), Citrullinemia (CIT), Homocystinuria (HCY), 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 (LCHAD), Trifunctional Protein Def. (TFP), Carnitine Uptake Def. (CUD), Carnitine Palmitoyl Transferase Def.1 (CPT1). **ORGANIC ACID DISORDERS**: Glutaric Acidemia I (GA-I), 3-OH 3-Methyl Glutaric Aciduria (HMG), Isovaleric 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

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

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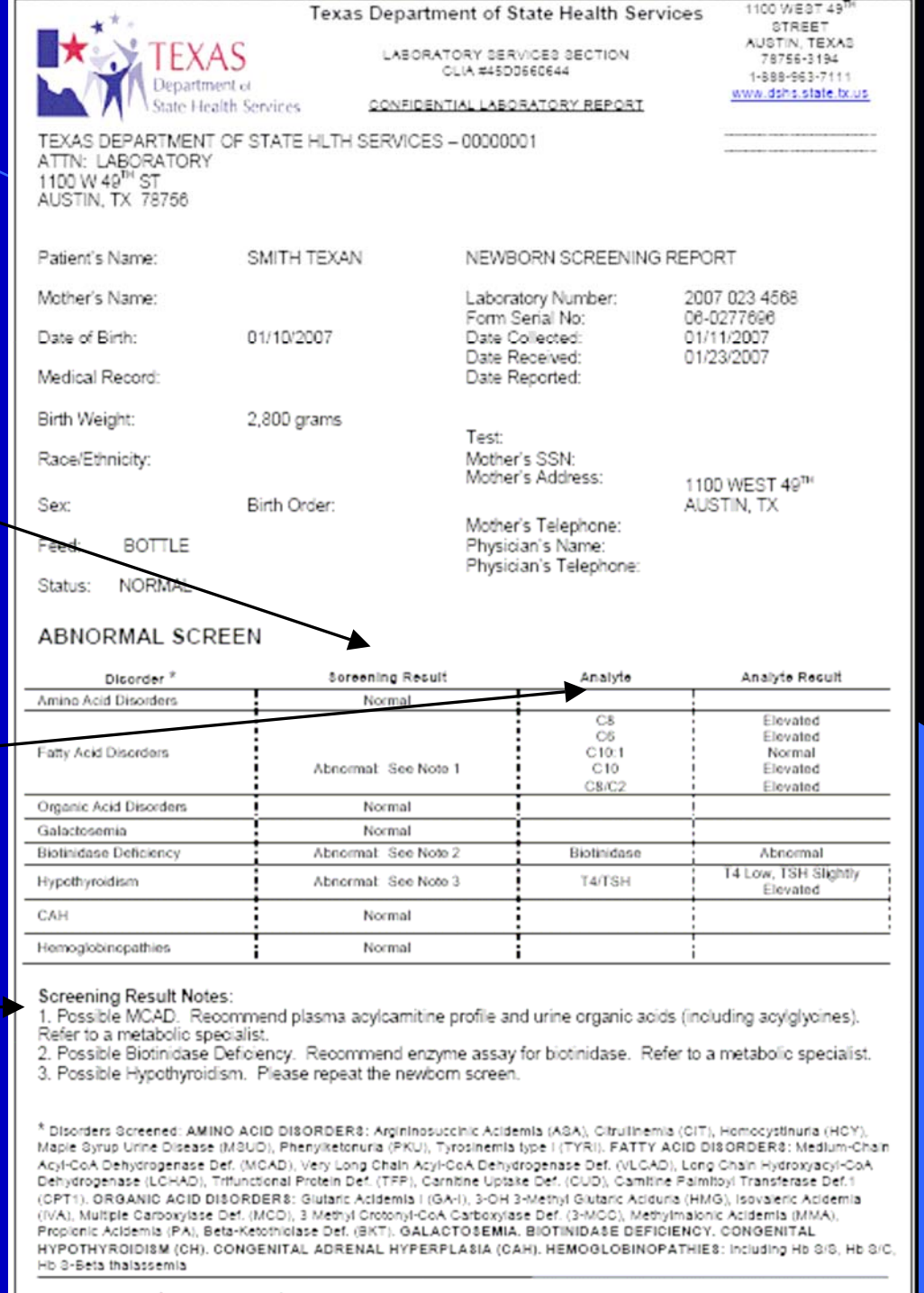
Abnormal Screen Report

The Screening Result column indicates if the disorder category tested is Normal, Abnormal, or Unsatisfactory.

The Analyte column lists analytes that indicate a specific disorder.

The Screening Result Notes provide more information on possible disorders, recommendations for follow-up testing and reasons for unsatisfactory specimens.



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TEXAS DEPARTMENT OF STATE HLTH SERVICES – 00000001
ATTN: LABORATORY
1100 W 49TH ST
AUSTIN, TX 78756

Patient's Name: SMITH TEXAN
Mother's Name:
Date of Birth: 01/10/2007
Medical Record:
Birth Weight: 2,800 grams
Race/Ethnicity:
Sex: Birth Order:
Feed: BOTTLE
Status: NORMAL

NEWBORN SCREENING REPORT

Laboratory Number: 2007 023 4568
Form Serial No: 06-0277696
Date Collected: 01/11/2007
Date Received: 01/23/2007
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	Abnormal See Note 1	C8 C6 C10:1 C10 C8/C2	Elevated Elevated Normal Elevated Elevated
Organic Acid Disorders	Normal		
Galactosemia	Normal		
Biotinidase Deficiency	Abnormal See Note 2	Biotinidase	Abnormal
Hypothyroidism	Abnormal See Note 3	T4/TSH	T4 Low, TSH Slightly Elevated
CAH	Normal		
Hemoglobinopathies	Normal		

Screening Result Notes:
1. Possible MCAD. Recommend plasma acylcarnitine profile and urine organic acids (including acylglycines). Refer to a metabolic specialist.
2. Possible Biotinidase Deficiency. Recommend enzyme assay for biotinidase. Refer to a metabolic specialist.
3. Possible Hypothyroidism. Please repeat the newborn screen.

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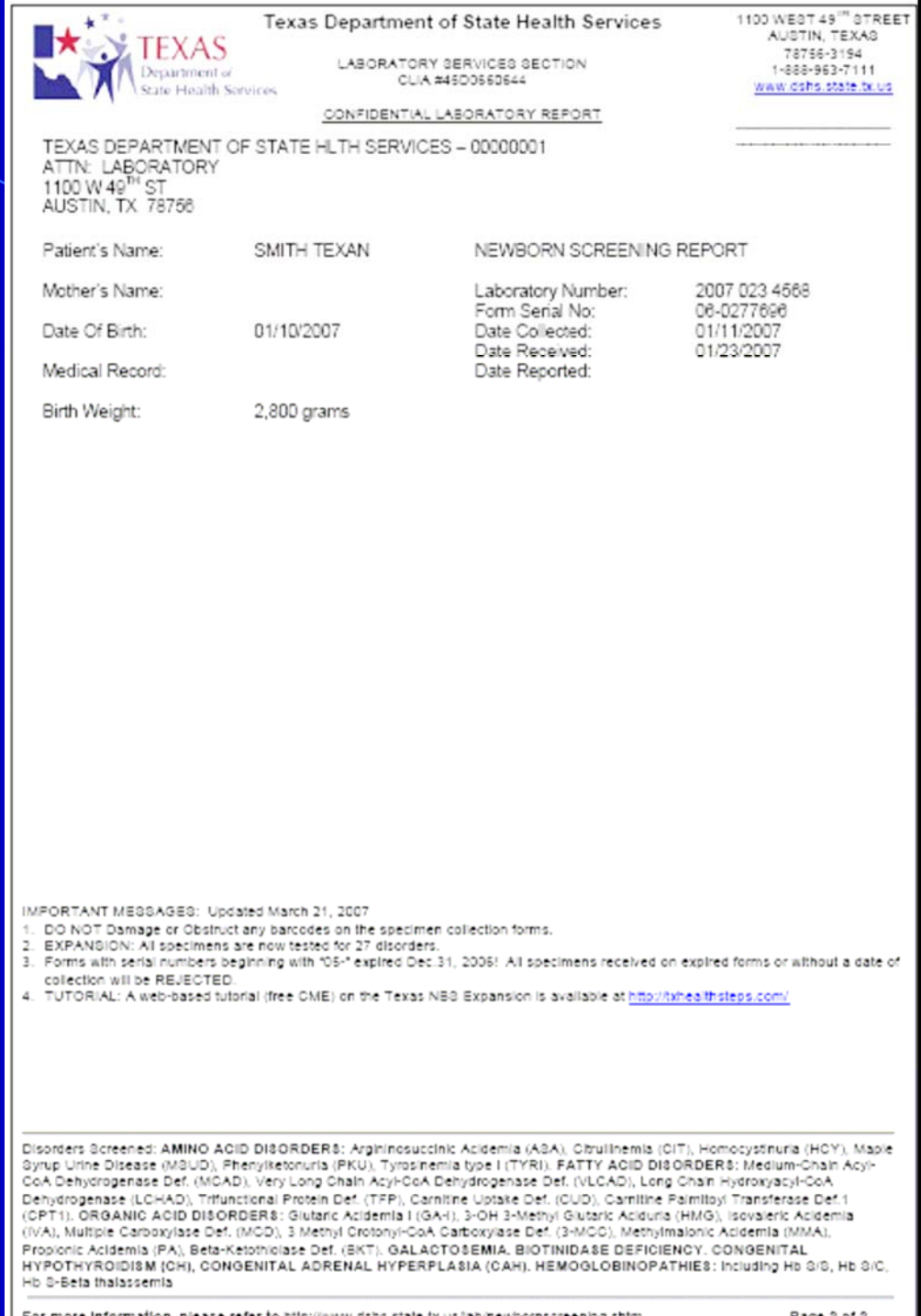
Abnormal Screen Report

Some abnormal reports may have a 2nd page.



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CONFIDENTIAL LABORATORY REPORT

TEXAS DEPARTMENT OF STATE HLTH SERVICES – 00000001
ATTN: LABORATORY
1100 W 49TH ST
AUSTIN, TX 78756

Patient's Name:	SMITH TEXAN	NEWBORN SCREENING REPORT	
Mother's Name:		Laboratory Number:	2007 023 4568
		Form Serial No:	06-0277696
Date Of Birth:	01/10/2007	Date Collected:	01/11/2007
		Date Received:	01/23/2007
Medical Record:		Date Reported:	
Birth Weight:	2,800 grams		

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Upcoming NBS issues

- Update to voice response system to include new disorders
- Enhancement to NBS LIMS to allow web-based patient demographic entry, access to results and file transfers
- Cystic Fibrosis screening
- Evaluation of the utility of the second screen

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Report Cards for Providers

- Summary of specimen quality issues
- Summary of transit time – when specimens are received at DSHS in comparison to when they were collected.
- Newborn Screening
  - Format finalized
  - 2006 report cards to be sent this month
- Clinical Chemistry – in development

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2006 NEWBORN SCREENING REPORT CARD

Submitter ID: 99999999

Submitter Name: NO SUBMITTOR FOUND

Submitter NPI:

SUBMITTER STATISTICS:

Submitter Category: LOW Volume *

Specimen Quality: 1 of 1 Unsatisfactory (100.00%)

Transit Time: 1 of 1 Rejected

Your Rank for Specimen Quality: 1795 of 1795

0-

UNSATISFACTORY SPECIMEN QUALITY (One unsatisfactory specimen may have up to three unsatisfactory codes.)

Month	TOTAL SPECIMENS SUBMITTED	TOTAL NUMBER OF UNSATISFACTORY SPECIMENS	UNSATISFACTORY RATE	BLOOD DID NOT COMPLETELY FILL SPECIMEN CIRCLE	BLOOD DID NOT SOAK THROUGH PAPER - INCOMPLETE SATURATION	FILTER PAPER IS SCRATCHED FROM THE POSSIBLE USE OF CAPILLARY TUBE	SPECIMEN APPEARS CONTAMINATED OR DISCOLORED	BLOOD WAS CAKED, CLOTTED, OR LAYERED ONTO THE FILTER PAPER	ASSAY INTERFERENCE DUE TO EDTA/CITRATE ANTICOAGULANT CONTAMINATION	ASSAY INTERFERENCE	INCOMPLETE ELUTION OF BLOOD FROM FILTER PAPER	PATIENT INFORMATION INCOMPLETE OR INVALID	SERIAL NUMBER OF PATIENT INFORMATION FORM DID NOT MATCH NUMBER ON SPECIMEN FILTER PAPER	SPECIMEN TOO OLD UPON RECEIPT	UNABLE TO ANALYZE SPECIMEN DUE TO LABORATORY ACCIDENT	NO BLOOD SAMPLE RECEIVED WITH REQUISITION	SPECIMEN SUBMITTED ON IMPROPER COLLECTION FORM	SPECIMEN SUBMITTED ON EXPIRED COLLECTION FORM	SPECIMEN REQUIRED REPEAT TEST	SERUM SEPARATION DUE TO IMPROPER DRYING OF SPECIMEN COLLECTION	SPECIMEN DAMAGED DURING TRANSPORT TO LABORATORY	TOTAL NUMBER OF UNSATISFACTORY CODES
06 - JUN	1	1	100.00 %													1						1
Totals:	1	1	100.00%													1						1
Category Totals:	95,973	3,267	3.40%																			
Totals for All Submitters:	776,624	12,634	1.63%																			

TIME FROM COLLECTION TO RECEIPT

	RECEIVED ON DAY 1-3 (Great)	RECEIVED ON DAY 4-5 (Good)	RECEIVED ON DAY 6-7 (Fair)	RECEIVED ON DAY 8-9 (Needs Improvement)	RECEIVED ON DAY 10-13 (Needs Improvement)	RECEIVED ON DAY 14 OR MORE (Rejected)	RECEIVED: MISSING DATE OF COLLECTION (Rejected)	AVERAGE TRANSIT TIME
06 - JUN	0	0	0	0	0	1	0	14.0
Totals:	0	0	0	0	0	1	0	14.0
	0.00%	0.00%	0.00%	0.00%	0.00%	100.00%	0.00%	
Category Totals:	12,671	39,065	32,204	8,100	2,179	606	1,148	5.6
Totals for All Submitters:	120,354	309,970	263,396	58,710	15,430	2,843	5,921	5.6

* Submitter Category Volume is determined by average number of monthly submissions:

HIGH : 150 or more
MEDIUM : 21 - 149
LOW : 20 or less

Any Questions?