Positive Newborn Screens: What do you do next?

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Objectives

• Understand current newborn screening
• Traditional vs. Expanded Screening
• Know how to interpret an expanded newborn screen
• Know how to organize the confirmatory testing
• Know some of the limitations to screening
• Improve your comfort in addressing the family's concerns
Q: Why do Non-selective Screening of Newborns?

- These conditions are important public health issues
  - ‘Reasonable’ prevalence
  - Cost to society if not screened or treated
    - Monetary
    - Resources
- Prevention of irreversible disease manifestations
  - mental retardation, disability or death
- Prompt institution of therapy improves outcome
  - Treatment is less expensive than the care of the untreated individual
Texas Expansion

• HB 790 - three years ago
  – To the ACMG list of 29 (or an equivalent)
• Started December 2006
• Newer technology
• Now DSHS screens for 27 disorders
  – Hearing screening is at birth institution
  – Cystic fibrosis not yet included
ACMG/March of Dimes
List of 29 Disorders for NBS

Prior to 2006 Texas Screened for:

- 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)
- 3-OH 3-CH3 glutaric aciduria (HMG)
- Argininosuccinic acidemia (ASA)
- Beta-ketothiolase deficiency (BKT)
- Biotinidase deficiency (BIOT)
- Carnitine uptake defect (CUD)
- Citrullinemia (CIT)
- **Hearing loss**
- Homocystinuria (HCY)
- Isovaleric academia (IVA)
- Long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHAD)
- Maple syrup urine disease (MSUD)
- Medium chain acyl-CoA dehydrogenase deficiency (MCAD)
- Methylmalonic acidemia (Cbl A,B)
- Methylmalonic acidemia (mutase deficiency) (MUT)
- Multiple carboxylase deficiency (MCD)
- Phenylketonuria (PKU)
- Propionic acidemia (PROP)
- Sickle cell anemia (SCA)
- Trifunctional protein deficiency (TFP)
- Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)
- Tyrosinemia type I (TYR I)
New Disorder Groupings

- **Expanded group of** Amino Acid Diseases
  - Phenylketonuria
  - Argininosuccinic acidemia (ASA)
  - Citrullinemia (CIT)
  - Homocystinuria (HCY)
  - Tyrosinemia type I (TYR I)
  - Maple syrup urine disease (MSUD)

- **New group of** Organic Acid Disorders

- **New group of** Fatty Acid Disorders

- **New / expanded group of** Miscellaneous Other Screened Disorders
Organic acid diseases

- 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)
- 3-hydroxy-3-methylglutaric aciduria (HMG)
- Beta-ketothiolase deficiency (BKT)
- Glutaric acidemia type I (GA I)
- Isovaleric acidemia (IVA)
- Methylmalonic acidemia (Cbl A,B)
- Methylmalonic acidemia (mutase deficiency) (MUT)
- Multiple carboxylase deficiency (MCD)
- Propionic acidemia (PROP)
Fatty Acid Oxidation Disorders

- Carnitine uptake defect (CUD)
- Long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHAD)
- Medium chain acyl-CoA dehydrogenase deficiency (MCAD)
- Trifunctional protein deficiency (TFP)
- Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)
New Technology

• Tandem Mass Spectrometry (MS/MS)
  – Two mass spectrometers in line
  – Separates and quantitates compounds
  – Detects mass(es) of their ionic fragments
• Rapid multiple analytes from a single machine
  – Often 2-4 min/specimen
  – Easily automated
• Low false negative rate
• Not a 1:1 match of analyte to disease
  – Elevated C5-OH carnitine seen in seven disorders
    • Beta-ketothiolase d., Biotinidase d., Holocarboxylase d., HMG-CoA lyase d.,
    • 2Me3Hydroxybutyric acidemia, 3MeGlutaconic acidemia, 3MeCrotonyl carboxylase d.
New Disorders = New Jargon

- Acylcarnitines
  - Fatty acid or fatty acid-like molecule
  - Linked to an amino acid derivative
  - Usually referred to by the carbon chain length
    - C3 = propionyl
    - C14:1 = tetradecanoyl
  - Used to denote metabolites found in the chromatogram
  - Are the first clue to abnormalities which may represent disease
KEEPING CENTRAL TEXAS CHILDREN WELL
First Annual Pediatric Conference

Order NBS

Flag charts of unscreened patients

Provide Parental Education

Await Results

Concern: NBS not conducted

3 to 5 day old visit

Yes

No

Parents Decline NBS?

Parents Decline NBS?

Order NBS

Order NBS

Yes

No

Yes

No

Parents Decline NBS?

Concern: NBS not conducted

Yes

No

Await Results

Parents Decline NBS?

Order NBS

Flag charts of unscreened patients

Order NBS

Provide Parental Education

Await Results

Concern: NBS not conducted

Yes

No
Interpreting a NBS report

• Added sections
• Amino acids
  – Normal or named elevations
  – Possible disorder listed
• Acylcarnitine profile
  – Identify the abnormal chemical species
  – Possible disorders listed
Texas Department of State Health Services

LABORATORY SERVICES SECTION

CLIA #:04D466644

CONFIDENTIAL LABORATORY REPORT

TEXAS DEPARTMENT OF STATE HLTH SERVICES – 00000001

ATTN: LABORATORY

1100 W 49TH ST

AUSTIN, TX, 78758

Overall Status

NEWBORN SCREENING REPORT

Patient's Name: SMITH TEXAN

Laboratory Number: 2007/023 4568

Mother's Name: 

Form Serial No: 06-077696

Date of Birth: 01/10/2007

Date Collected: 01/11/2007

Medical Record: 

Date Received: 01/23/2007

Birth Weight: 2,800 grams

Test: 

Race/Ethnicity: 

Date Reported: 

Sex: 

Birth Order: 

Feed: BOTTLE

Mother's SSN: 

Physician's Name: 

Mother's Address: 1100 W 49TH AUSTIN, TX

Physician's Telephone: 

Status: NORMAL

NORMAL SCREEN

Disorder | Screening Result
---|---
Amino Acid Disorders | Normal
Fatty Acid Disorders | Normal
Organic Acid Disorders | Normal
Galactosemia | Normal
Biotinidase Deficiency | Normal
Endocrine Disorders | Normal
Hemoglobinopathies | Normal

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

Updates: Important messages and newborn screening updates are listed in this area.

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


For more information, please refer to http://www.dshs.state.tx.us/abimewbonscreening.htm
Result Received

Are NBS results available?

Yes

Consult ACT Sheets and DSHS

Disorder Identified?

Yes

Provide Parental Education

No

Discuss false positive & reassure family

No

Identify as CSHCN

Initiate chronic management

Out of Range

Invalid

In range

Document result & Reassure family

Reorder NBS

Call for NBS results

2 to 4 week Visit
Normal Results

• First Screen Normal
  – Do the second screen
• First AND Second Screen Normal
  – Document in your record and let family know that the required screening was normal
• But.........
**Overall Status**

**ABNORMAL SCREEN**

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Screening Result</th>
<th>Analyte</th>
<th>Analyte Result</th>
</tr>
</thead>
<tbody>
<tr>
<td>Amino Acid Disorders</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Fatty Acid Disorders</td>
<td>Abnormal: See Note 1</td>
<td></td>
<td>Normal</td>
</tr>
<tr>
<td>Organic Acid Disorders</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Galactosemia</td>
<td>Abnormal: See Note 2</td>
<td></td>
<td>Abnormal</td>
</tr>
<tr>
<td>Biotinidase Deficiency</td>
<td>Abnormal: See Note 3</td>
<td></td>
<td>T4/TSH</td>
</tr>
<tr>
<td>Endocrine Disorders</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hemoglobin Deficiency</td>
<td></td>
<td></td>
<td>Normal</td>
</tr>
</tbody>
</table>

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

The Screen Result column indicates if the disorder category tested is Normal, Abnormal, or Unsatisfactory.
The Analyte column lists analytes that indicate a specific disorder.
The Result Table includes an "Analyte" and "Analyte Result" column for Abnormal Screens.

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.

Important messages – Updated periodically:

The List of Disorders will print on all pages.

For more information, please refer to [http://www.dshs.state.tx.us/vs/bornscreening.shtml](http://www.dshs.state.tx.us/vs/bornscreening.shtml)
The Abnormal Screen

• What is the abnormality?
  – Endocrine: Refer appropriately
    • Some are emergencies
  – Hematologic: Refer appropriately
  – Metabolic: Evaluate the patient now
Are NBS results available?

- Yes: Call for NBS results
- No: 2 to 4 week Visit

NBS Result

- In range: Document result & Reassure family
- Invalid: Reorder NBS
- Out of Range: Consult ACT Sheets & DSHS Contact Consultant

Disorder Identified?

- Yes: Provide Parental Education
- No: Discuss false positive & reassure family

Result Received

- Yes: Do definitive testing Repeat NBS
- No: 2 to 4 week Visit
What to do

• Algorithmic approach
• Even a short delay may harm an infant
• Follow the ACT sheet and algorithm
  – Find the patient
  – Evaluate the patient
  – Obtain labs
  – Speak with a metabolic geneticist
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 acylglycines, 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 acylglycines will show elevated hexanoylglycerine. 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/newenglandconsortiumNSI/MCAD.html

Gene Tests
http://www.genetests.org/search/access?idx=genetests&state=ct&sid=18880918&key=New%70Ut%70Fr%70%70Gr%70%70Fr%70%70%70&f환=88%70Fr%70%70Fr%70%70%

Genetics Home Reference

STAR & FEISI
http://www.newbornscreening.info/Profiles/fattyaciddisorders/MCAD.html
http://www.newbornscreening.info/Parents/fattyaciddisorders/MCAD.html

Disclaimer: This information is adapted from American College of Medical Genetics website ACT sheets. http://www.acmg.com/resources/pediatrics/ACT/condition-analyte-link.htm
Algorithms

Flow Diagram format

Actions
in shaded boxes

Results
in Unshaded box

Plasma AC (C8) – high
Urine OA – Normal/high dicarboxylic acids
Urine AG – high hexanoylglycine

MCAD Deficiency

http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm
Newborn Screening Directory
1-800-252-8023

- Case Management Extensions
- General Information 2129
- Congenital Adrenal Hyperplasia (CAH) 2819
- Congenital Hypothyroidism 3666
- Galactosemia 6827
- Hemoglobinopathies 6832
- Phenylketonuria (PKU) 6827
- Biotinidase Deficiency 2071
- Fatty Acid Disorders, Organic Acid Disorders, Amino Acid Disorders 7715
Admission or Confirmatory Testing

• Some NBS values will be very high
  – **Metabolic Emergencies** requiring immediate admission
    • Intravenous fluids
    • May require specialized medications

• Many values will require repeat testing, or
  – Acylcarnitine profile
  – Urine organic acids
  – Plasma amino acids
  – Ammonia
  – Urine Orotic Acid
Who is going to become ill?

- **Timing**: days, weeks or never
- **If days**, then rapid turn-around and diagnosis needed
- **If weeks**, consistent with model of screening from PKU
- **If never**, can we select those who may ever get ill?
  - What criteria?
Now What Do I Do?

• Infant looks well
  – Information to parents

• Confirmatory testing results back
  – Normal results?
    • Good except for some Fatty Acid Oxidation Defects as normalization can occur
  – Abnormal … But not the same as the NBS
  – Abnormal and the same as the NBS
    • May need repeat or additional testing
Incomplete Evaluations

• Very Long Chain Acyl-CoA Dehydrogenase
  – Second screen is normal
  – But 2 known mutations found
• 1 of 2 recommended tests obtained
  – Miss a diagnosis
• Lab does not look for a metabolite
  – Know what the reference lab detects
    • Succinylacetone in Tyrosinemia, type I
Incomplete Evaluations 2

• Lab can do the test
  – Their reference ranges are non-neonatal
  – “Book” reference ranges
  – Limited numbers of neonates
  – Preterm vs. term infants
Information without Answers

• Abnormal screen: elevated tyrosine
  – Repeat screen - same result
• DDX:
  – Tyrosinemia, type I
  – Tyrosinemia, Type II or III
  – Transient tyrosinemia of the neonate
  – Liver disease including hyperbilirubinemia
  – Feeding practices (excessive protein intake)
  – Other
Elevated Tyrosine

- Exclude other causes of liver damage
- Get urine for succinylacetone
- Tyrosine level as plasma amino acids
- If succinylacetone is negative, may take weeks to resolve the issue

- There may be no disease!
Clinical Judgment vs. NBS

- NBS will detect
  - most of the severe or moderate cases of screened disorders
- NBS will not detect
  - non-screened disorders
  - all mild cases of a disorder (later presentations)
- NBS may not give an answer until after the infant is ill
  - critical window of time
- Physicians must still recognize and treat the infant with an IEM
Timing of Samples

- Preterm infants
  - Transfusions alter results
    - Galactosemia
    - Biotinidase
  - Nutrition
    - TPN and Carnitine
      - Carnitine palmitoyl transferase type I
- Fed or fasted
  - Need protein intake for metabolites to accumulate in some disorders
  - 48hr vs 24hr of age samples
Resources for You and Parents

- Screening Technology and Research in Genetics: STAR-G
  - HRSA funded multi-state consortium with consumer input

- http://www.newbornscreening.info/index.html

- Description of newborn screening process
- ‘Parent fact’ sheets for each disorder
- Overview of Genetics/Genes/Inheritance
- Glossary of screening terms, amino acid, etc.
Amino Acid Disorders

Disorder name: Arginemia / Arginase deficiency
Acronym: ARG 1 deficiency

- What is arginase deficiency?
- What causes arginase deficiency?
- If arginase deficiency is not treated, what problems occur?
- What is the treatment for arginase deficiency?
- What happens when arginase deficiency is treated?
- What causes the arginase enzyme to be absent or not working correctly?
- How is arginase deficiency inherited?
- Is genetic testing available?
- What other testing is available?
- Can you test during pregnancy?
- Can other members of the family have arginase deficiency or be carriers?
- Can other family members be tested?
- How many people have arginase deficiency?
- Does arginase deficiency happen more often in a certain ethnic group?
- Does arginase deficiency go by any other names?
- Where can I find more information?

This fact sheet has general information about arginase deficiency. Every child is different and some of these facts may not apply to your child specifically. Certain treatments may be recommended for some children but not others. All children with arginase deficiency should be followed by a metabolic doctor in addition to their primary doctor.

What is arginase deficiency?

Arginase deficiency is one type of amino acid disorder. People with this condition have problems removing ammonia from the body. Ammonia is a harmful substance. It is made when protein and its building blocks, amino acids, are broken down for use by the body.
NEWBORN SCREENING TESTS:
For Your Baby’s Health

NBS Brochure
Texas DSHS

KEEPING CENTRAL TEXAS CHILDREN WELL
First Annual Pediatric Conference

Version of 22 May 2007

NEWBORN SCREENING
For Your Baby’s Health

WHY IS MY BABY TESTED?
• Newborn Screening is one of the most important things done for your new baby’s health.
• We test all newborns because a few who look healthy have rare birth defects or disorder.
• If we find a defect, we can help prevent serious problems, such as mental retardation, illness, or death.

HOW IS MY BABY TESTED?
Two tests are required. The tests are done using drops of blood taken from your baby’s heel 1 to 5 days after birth in the hospital and again at 7 to 14 days of age in your doctor’s office or clinic. Some tests may not give true results.

BUT WE’VE NEVER HAD ANY BIRTH DEFECTS IN OUR FAMILY...
• Many of the babies born with these problems are from healthy families.
• Since these disorders are not very common, the chances are good that your child does NOT have one.

MY BABY SEEMS FINE. ARE THE TESTS STILL NEEDED?
• Yes. Most babies with these disorders look healthy at birth. Many disorders can’t be seen.

• If the newborn screen shows a problem, your baby’s doctor will be alerted to do more testing or start special treatment.

IF MY BABY HAS ONE OF THESE DISORDERS, CAN IT BE CURED?
We can’t cure these conditions. But early treatment may prevent or control serious health problems.

IF MORE TESTING MUST BE DONE, DOES THAT MEAN THAT MY BABY HAS A PROBLEM?
• No. An abnormal newborn screen only shows the need for more testing. This does NOT mean there is anything wrong. It just means your baby needs to be tested again to double-check for possible problems.

• If your baby’s test is unusual for a second time, the doctor may refer you or your baby to a doctor who specializes in the disorder for more testing.

HOW CAN I HELP THE DOCTOR TO HELP MY BABY?
1. Make sure your baby is tested before he or she leaves the hospital. You should get a form from your baby’s first screen. Take this form and your baby to your doctor or clinic for a second test at 7 to 14 days of age.

2. Your doctor may ask you to bring in your baby for more testing. Do it quickly! If your child has a disorder, fast action can be very important.

3. Be sure to give your current address and phone number to the hospital or doctor. If you don’t have a phone, leave the number of a friend, relative, or neighbor with the doctor or hospital.

4. If you move soon after your baby is born, be sure your doctor knows your new address. Then, if your child needs to be tested again, your doctor will know whom to reach you. Remember, these steps are very important!
**Hoja informativa sobre el examen médico del recién nacido**

**Insuficiencia de acil-CoA deshidrogenasa de cadena media (MCAD, las siglas corresponden a nombres en inglés)**

¿Qué es la MCAD? La MCAD es un tipo de alteración de la oxidación de los ácidos grasos. Las personas con MCAD tienen dificultad para degradar (o digerir) las grasas y convertirlas en energía para el organismo.

¿Qué causa la MCAD? Las enzimas ayudan a iniciar las reacciones químicas en el organismo. La MCAD surge cuando la enzima denominada “acil-CoA deshidrogenasa de cadena media” está ausente o no funciona. Esta enzima degrada ciertas grasas de los alimentos que consumimos para convertirlas en energía. También degrada la grasa ya almacenada en el cuerpo.

¿Síntomas o problemas produce la MCAD? [En algunos niños con esta enfermedad pueden presentarse ciertos síntomas. No son habituales en todas] Por eso, algunos niños con MCAD pueden presentar ciertos síntomas:

- falla de apetito
- pérdida de peso o demasiado crecimiento
- cambios en la coloración de la piel
- irritabilidad
- problemas respiratorios
- convulsiones
- retraso mental
- parálisis cerebral
- otros, que van a mejorar con el tiempo.

¿Cuál es el tratamiento para la MCAD? A menudo se usan los siguientes tratamientos para los niños con MCAD:

1. No dejar que el niño coma ni coma con bebés durante la noche si no está despierto. No debe pasar más de 4 a 6 horas sin comer. Algunos bebés necesitan comer más allá de 6 horas sin comer al día. Un bebé pequeño con MCAD debe comer algo con almíbar y bebés más grandes pueden tomar una forma importante de la enzima en el organismo. Cuando estén en la glía, esto ayuda a los niños a mantenerse enfermos en la noche, los niños a comer y menos retraso en el crecimiento. Si no se dan retrasos en el crecimiento, las mismas alteraciones del crecimiento pueden ser causadas por el bebé con MCAD con una enfermedad genética o una desviación congénita.

¿Cómo se hacen estos exámenes al bebé? Se pueden hacer de dos a tres veces al día durante el primer año de vida.

- Analítico de orina: se hace una muestra de la orina del bebé para examinar la orina de la orina y la orina de la orina para detectar la enfermedad. La orina de la orina es la orina de la orina para detectar la enfermedad. Si se encuentra algo en la orina que no debería estar allí, se puede hacer un análisis más detallado.

**EXÁMEN MÉDICO DEL RECÍN NACIDO**

Por la salud de su bebé

**¿Por qué el médico quiere ver a mi bebé?**

El examen médico para los recién nacidos es uno de los más importantes para que el bebé no sea enfermo. Se hacen exámenes a todos los bebés nacidos para que, en caso de que algún cambio en el bebé, pueda detectarse lo antes posible.

- La descripción empírica se refiere a las enfermedades genéticas que son difíciles de tratar.
- La descripción empírica se refiere a los síntomas que se pueden detectar.

**¿Cómo puedo hacer que mi bebé esté en un problema?**

- Por favor, en el caso de que su bebé sea enfermo, por favor, hágale más pruebas.
- No sólo puede ser un problema para el médico, sino que también puede ser un problema para el bebé.
- Por favor, hágale más pruebas.

**Si mi bebé tiene alguna de estas enfermedades, ¿puedo curarlas?**

No podemos curar estas enfermedades, pero podemos tratarlas y controlarlos problemas genéticos del bebé.

**SÍ DIBUJAMOS REALES MÁS PRUEBAS, ¿SIGNIFICA QUE MI BÉBÉ TIENE UN PROBLEMA?**

- Sí, Un resultado anormal en el examen de un recién nacido no significa que haya un problema. Sólo significa que hay que hacer pruebas para ver si es algo en el bebé.

**Si el resultado del examen de su bebé es anormal por segundo vez, el médico puede iniciar un tratamiento para el bebé o indicar que un profesional de salud es especialista en enfermedades genéticas.**

**¿Cómo puedo asistir al médico para que atienda a mi bebé?**

1. Asista a que se le hagan pruebas a su bebé en el lugar de su médico.
2. Llame al número de teléfono correcto del médico si es el caso.
3. Asiste a la consulta de su médico y ayude a su bebé.
4. Si su bebé tiene un problema, el médico debe tomar pruebas para ver si es algo en su bebé.

**Recuerde, por favor, que su médico lo enumera.**
March of Dimes Resources

• A Parents Guide to Newborn Screening
  – 5 minutes long DVD (English and Spanish)
  – Also as streaming video from the Pregnancy/Newborn section
    – www.marchofdimes.com
    – www.nacersano.org

• Or from DSHS
  http://www.dshs.state.tx.us/newborn/expandparent.shtm
Starting a discussion with parents:

**8 Things Parents Want To Know About Newborn Screening**

1. The Texas Newborn Screening Program checks all newborn babies for 27 rare disorders. The screening tests are very important for your baby's health.
2. Babies with these disorders may look healthy at birth. Many disorders cannot be seen.
3. Screening problems, such as unusual weakness, illness, or death, may be prevented if we find the disorders right away.
4. Newborns are first tested 1 to 3 days after birth before they leave the hospital and again at 7 to 14 days of age in their doctor's office or clinic.
5. To do the test, a health professional will take a few drops of blood from your baby's heel.
6. Your baby's health professional or the hospital will get a copy of the test results. Tell your baby's health professional if you would like to talk about the results.
7. Some babies need more tests. You will be notified if your baby needs more tests.
8. It is very important for you and your baby to take these tests.

**Newborn Screening**

**Biotinidase Deficiency (BIO) BIO** is a rare enzyme deficiency that occurs in about 1 in 50,000 newborns and can result in seizures, hearing loss, and death in severe cases. Treatment is simple and involves daily doses of biotin.

**Congenital Adrenal Hyperplasia (CAH)** CAH is caused by a deficiency in the production of certain adrenal hormones. The most common type is caused by a deficiency in the production of cortisol. Early detection can prevent death in babies and girls and severe skin- and hair-related symptoms. Treatment includes lifelong hormone replacement therapy.

**Congenital Hypothyroidism (CH)** is a rare disorder that occurs in about 1 in 100,000 newborns. Early detection can prevent death in babies and severe intellectual disability. Treatment includes lifelong hormone replacement therapy.

**Galactosemia (GAL)** is a rare disorder that occurs in about 1 in 50,000 newborns. Early detection can prevent death and intellectual disability. Treatment includes lifelong exclusion of milk products.

**Homozygous (HCH)** HCH is a rare disorder that occurs in about 1 in 50,000 newborns. Early detection can prevent death and intellectual disability. Treatment includes lifelong exclusion of milk products.

**Maple Syrup Urine Disease (MSUD)** MSUD is a rare disorder that occurs in about 1 in 50,000 newborns. Early detection can prevent death and severe intellectual disability. Treatment includes lifelong exclusion of milk products.

**Medium Chain Acyl-CoA Dehydrogenase (MCAD)** is a rare disorder that occurs in about 1 in 50,000 newborns. Early detection can prevent death and intellectual disability. Treatment includes lifelong exclusion of milk products.

**Organic Acid Urinary Syndrome (OA)** is a rare disorder that occurs in about 1 in 50,000 newborns. Early detection can prevent death and intellectual disability. Treatment includes lifelong exclusion of milk products.

**Phenylketonuria (PKU)** is a rare disorder that occurs in about 1 in 100,000 newborns. Early detection can prevent death and intellectual disability. Treatment includes lifelong exclusion of milk products.

**Sickle Cell Disease (SCD)** is a rare disorder that occurs in about 1 in 100,000 newborns. Early detection can prevent death and intellectual disability. Treatment includes lifelong exclusion of milk products.

**Urea Cycle Disorders (UCD)** is a rare disorder that occurs in about 1 in 100,000 newborns. Early detection can prevent death and intellectual disability. Treatment includes lifelong exclusion of milk products.

**What Parents Want to Know About Newborn Screening**

From Their Baby's Health Care Provider
Help for Brief Discussions:

1. Screening is required
2. Infant is generally healthy at birth
3. Serious consequences
4. **Tested at 1-2 and 7-14 D**
5. Blood sample from the heel
6. Results go to MD and birth hospital
7. Retesting may be needed
8. For more information: call your PCP or Department of State Health Services

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**8 Things Parents Want To Know About Newborn Screening**

- The Texas Newborn Screening Program checks all newborn babies for 27 rare disorders. The screening tests are very important for your baby’s health.
- Babies with these disorders may look healthy at birth. Many disorders can’t be seen.
- Some problems, such as problems with hearing, vision, or blood sugar, can be prevented if we find the disorder early enough.
- Newborns are first tested 1 to 2 days after birth to learn if they are at the hospital and again at 7 to 14 days of age in their doctor’s office or clinic.
- To do the test, a health professional will take a small drop of blood from your baby’s heel.
- Your baby’s health professional at the hospital will get a copy of the test results. Call your baby’s health professional if you would like to talk about the results.
- Some babies may need more tests. You will be notified if your baby needs more tests.
- It is very important for your baby to get these tests early on.
- If you have any questions, you can call your baby’s health professional or the Texas Department of State Health Services – Newborn Screening Program toll-free at 1-800-258-5023 or visit txscreen.org.
Resources for the MD

• New information on the DSHS website
  – www.dshs.state.tx.us/newborn/default.shtm
  – ACT and FACT sheets
  – CME for an education module
    • http://txhealthsteps.com/

• AAP
  – www.medicalhomeinfo.org/screening/newborn.html
  – Link to an overview of many of the disorders
    www.dshs.state.tx.us/newborn/pdf/AAPFactSheets.pdf

• National Newborn Screening and Genetic Resource Center
  – http://genes-r-us.uthscsa.edu/
  – Links to states NBS programs & to the ACT sheets
Summation

- More disorders: individually rare
- New methods – same specimens
- New information
- New potential problems
- Resources available on line and in print
- Texas DSHS Site:
  - www.dshs.state.tx.us/newborn/default.shtm
  - Consultants