Newborn Screening in Texas

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Texas Society of Genetic Counselors
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TEXAS
Department of
State Health Services

BASICS: PURPOSE OF NEWBORN SCREENING

- Program to screen for 55 congenital and heritable disorders.
- Disorders may cause severe intellectual disability, chronic illness, or death with no clinical symptoms.
- Early detection and treatment leads to dramatic positive outcomes for most affected babies.
- Typically treatment through diet control, hormone replacement, and medical supervision.

BASICS: TIMING IS EVERYTHING

- Many disorders may cause irreparable damage in the first days of life.
- Changes in diet and/or other simple interventions can prevent lifelong consequences.
- If an abnormal result occurs, prompt follow-up is critical.
- Accuracy in testing and correct demographic information are essential.

AS OF MAY 2015
TEXAS SCREENS FOR 55 DISORDERS

- 53 rare disorders: Newborn Screening blood spot specimen
- 2 Points of Contact Screens
  - Congenital Hearing Loss
  - Critical Congenital Heart Disease

TEXAS EARLY HEARING DETECTION AND INTERVENTION

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TEXAS NEWBORN HEARING SCREENING

Hearing screening by one of two tests:

- Otoacoustic Emissions (OAE)
- Automated Auditory Brainstem Response (AABR)

TEXAS NEWBORN HEARING SCREENING

Mandated by Texas Senate Bill 793, 83rd Legislative Session

As codified by Health and Safety Code 43.003

- Requires birth facilities to perform directly or through a referral to another program certified under that section, a hearing screening for the identification of hearing loss
- Facilities that must perform Newborn Hearing Screening are:
  - ✓ Hospitals licensed under Chapter 241 offering obstetrical services
  - ✓ Birthing Centers licensed under Chapter 244

TEHDI: 1 – 3 – 6 MONTH PATH

BEFORE 1 MONTH
SCREENING

PASS ← **HEARING SCREEN** at Birth Facility

↓

PASS ← **2ND HEARING SCREEN** at Birth Facility before discharge → Report to Parents, Doctor and TEHDI

↓

REFER

↓

PASS ← **FOLLOW-UP SCREEN** as an outpatient → Report Results & Referrals- TEHDI

↓

REFER

→ Early Childhood Intervention (ECI)

→ Audiological/ Hearing Diagnostic Evaluation

BEFORE 3 MONTHS
CONFIRMATION DIAGNOSIS

TO AUDIOLOGIST: DIAGNOSTIC EVALUATION Using Texas Evaluation Protocol

↓

HEARING LOSS CONFIRMED → Report Results & Referrals- TEHDI

↓

REFERRAL TO EARLY CHILDHOOD INTERVENTION (ECI)

BEFORE 6 MONTHS

ENROLLED IN ECI AND RECEIVING APPROPRIATE EARLY INTERVENTION, HEARING & MEDICAL SERVICES

↓

Report Services – TEHDI

HEARING AID EVALUATION/FITTING WHEN APPROPRIATE

↓

Report Services – TEHDI

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Critical Congenital Heart Disease - CCHD

What is CCHD?

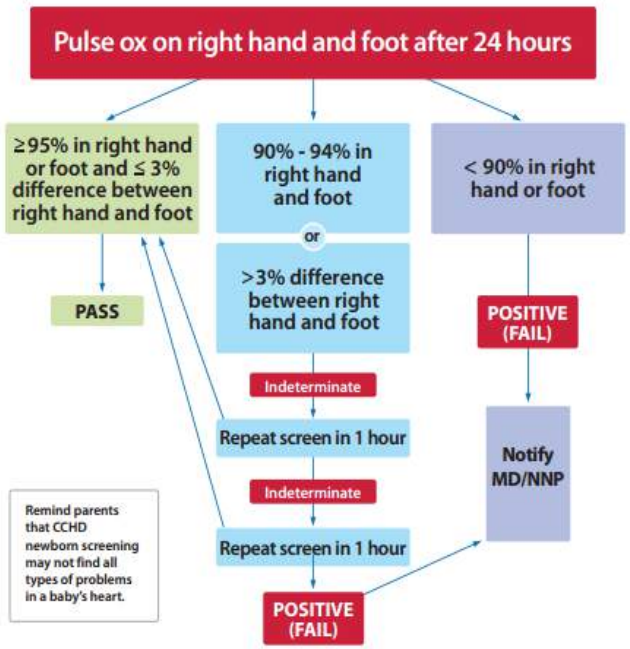
- Heart defects that lead to low oxygen in the newborn and require expert cardiac care and intervention in the immediate Newborn period or early infancy.
- 1 in every 4 babies born with a heart defect has a CCHD
- In the US, about 7,200 babies born every year have CCHD
- Identified using pulse oximetry screening at least 24 hours after birth before leaving the hospital

CCHD Screening

Critical Congenital Heart Disease (CCHD)

- Texas implemented CCHD screening in September 1, 2014
- Point of Contact Testing (at bedside)
- Pulse Oximetry utilized for screening
- Requires reporting of confirmed cases to DSHS
- Birth Defects via active surveillance will confirm number of babies with CCHD (not real time)

Critical Congenital Heart Disease
Newborn Screening Algorithm



A Joint Educational Initiative of
The University of Texas Health Science Center at San Antonio/Department of Pediatrics, Baylor College of Medicine/Department of Pediatrics and Texas Department of State Health Services

texas pulse oximetry project txpop

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Confirmed CCHD in Texas: Sep 2014 - Sep 2016

Primary	
Hypoplastic Left Heart	30
Pulmomary Atresia	13
Tetralogy of Fallot	50
Total Anomalous Pulmonary	30
Transposition of Arteries	55
Tricuspid Atresia	13
Truncus Arteriosus	8
Unspecified Primary	23

Secondary	
Coarctation of Aorta	54
Double Outlet Ventricle	19
Ebstein Anomaly	8
Interrupted Aortic Arch	11
Single Ventricle	7
Unspecified Secondary	38

*Some patients may have more than one condition.

CCHD in Texas: Sep 2014 - Sep 2016

Timeframe	
Post-natal after norm pulse ox	23
Post-natal prior to pulse ox	101
Post-natal with pulse ox	41
Prenatal	7
Prenatal matched to post	109
Prenatal not matched to post	11
Blanks	18
Grand Total	**310**

Treatment	Count	%
Cardiac surgery	154	50%
Medical management	95	31%
Supportive care	21	7%
Blanks	40	12%
Grand Total	**310**	

Ethnicity	Count	%
African American	25	8%
Asian	3	1%
Hispanic	154	50%
Other	9	3%
Blanks	31	10%
White	88	28%
Grand Total	**310**	

Condition	Count	%
Isolated Heart Disease	210	68%
Multiple Anomalies	36	12%
Syndrome/ Chromosomal	38	12%
Blanks	26	8%
Grand Total	**310**	

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Blood Spot Screening

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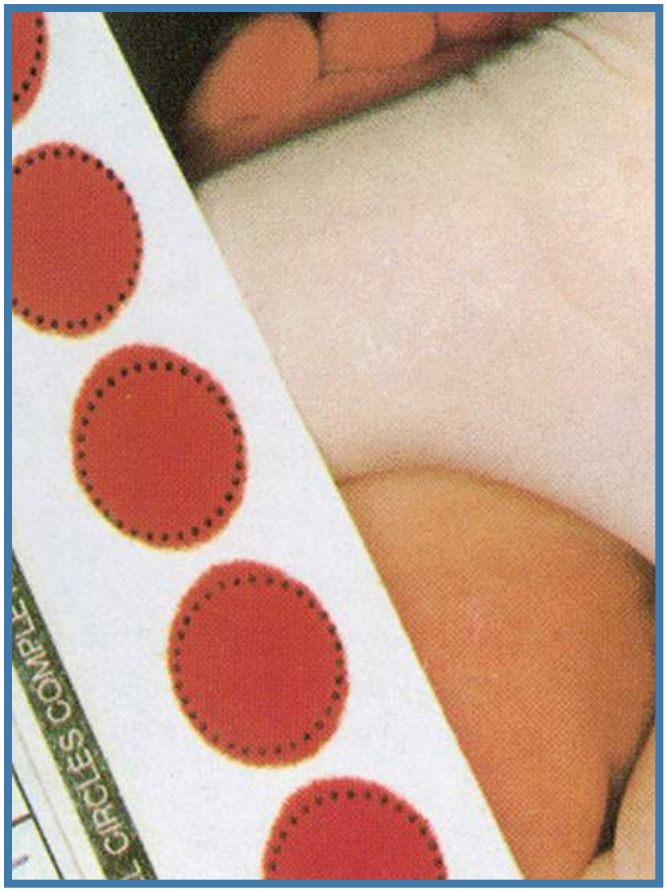
PERCENTAGE OF BIRTHS SCREENED

- Parents can only refuse to have their child screened if the screening conflicts with a parent's religious tenets or practices
- In 2014, 409,111 births were registered in TX and 8,241 (2.0%) were not linked to TX newborn screen database
  - 645 unlinked births were out of state births
  - 901 deaths occurred within 24 hours after birth
  - Were the remaining 6,695 (1.6%) newborns not screened due to parental refusal?

NEWBORN SCREENING PANEL

Currently screen for 53 disorders at the DSHS Lab

- Congenital Hypothyroidism
- Congenital Adrenal Hyperplasia
- 3 primary and various other Hemoglobinopathies
- Galactosemia
- Biotinidase Deficiency
- 14 Amino Acid Disorders
- 13 Fatty Acid Oxidation Disorders
- 15 Organic Acid Disorders
- Cystic Fibrosis
- SCID and T-cell related lymphocyte deficiencies



THE 29 CORE CONDITIONS

PROP	Propionic Acidemia	**CIT**	Citrullinemia, Type I
MUT	Methylmalonic Acidemia (methylmalonyl-CoA mutase)	**MSUD**	Maple Syrup Urine Disease
Cbl A,B	Methylmalonic Acidemia (Cobalamin disorders)	**HCY**	Homocystinuria
IVA	Isovaleric Acidemia	**PKU**	Classic Phenylketonuria
3-MCC	3-Methylcrotonyl-CoA Carboxylase Deficiency	**TYR I**	Tyrosinemia, Type I
HMG	3-Hydroxy-3-Methyglutaric Aciduria	**CH**	Primary Congenital Hypothyroidism
MCD	Holocarboxylase Synthase Deficiency	**CAH**	Congenital adrenal hyperplasia
ßKT	ß-Ketothiolase Deficiency	**Hb SS**	S,S Disease (Sickle Cell Anemia)
GA1	Glutaric Acidemia Type I	**Hb S/ßTh**	S, βeta-Thalassemia
CUD	Carnitine Uptake Defect/Carnitine Transport Defect	**Hb S/C**	S,C Disease
MCAD	Medium-chain Acyl-CoA Dehydrogenase Deficiency	**BIOT**	Biotinidase Deficiency
VLCAD	Very Long-chain Acyl-CoA Dehydrogenase Deficiency	**CF**	Cystic Fibrosis
LCHAD	Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency	**GALT**	Classic Galactosemia
TFP	Trifunctional Protein Deficiency	**SCID**	Severe Combined Immunodeficiencies
ASA	Argininosuccinic Aciduria		

THE 24 SECONDARY CONDITIONS

Cbl C,D	Methylmalonic acidemia with homocystinuria	**CPT II**	Carnitine palmitoyltransferase type II deficiency
MAL	Malonic acidemia	**CACT**	Carnitine acylcarnitine translocase deficiency
IBG	Isobutyrylglycinuria	**ARG**	Argininemia
2MBG	2-Methylbutyrylglycinuria	**CIT II**	Citrullinemia, type II
3MGA	3-Methylglutaconic aciduria	**MET**	Hypermethioninemia
2M3HBA	2-Methyl-3-hydroxybutyric aciduria	**H-PHE**	Benign hyperphenylalaninemia
SCAD	Short-chain acyl-CoA dehydrogenase deficiency	**BIOPT (BS)**	Biopterin defect in cofactor biosynthesis
M/SCHAD	Medium/short-chain L-3-hydroxyacyl- CoA dehydrogenase deficiency	**BIOPT (REG)**	Biopterin defect in cofactor regeneration
GA2	Glutaric acidemia type II	**TYR II**	Tyrosinemia, type II
MCAT	Medium-chain ketoacyl-CoA thiolase deficiency	**TYR III**	Tyrosinemia, type III
DE RED	2,4 Dienoyl-CoA reductase deficiency	**Var Hb**	Various other hemoglobinopathies
CPT IA	Carnitine palmitoyltransferase type I deficiency		T-cell related lymphocyte deficiencies

TIMING

- 1st blood sample is collected at **24 – 48 hours** after birth or before transfusion or discharge, regardless of weight or feeding status.

- 2nd sample is recommended to be collected at **7 – 14 days** of age.

The later a specimen is drawn outside this timeframe, the greater the chance the screen may not identify a disorder.

WHY TWO SCREENING TESTS?

1st Screen

- The tests for certain disorders pick up abnormal levels produced by the stress of birth.
- Abnormal levels for some disorders may normalize by the second screen.
- Early testing may mean the difference between life and death for a patient.

WHY DOES TEXAS LAW MANDATE TWO SCREENING TESTS?

2nd Screen

- Some disorders may be missed on the 1st screen due to infant physiology.
- The second screen is necessary to capture some disorders not picked up on the first screen.
- The CF testing protocol requires two screens.

IT TAKES A TEAM

- NBS Laboratory Services.
- NBS Clinical Care Coordination.
- Medical Providers/Medical Facilities.
- Parents and/or Caregivers.

DSHS NEWBORN SCREENING LABORATORY

- Operates 6 days a week
- Testing processes begin on all specimens within 1 business day of receipt
- Initial (critical) results may be available in as little as 24 hours
- All results reported within 4-5 business days

NBS LABORATORY SERVICES

TEXAS Department of State Health Services

LAB TESTING THE BLOOD SPOTS 2016

- Received 782,187 specimens (~400,000 newborns)
- Specimens Assayed and Reported: 775,084
  - Average 2,573 specimens per day
  - 7,103 unsatisfactory specimens (~0.91%)
- ~17,500 (2.2%) specimens reported with presumptive positive results
- Testing & follow-up performed 6 days a week

NORMAL SCREEN REPORT

TEXAS Department of State Health Services

PHYSICAL ADDRESS
1100 W. 49th St
Austin, TX 78756

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

NURSERIES - BEN TAUB HOSP - 10107422

NEWBORN SCREENING RE

Patient's Name:		Laboratory Number:	
Mother's Name:		Form Serial No:	
Date Of Birth:	07/03/2009	Date Collected:	07/05/2009
Medical Record:		Date Received:	07/08/2009
Birth Weight:	3,770 grams	Date Reported:	07/14/2009
Race/Ethnicity:	HISPANIC		
Sex: MALE	Birth Order:	Test:	1ST TEST (
Feed: Breast		Mother's SSN:	
Status: NORMAL		Mother's Address:	
		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

-- *Demographic information entered through remote data systems.*

The newborn screen identifies newborns at increased risk for specified 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 a clinical concern, diagnostic testing should be initiated.

* 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 Deficiency (LCHAD), Trifunctional Protein Def. (TFP), Carnitine Uptake Def. (CUD). **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.** Cystic Fibrosis (CF)

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

Page 1 of 1

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

ABNORMAL SCREEN REPORT

Disorder *	Screening Result	Analyte	Analyte Result
Amino Acid Disorders	Abnormal: See Note 1	Methionine	Elevated
Fatty Acid Disorders	Abnormal: See Note 2	C8	Borderline
		C6	Normal
		C10:1	Normal
		C10	Elevated
		C8/C2	Normal
Organic Acid Disorders	Normal		
Galactosemia	Normal		
Biotinidase Deficiency	Unsatisfactory: See Note 3		
Hypothyroidism	Abnormal: See Note 4	T4/TSH	T4 Low,TSH Moderately Elevated
CAH	Unsatisfactory: See Note 5		
Hemoglobinopathies	Abnormal: See Note 6	Hemoglobin	A,F,Other
Cystic Fibrosis	[illegible]	[illegible]	[illegible]

Screening Res

1. Possible H
Consult with
2. Borderline
3. Unsatisfac
4. Possible H

Screening Result Notes Continued:

5. Unsatisfactory - Please Resubmit: Patient Information incomplete or invalid (e.g. date of collection missing).

6. Probable Unidentified Hb Variant Trait. Notify family of test results. Recommend consultation with pediatric hematologist.

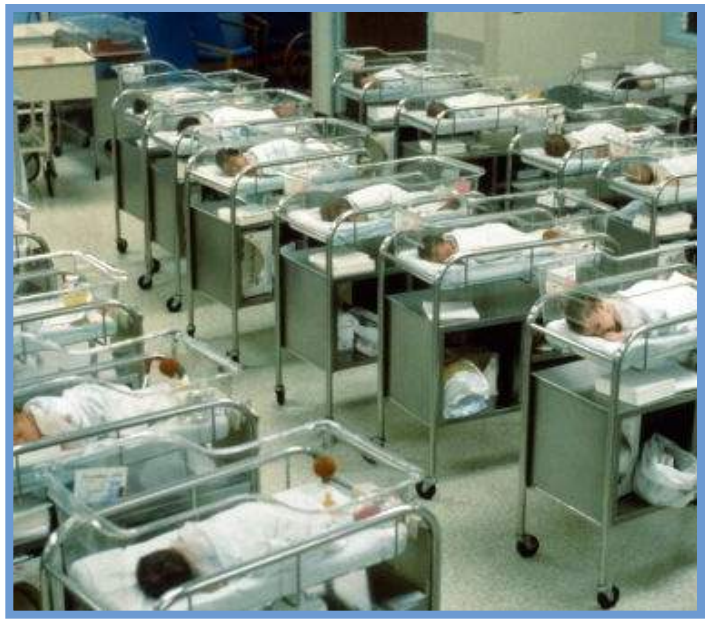
7. Two potential Cystic Fibrosis-causing mutations, ΔF508 and R117H (7T/9T), in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene were identified. Recommend referral for confirmatory sweat testing and consider genetic counseling.

TEXAS NEWBORN SCREENING CLINICAL CARE COORDINATION

TEXAS Department of State Health Services

FINDING THE MEDICAL PROVIDER

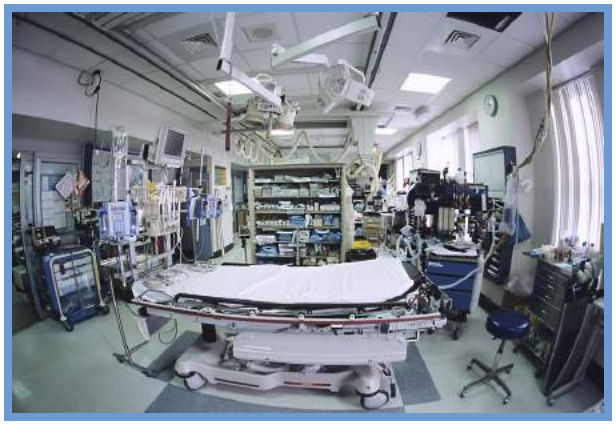
- Find the Medical Provider responsible for the medical care of the baby.
  - Determine if the baby is in the hospital.
- If a Medical Provider <u>can be</u> located:
  - Provide results.
  - Provide guidance for recommended actions.



FINDING THE FAMILY

If a Medical Provider <u>cannot be</u> located:

- Contact parents to obtain Primary Care Provider (PCP) information.
- If a PCP is not identified:
  - Provide results to family.
    - Direct family to an Emergency Department (ED) if necessary.
    - Clinical Care Nurse will coordinate with ED staff if family directed to ED.



WHEN ALL ELSE FAILS

If baby cannot be located:

- Utilize DSHS Regional Social Workers to assist with:
  - Locating the baby.
  - Connecting baby with health-care providers and services.
- Involve other agencies, including law enforcement and/or CPS if necessary.

RESOURCES DISTRIBUTED

Out-of-Range NBS

- Information mailed to parent
- NBS letter
- General NBS Brochures

URGENT FOLLOW-UP

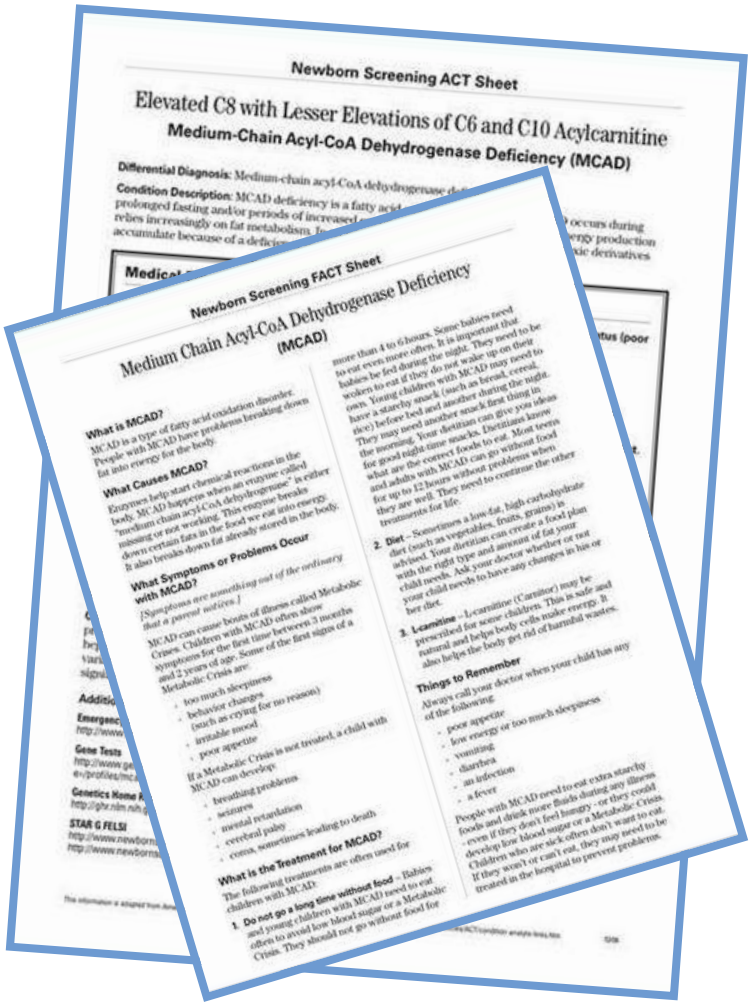
POSITIVE SCREEN WITH VERY ELEVATED LEVELS: MEDICAL EMERGENCY

- Reported immediately to nurses in NBS Clinical Care Coordination.
- Nurse will notify PCP by phone and fax the same day the laboratory results reports are received from the DSHS lab.
- If no PCP is on record for the newborn or cannot be located, the nurse will notify the parents directly.

RESOURCES DISTRIBUTED FOR A NEWBORN REQUIRING URGENT FOLLOW-UP

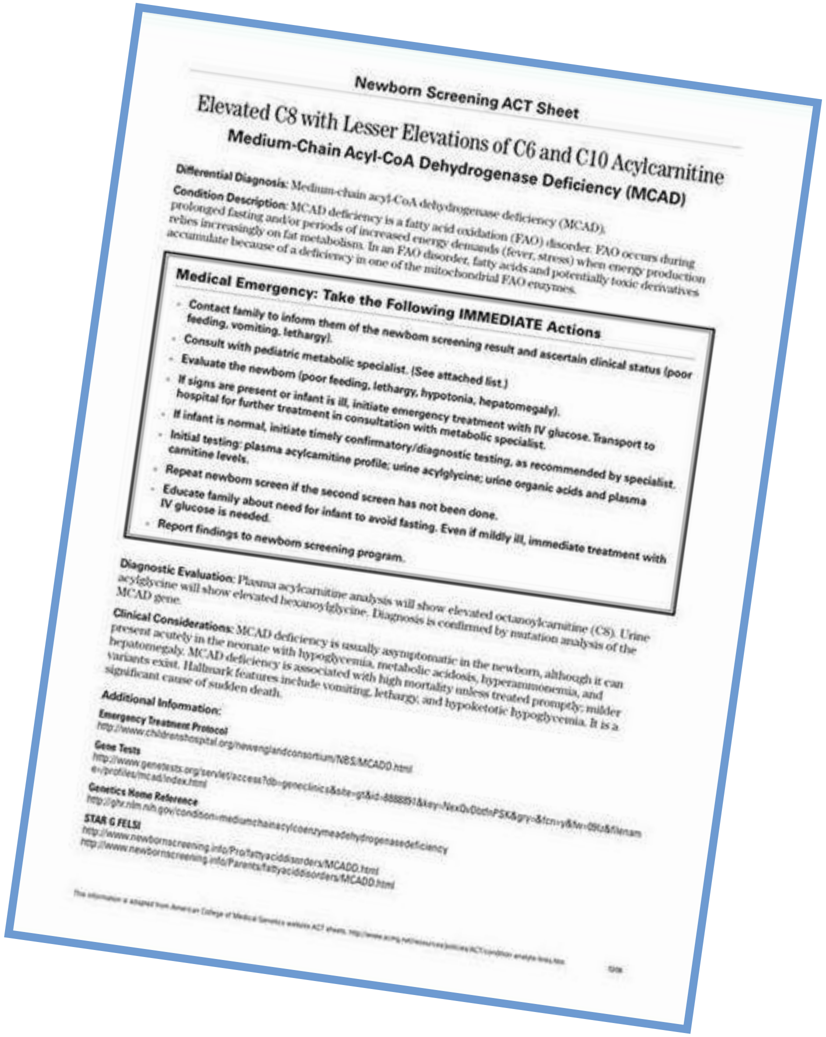
Faxed to Medical Provider

- NBS letter with:
  - NBS disorder-specific lab results.
  - Contact information for the NBS Nurse responsible for the NBS case.
  - Disorder-specific ACT/FACT Sheet.
- List of regional subspecialist consultants.



ACT (ACTION) SHEETS FOR PROVIDERS

- Adapted from the American College of Medical Genetics (ACMG).
- Designed for the medical provider.
- Contain the following:
  - Differential Diagnosis.
  - Condition Description.
  - For medical emergencies, follow the instructions in the black outlined box.
- Available on the NBS Clinical Care Coordination website.



Newborn Screening ACT Sheet

Elevated C8 with Lesser Elevations of C6 and C10 Acylcarnitine

Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)

Differential Diagnosis: Medium-chain acyl-CoA dehydrogenase deficiency (MCAD).

Condition Description: MCAD deficiency is a fatty acid oxidation (FAO) disorder. FAO occurs during prolonged fasting and/or periods of increased energy demands (fever, stress) when energy production relies increasingly on fat metabolism. In an FAO disorder, fatty acids and potentially toxic derivatives accumulate because of a deficiency in one of the mitochondrial FAO enzymes.

Medical Emergency: Take the Following IMMEDIATE Actions

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy).
- Consult with pediatric metabolic specialist. (See attached list.)
- Evaluate the newborn (poor feeding, lethargy, hypotonia, hepatomegaly).
- If signs are present or infant is ill, initiate emergency treatment with IV glucose. Transport to hospital for further treatment in consultation with metabolic specialist.
- If infant is normal, initiate timely confirmatory/diagnostic testing, as recommended by specialist.
- Initial testing: plasma acylcarnitine profile; urine acylglycine; urine organic acids and plasma carnitine levels.
- Repeat newborn screen if the second screen has not been done.
- Educate family about need for infant to avoid fasting. Even if mildly ill, immediate treatment with IV glucose is needed.
- Report findings to newborn screening program.

Diagnostic Evaluation: Plasma acylcarnitine analysis will show elevated octanoylcarnitine (C8). Urine acylglycine will show elevated hexanoylglycine. Diagnosis is confirmed by mutation analysis of the MCAD gene.

Clinical Considerations: MCAD deficiency is usually asymptomatic in the newborn, although it can present acutely in the neonate with hypoglycemia, metabolic acidosis, hyperammonemia, and hepatomegaly. MCAD deficiency is associated with high mortality unless treated promptly; milder variants exist. Hallmark features include vomiting, lethargy, and hypoketotic hypoglycemia. It is a significant cause of sudden death.

Additional Information:

Emergency Treatment Protocol
http://www.childrenshospital.org/newenglandconsortium/NBS/MCADD.html

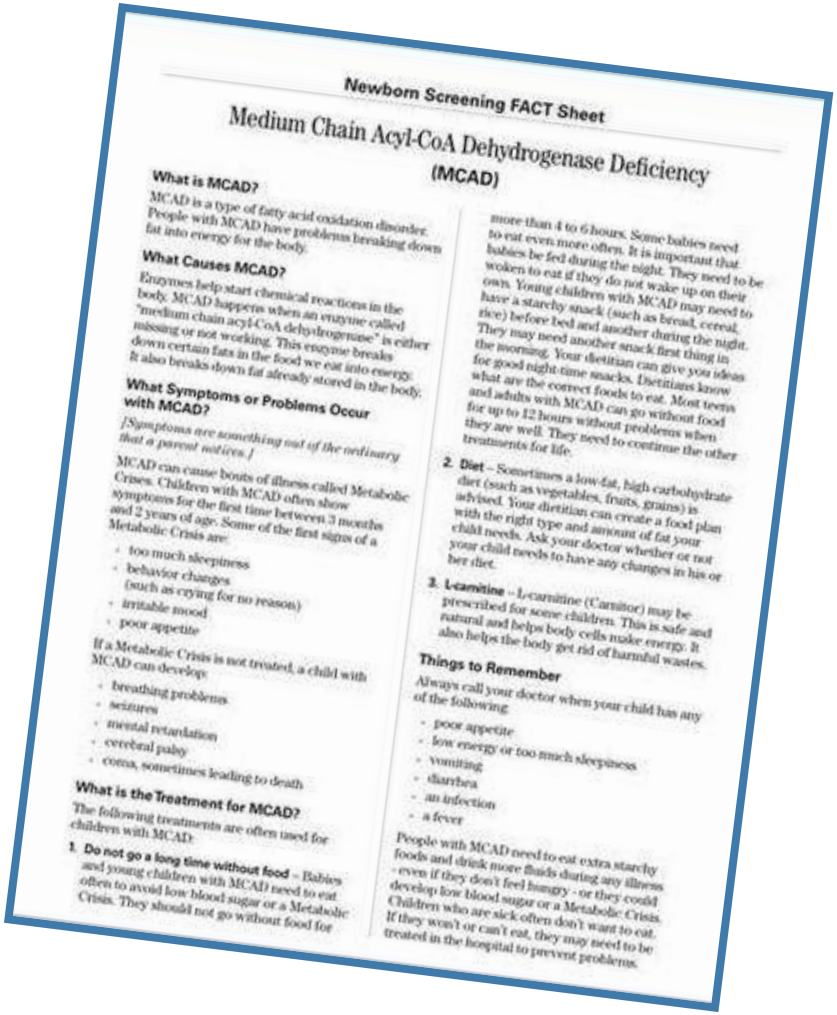
Gene Tests
http://www.genetests.org/servlet/access?db=geneclinics&site=gt&id=[illegible]
e=/profiles/mcad/index.html

Genetics Home Reference
http://ghr.nlm.nih.gov/condition=mediumchainacylcoenzymeadehydrogenasedeficiency

STAR G FELSI
http://www.newbornscreening.info/Pro/fattyaciddisorders/MCADD.html
http://www.newbornscreening.info/Parents/fattyaciddisorders/MCADD.html

FACT SHEETS FOR PARENTS

- Each disorder has a FACT sheet that is modeled from the ACMG Fact Sheet.
- Designed for the PCP to share with the family.
- Information for the parents about symptoms, treatment, and things to remember for the specific disorder.
- Available on the NBS Clinical Care Coordination website.
- Available in English and Spanish.



Newborn Screening FACT Sheet

Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)

What is MCAD?

MCAD is a type of fatty acid oxidation disorder. People with MCAD have problems breaking down fat into energy for the body.

What Causes MCAD?

Enzymes help start chemical reactions in the body. MCAD happens when an enzyme called "medium chain acyl-CoA dehydrogenase" is either missing or not working. This enzyme breaks down certain fats in the food we eat into energy. It also breaks down fat already stored in the body.

What Symptoms or Problems Occur with MCAD?

[Symptoms are something out of the ordinary that a parent notices.]

MCAD can cause bouts of illness called Metabolic Crises. Children with MCAD often show symptoms for the first time between 3 months and 2 years of age. Some of the first signs of a Metabolic Crisis are:

- too much sleepiness
- behavior changes (such as crying for no reason)
- irritable mood
- poor appetite

If a Metabolic Crisis is not treated, a child with MCAD can develop:

- breathing problems
- seizures
- mental retardation
- cerebral palsy
- coma, sometimes leading to death

What is the Treatment for MCAD?

The following treatments are often used for children with MCAD:

1. **Do not go a long time without food** – Babies and young children with MCAD need to eat often to avoid low blood sugar or a Metabolic Crisis. They should not go without food for more than 4 to 6 hours. Some babies need to eat even more often. It is important that babies be fed during the night. They need to be woken to eat if they do not wake up on their own. Young children with MCAD may need to have a starchy snack (such as bread, cereal, rice) before bed and another during the night. They may need another snack first thing in the morning. Your dietitian can give you ideas for good night-time snacks. Dietitians know what are the correct foods to eat. Most teens and adults with MCAD can go without food for up to 12 hours without problems when they are well. They need to continue the other treatments for life.
2. **Diet** – Sometimes a low-fat, high carbohydrate diet (such as vegetables, fruits, grains) is advised. Your dietitian can create a food plan with the right type and amount of fat your child needs. Ask your doctor whether or not your child needs to have any changes in his or her diet.
3. **L-carnitine** – L-carnitine (Carnitor) may be prescribed for some children. This is safe and natural and helps body cells make energy. It also helps the body get rid of harmful wastes.

Things to Remember

Always call your doctor when your child has any of the following:

- poor appetite
- low energy or too much sleepiness
- vomiting
- diarrhea
- an infection
- a fever

People with MCAD need to eat extra starchy foods and drink more fluids during any illness - even if they don't feel hungry - or they could develop low blood sugar or a Metabolic Crisis. Children who are sick often don't want to eat. If they won't or can't eat, they may need to be treated in the hospital to prevent problems.

MEDICAL PROVIDERS AND FACILITIES

TEXAS Department of State Health Services

BIRTH FACILITY AND PRIMARY CARE PROVIDER RESPONSIBILITIES

Birth Hospital:

- Assist with locating baby if needed.
- Identify PCP for infant.

PCP:

- Agree to follow-up with newborn/family.
- Agree to accept patient into practice.
- Refer to subspecialists as appropriate.

PARENTS AND CAREGIVERS

TEXAS Department of State Health Services

PARENTAL RESPONSIBILITIES

- Parent provides PCP information to Clinical Coordination Staff.
- If the newborn does not have a PCP:
  - Parent is asked to identify a PCP.
  - Take infant to ED if necessary.
- Parent must follow-up to ensure newborn:
  - Attends appointments.
  - Receives treatment and care if diagnosed.

LONG TERM FOLLOW-UP

Goal: To ensure the best possible outcome for individuals with disorders identified through newborn screening.

Components:

1. Care coordination through a medical home.
2. Evidence-based treatment.
3. Continuous quality improvement.
4. New knowledge discovery.

LONG TERM FOLLOW-UP

What is Involved?

- Continuing PCP/specialist visits.
- Continuing documentation of treatment.
- Parental involvement.
- Physician/specialist participation.

How long is a child in long term follow-up?

- Begins when an infant receives a confirmatory diagnosis.
- Continues until child is 4-18 years old, depending on the disorder.

LONG TERM FOLLOW-UP

Why Track Long Term?

- Evaluate effectiveness of the NBS Program.
- Develop evidence-based treatment.
- Improve treatment of affected individuals.
- Provide continuous quality improvement.

2015 Diagnosed Cases

Disorder	Diagnosed Cases
Biotinidase deficiency	39
Congenital Adrenal Hyperplasia	87
Cystic Fibrosis	66
Galactosemia	7
Hypothyroidism	283
Sickling Hemoglobinopaties	175
Non-Sickling Hemoglobinopaties	28
Metabolic Disorders	119
Severe Combined Immune Def.	5
Secondary T-Cell Lymphopenias	118
	927

NBS Fee Increase

In order to recover costs for the current testing and follow-up, an increase of the newborn screening fee was needed.

- Old fee - $33.60
  - Established 10/10/2012
  - 10th lowest NBS fee in US
- New fee - $55.24
  - Effective date 10/1/2016
  - 12th lowest NBS fee in US

New Conditions Added onto the RUSP

The Recommended Uniform Screening Panel (RUSP) added:

- Pompe:
  - Approved for addition in March 2015
- Mucopolysaccharidosis Type I (MPS1)
  - Approved for addition in February 2016
- X-linked Adrenoleukodystrophy (X-ALD)
  - Approved for addition in February 2016

Timeliness Recommendations: Overall

To achieve the goals of timely diagnosis and treatment of screened conditions and to avoid associated disability, morbidity and mortality, the following time frames should be achieved by NBS systems for the initial NBS specimen:

- Presumptive positive results for time-critical conditions should be communicated immediately to the newborn's healthcare provider but no later than five days of life.
- Presumptive positive results for all other conditions should be communicated to the newborn's healthcare provider as soon as possible but no later than seven days of life.
- All NBS tests should be completed within seven days of life with results reported to the healthcare provider as soon as possible.

ACHDNC Timeliness Recommendations: Pre-analytical

In order to achieve these goals:

- Initial NBS specimens should be collected in the appropriate time frame for the newborn's condition but no later than 48 hours after birth, and
- NBS specimens should be received at the laboratory as soon as possible; ideally within 24 hours of collection.

Timeliness: Ongoing Activities

- Work with stakeholders to increase awareness
- Submitter Monthly Report Cards
- Spotlight Recognition
- Letter to Birth Hospital CEO's
- Online quarterly report on submitter performance
- NewSTEPs 360 Project
- Improve courier use and efficiency
- Improve turnaround time in the lab

Courier Services

- Funded by DSHS with focus on first screen specimens
- Lone Star Delivery and Processing
  - Hospitals, Pediatric Clinics
  - 530 NBS submitters
  - 69% of NBS specimens (85% of 1st screens)
  - Pick-up Sun – Fri, deliver Mon - Sat
- FedEx
  - 150 NBS submitters
  - 13.8% of specimens (14.5% of 1st screens)
  - Pick-up and deliver Mon - Sat
- Want to add more NBS submitters to FedEx

NBS BENEFITS PROGRAM

TEXAS Department of State Health Services

WHAT IS THE NBS BENEFITS PROGRAM?

- Targets families *without* Medicaid or private insurance.

WHAT ARE THE NBS BENEFITS FOR PATIENTS?

- Confirmatory testing.
- Dietary supplements.
- Metabolic foods.
- Low-protein foods.
- Medications.
- Vitamins.
- Follow-up care.

WHO IS ELIGIBLE FOR NBS BENEFITS?

- Texas Resident
- Those with a presumed positive screen or a confirmed diagnosis of a disorder screened for in the Texas Newborn Screening Program.
- An income at or below 350% of the federal poverty income level (FPL).

NBS Educational Efforts

- Newborn Screening Grand Rounds
  - Anticipated Winter/Spring 2017
  - Dr. Richard Parad and Dr. Donna Beth Willey Courand (Cystic Fibrosis)
  - Dr. Kathy Hassell (Hemoglobinopathies)
- Newborn Screening Journal Club
- Tales from the Crib -NBS Morbid & Mortality
- Educational Outreach
  - External Grand Rounds
  - General NBS presentation
  - Webinar General NBS Grand Round
  - Education Positions are now filled

NBS/Genetics Educational Efforts

DSHS Funds:

- Yearly State of the Art Genetics Conferences-designed for primary care providers
- Baylor Seminars with Genetics-community based genetic seminars
    - Collaborative project with UT Austin Center for Disability Studies (TCDS)
- Teratogen Information Program
- Clinical genetics medical student summer internships

NBS Educational Efforts

Texas Health Steps Modules-CME accredited Provider Education

- Newborn Screening
- Sickle Cell Disease and Trait
- Critical Congenital Heart Disease
- Newborn Hearing Screening
- Genetic Screening, Testing, Treatment and Referral

RESOURCES

Newborn Screening Laboratory

http://www.dshs.state.tx.us/lab/newbornscreening.shtm

NewbornScreeningLab@dshs.state.tx.us

1-888-963-7111 ext. 7333

Clinical Care Coordination

www.dshs.state.tx.us/newborn

Newborn@dshs.state.tx.us

1-888-963-7111 ext. 3957

NBS Benefits Program

www.dshs.state.tx.us/newborn/benefits.aspx

1-800-252-8023 ext. 2983

TEXAS Department of State Health Services

Questions?