

Positive Newborn Screens: What do you do next?

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A member of the  Seton Family of Hospitals

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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

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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

Prior to 2006 Texas Screened for:

List of 29 Disorders for NBS

- 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)
- **Congenital adrenal hyperplasia (CAH)**
- **Congenital hypothyroidism (HYPOTH)**
- *Cystic fibrosis (CF)*
- **Galactosemia (GALT)**
- Glucose-6-phosphate dehydrogenase deficiency (G6PD)
- Glutaric acidemia type I (GA I)
- **Hb S/Beta-thalassemia (Hb S/Th)**
- **Hb S/C disease (Hb S/C)**
- **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

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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.

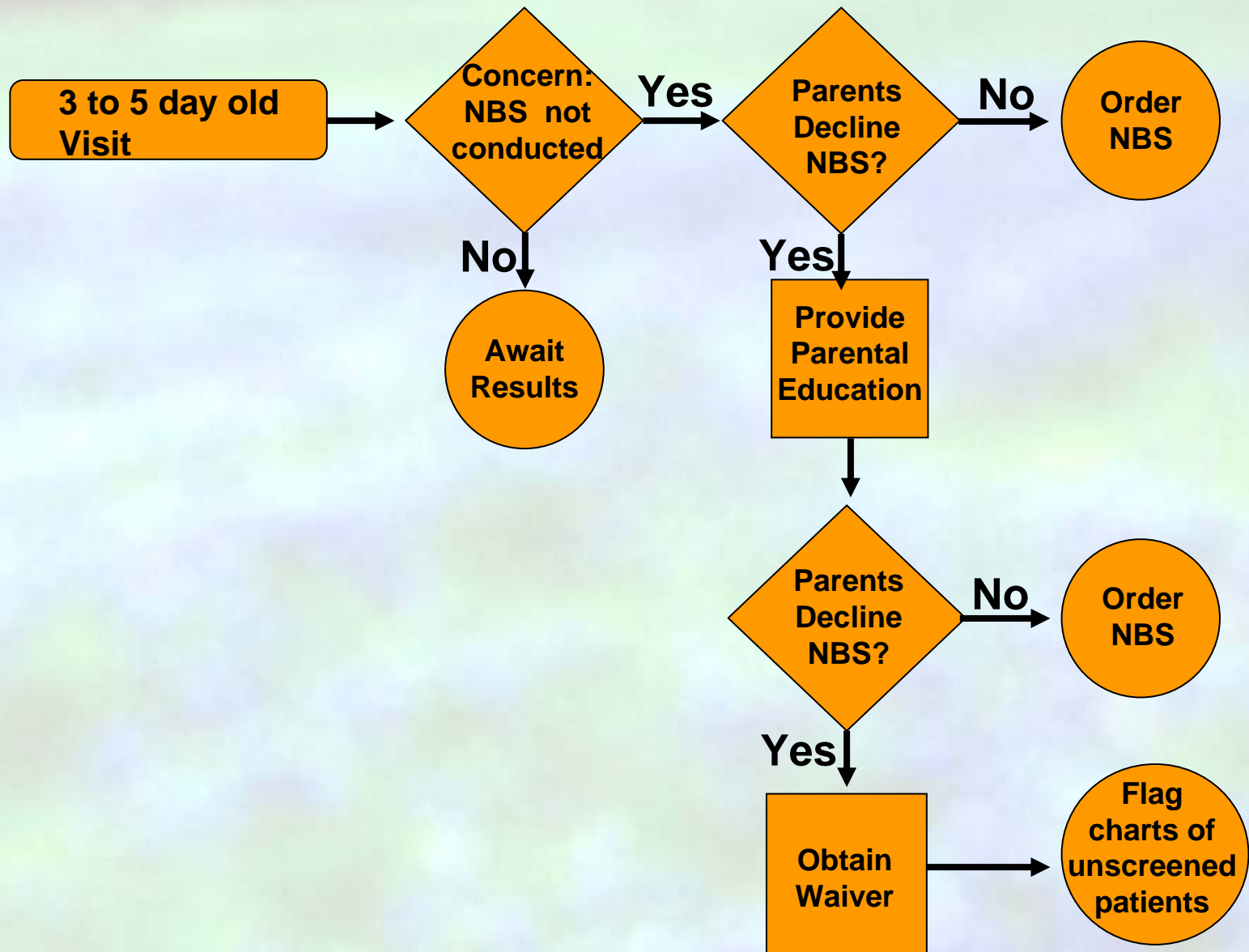
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New Disorders = New Jargon

- 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



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 #45D0660644

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

CONFIDENTIAL LABORATORY REPORT

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

Overall Status

NEWBORN SCREENING REPORT

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

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:

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

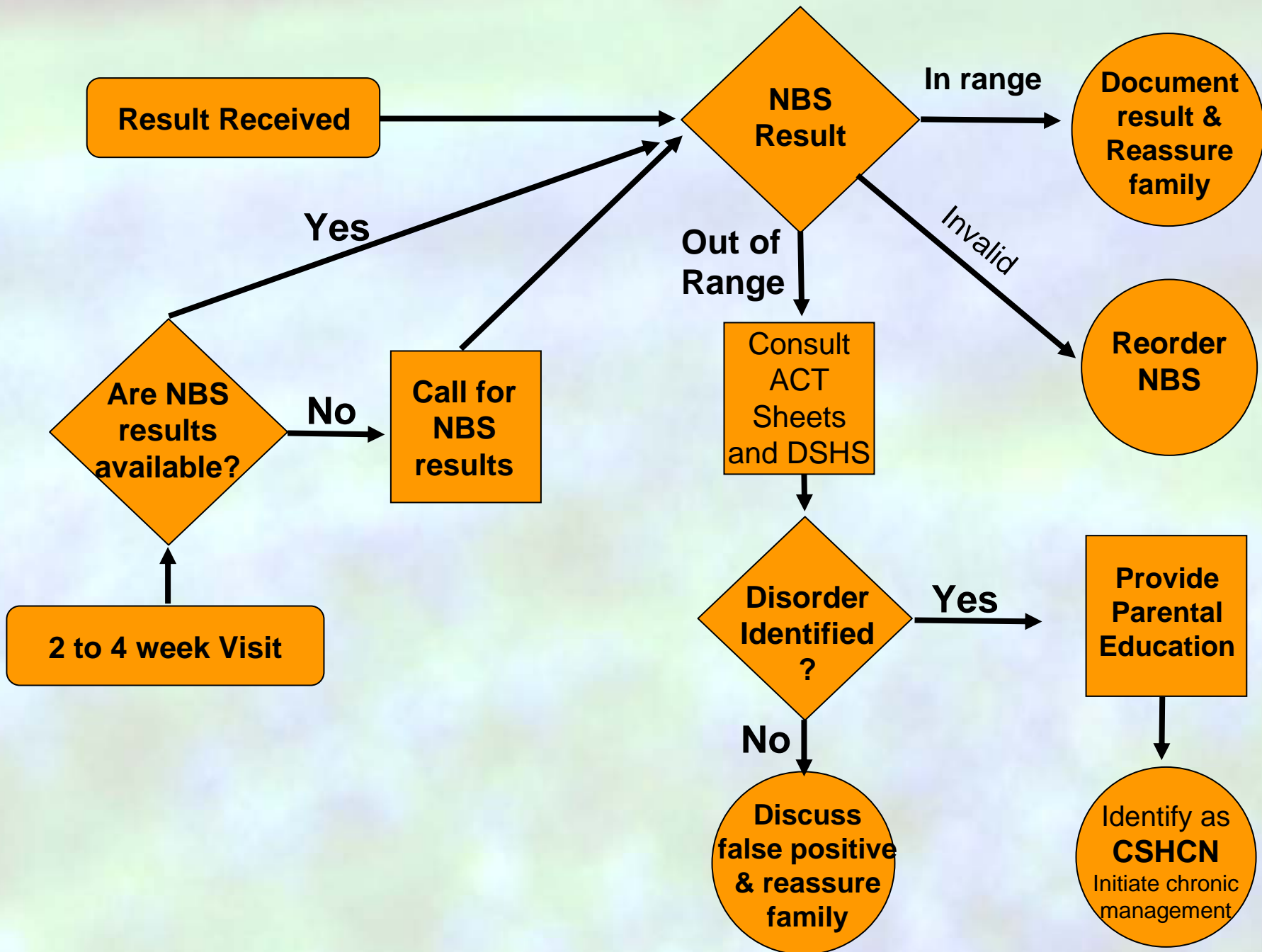
Result Table: Results in the table are listed by category of the disorder.

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.

IMPORTANT MESSAGES: Updated February 7, 2007
EXPANSION: All specimens are now tested for 27 disorders.
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.
TUTORIAL: A web-based tutorial (free CME) on the Texas NBS Expansion is available at <http://txhealthsteps.com/>.
24-HOUR, 7-DAY RESULT ACCESS (not including the expansion tests) is available via the Voice Response System (VRS).
Call 1-888-963-7111 ext. 6988 or e-mail labappsupport@dshs.state.tx.us to obtain a PIN and instructions.

Disorders Screened: **AMINO ACID DISORDERS:** Argininosuccinic Acidemia (ASA), Citrullinemia (CIT), Homocystinuria (HCY), Maple Syrup Urine Disease (MSUD), Phenylketonuria (PKU), Tyrosinemia type 1 (TYR). **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 1 (GA-1), 3-OH 3-Methyl Glutaric Acidemia (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, ENDOCRINE DISORDERS:** Congenital Hypothyroidism (CH), Congenital Adrenal Hyperplasia (CAH). **HEMOGLOBINOPATHIES:** Including Hb SS, Hb SC, Hb S-Beta thalassemia



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.....



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Laboratory Number: 2007 023 4568
Form Serial No: 06-0277696
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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	CE C10 C16 C26/2	Elevated Elevated Normal Elevated Elevated
Organic Acid Disorders	Normal		
Galactosemia	Normal		
Biotinidase Deficiency	Abnormal: See Note 2	Biotinidase	Abnormal
Endocrine Disorders	Abnormal: See Note 3	T4/TSH	T4 Low, TSH Slightly Elevated
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.

The List of Disorders will print on all pages.

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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 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.

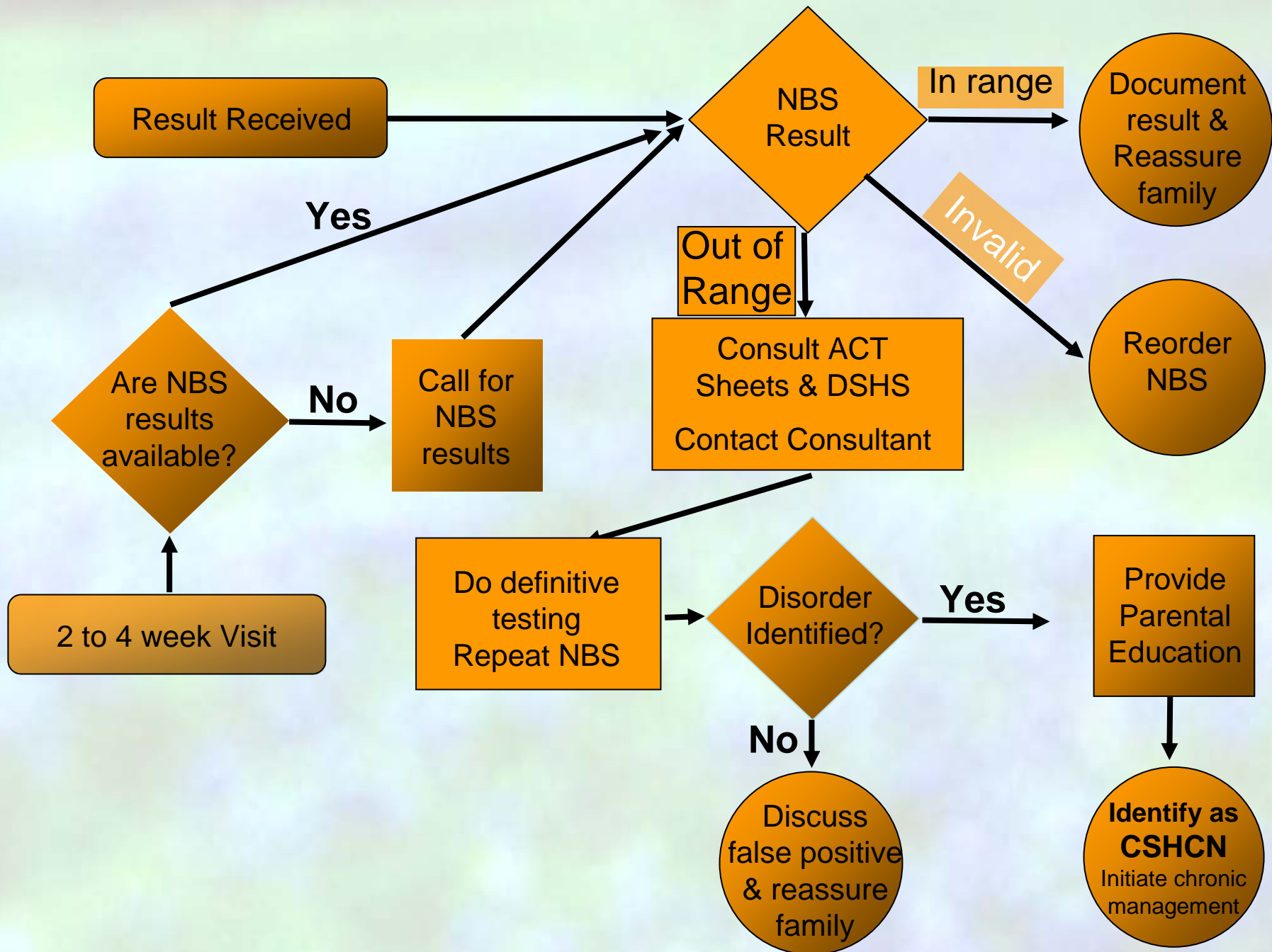
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The Abnormal Screen

- What is the abnormality?
 - Endocrine: Refer appropriately
 - Some are emergencies
 - Hematologic: Refer appropriately
 - Metabolic: Evaluate the patient now



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



DSHS ACT Sheets

What Analyte

PCP's Actions

Diagnostic
Evaluations

Clinical
Snapshot

More
Information

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 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/serve/ac cess?d b= genecli nic s& site= gt& id= 8888991& key= NexQvD btf nPSK& gry= & fc n=y& fw= 09 lz& file nam e= profiles/mcad/index.html>

Genetics Home Reference

<http://ghr.nlm.nih.gov/condition=mediumchainacylcoenzymeadehydrogenasedeficiency>

STAR G FELSI

<http://www.newbornscreening.info/Pro/fattyacid disorders/MCADD.html>

<http://www.newbornscreening.info/Parents/fattyacid disorders/MCADD.html>

Algorithms

Flow Diagram format

Actions

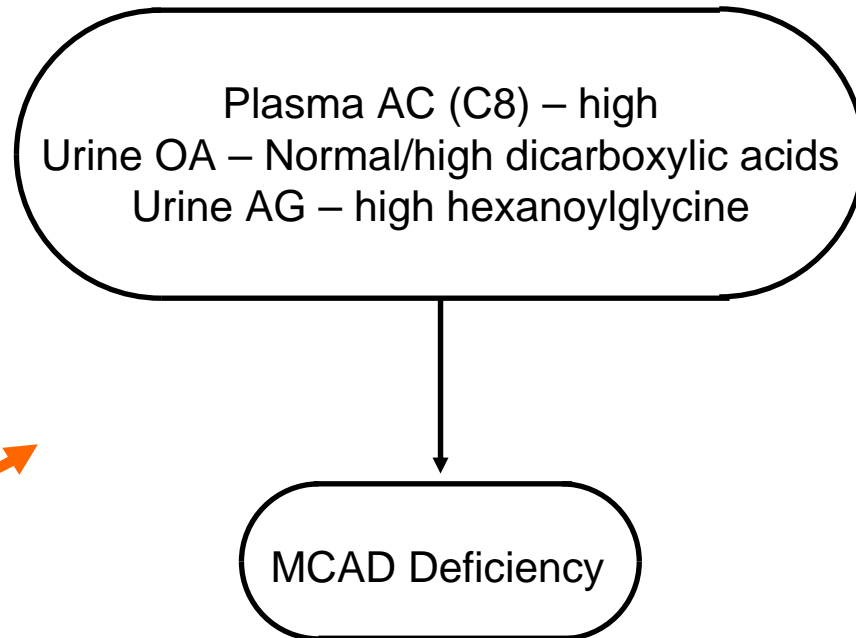
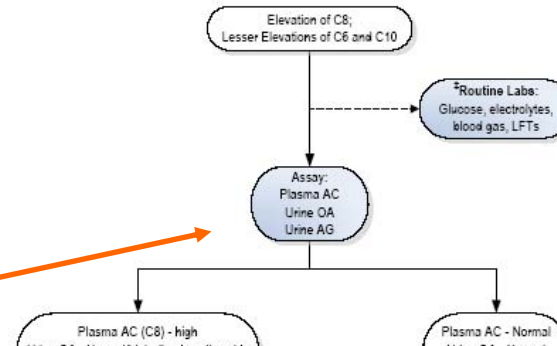
in shaded boxes

Results

in Unshaded box



C8 Elevated + Lesser Elevations of C6 and C10



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

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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.

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Genetic Fact Sheets for Parents Amino Acid Disorders

Screening, Technology, and Research in Genetics is a multi-state project to improve information about the financial, ethical, legal, and social issues surrounding expanded newborn screening and genetic testing - <http://bit.ly/starproject.org>

Disorder name: Argininemia / 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.

10-11 pages

Written for parents

Covers the same points for each disorder

Cause

Problems

Treatment

Inheritance

Testing

More Information

Printable form

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NEWBORN SCREENING TESTS:
For Your Baby's Health

Texas Department of State Health Services
Newborn Screening Program
1100 West 49th Street
Austin, Texas 78756
1-800-252-8023 ext. 2129
www.dshs.state.tx.us/newborn



NBS Brochure

Texas DSHS

NEWBORN SCREENING

For Your Baby's Health



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 the baby 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 start treating your baby at once or send you 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 be given 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 correct 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, let your doctor know right away. Then, if your child needs to be tested again, your doctor will know where to reach you.

Remember, these steps are very important!

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 a rare birth defect or disorder.
- If we find it early, we can help prevent serious problems, such as mental retardation, illness, or death.

HOW IS MY BABY TESTED?

Two screens are required. The tests are done using drops of blood taken from your baby's heel: 1 to 2 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.

Version of 22 May 2007

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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.

¿Cuál es la causa de 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 comemos para convertirlas en energía. También degrada la grasa ya almacenada en el cuerpo.

¿Qué síntomas o problemas produce la MCAD?

[Un síntoma es algo que los niños manifiestan cuando están enfermos y que es fácil de notar por los padres.]

La MCAD puede causar períodos de enfermedad llamados crisis metabólicas. Los niños con MCAD a menudo muestran síntomas por primera vez entre los 8 meses y los 2 años de edad. Algunos de los primeros indicios de una crisis metabólica son:

- demasiado sueño
- cambios de conducta (por ej., llorar sin motivo)
- irritabilidad
- falta de apetito

Si una crisis metabólica no se trata, un niño con MCAD puede desarrollar:

- problemas respiratorios
- convulsiones
- retraso mental
- parálisis cerebral
- coma, que a veces lleva a la muerte

¿Cuál es el tratamiento para la MCAD?

A menudo se usan los siguientes tratamientos para los niños con MCAD:

1. No dejar pasar mucho tiempo sin comer – los bebés y los niños pequeños con MCAD necesitan comer a menudo para evitar una baja del azúcar en la sangre o una crisis metabólica. No debe pasar más de 4 a 6 horas sin comer. Algunos bebés necesitan comer incluso más seguido. Es importante que los bebés reciban alimento durante la noche. Si no se despiertan solos, es necesario despertarlos para alimentarlos. Los niños pequeños con MCAD pueden necesitar un refrigerio con almidón (tal como pan,

cereales, arroz) antes de irse a dormir y otro durante la noche. También pueden necesitar ingerir otro refrigerio apenas se levantan en la mañana. Su dietista puede darle ideas de refrigerios adecuados para la noche. Los dietistas saben cuáles son los alimentos correctos para cada caso. La mayoría los adolescentes y adultos con MCAD pueden a las 12 horas sin comer sin problemas, cuando está bien. Los demás tratamientos, deben continuar de por vida.

2. Dieta – a veces se aconseja una dieta baja en grasas y alta en carbohidratos (como verduras, frutas y cereales). Su dietista puede crear un plan de alimentación con el tipo y la cantidad correcta de grasas que su hijo necesita. Pregúntele a su médico si su hijo necesita algún cambio en la dieta.

3. L-carnitina – a algunos niños se les puede recetar la L-carnitina (Carnitor). Esta sustancia es segura y natural y ayuda a las células del organismo a generar energía. La L-carnitina también ayuda al cuerpo a eliminar los residuos dañinos.

Cosas para recordar

Siempre llame al médico cuando su hijo tenga alguno de estos síntomas:

- falta de apetito
- poca energía o demasiado sueño
- vómitos
- diarrea
- una infección
- fiebre

Las personas con MCAD deben comer más alimentos con almidón y beber más líquidos durante cualquier enfermedad—aunque no tengan hambre—o podr sufrir una baja del azúcar en la sangre o una crisis metabólica. Cuando están enfermos, los niños a menudo no desean comer. Si se niegan a comer o si pueden hacerlo, podría ser necesario tratarlos en hospital para evitar problemas.

State Materials in Spanish

Fact Sheets and NBS Brochures

EXAMEN MÉDICO DEL RECIÉN NACIDO

Por la salud de su bebé



¿POR QUÉ LE HACEN EXÁMENES A MI BEBÉ?

- El examen médico para los recién nacidos es uno de los más importantes para la salud de su bebé.
- Hacemos exámenes a todos los recién nacidos porque algunos de ellos, a pesar de tener un aspecto saludable, pueden tener alguna alteración o defecto congénito raro.
- La detección temprana ayuda a evitar problemas graves como el retraso mental, enfermedades crónicas o la muerte.

¿CÓMO LE HACEN ESTE EXAMEN A MI BEBÉ?

Se requieren dos tipos de exámenes. Éstos se hacen extrayendo unas gotas de sangre del talón de su bebé, el primero 1 ó 2 días después del nacimiento, y el segundo en la oficina de su médico o en una clínica a los 7 a 14 días después de haber nacido. Algunos de estos exámenes pueden dar resultados falsos y en estos casos algunos estudios serán repetidos.

USTED SE PRECUNTARÁ: ¿PERO CÓMO? NADIE EN NUESTRA FAMILIA HA TENIDO PROBLEMAS CONGÉNITOS...

- Muchos de los bebés que nacen con estas enfermedades vienen de familias saludables.
- Dado que estas enfermedades no son muy comunes, lo más probable es que su bebé NO las padezca.

MI BEBÉ SE VE MUY BIEN. ¿DE TODAS FORMAS ES NECESARIO REALIZAR LAS PRUEBAS?

- Sí. La mayoría de los bebés con estas enfermedades se ven saludables al nacer, ya que muchas de ellas no son visibles.
- Si el examen del recién nacido muestra que hay un problema, se alertará al médico de su bebé para que realice más pruebas o empiece un tratamiento especial.

SI MI BEBÉ TIENE ALGUNA DE ESTAS ENFERMEDADES, ¿PUEDE CURARSE?

No podemos curar estas enfermedades, pero el tratamiento temprano puede evitar o controlar problemas graves de salud.

SI SE DEBEN REALIZAR MÁS PRUEBAS, ¿SIGNIFICA QUE MI BEBÉ TIENE UN PROBLEMA?

- No. Un resultado anormal en el examen de un recién nacido sólo indica la necesidad de hacer más pruebas. Esto NO implica que haya un problema. Sólo significa que hay que repetir las pruebas para verificar por segunda vez si hay algún problema.
- Si el resultado del examen de su bebé es anormal por segunda vez, el médico puede iniciar de inmediato un tratamiento para su bebé o indicarle que vea a un especialista en estas enfermedades para que le haga más pruebas.

¿CÓMO PUEDO ASISTIR AL MÉDICO PARA QUE AYUDE A MI BEBÉ?

1. Asegúrese de que le hagan las pruebas a su bebé antes de salir del hospital. Le deben dar una forma con los primeros exámenes de su bebé. Luego de 7 ó 14 días de nacido, lleve a su bebé y esta forma al médico o a la clínica para que le hagan una segunda prueba.
2. Su médico podría pedirle que lleve a su bebé para hacerle más pruebas. En ese caso, ¡llévelo pronto! Si su hijo tiene una enfermedad, el actuar rápidamente puede ser muy importante.
3. Asegúrese de darle su dirección y número de teléfono correcto al hospital o al médico. Si no tiene teléfono, dé al hospital o al médico el número de algún amigo, familiar o vecino.
4. Si se muda enseguida tras el nacimiento de su bebé, comuníquese de inmediato a su médico. Así, si su hijo requiere más pruebas, su médico sabrá dónde localizarlo.

Recuerde, estos pasos a seguir son muy importantes!

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

From Their Baby's Health Professional

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 can't be seen.
3. Serious problems, such as mental retardation, 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. Call your baby's health professional if you would like to talk about the results.
7. 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 quickly.*
8. If you have more questions, you can call your baby's health professional or the Texas Department of State Health Services - Newborn Screening Program toll-free at 1-800-252-8023 ext. 2129.

NEWBORN SCREENING



QUICK REFERENCE TO NEWBORN SCREENING DISORDERS

Biotinidase Deficiency (BIOT) BIOT is an 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 decreased or absent production of certain adrenal hormones. The most common type is detected by newborn screening in about 1 in 15,000 newborns. Early detection can prevent death in boys and girls and sex assignment in girls. Treatment involves a lifelong hormone replacement therapy.

Congenital Hypothyroidism (CH) Inadequate or absent production of thyroid hormone results in CH and is present in about 1 in 3,500 newborns. Thyroid hormone replacement therapy begun by 1 month of age can prevent mental and growth retardation.

Galactosemia (GALT) Failure to metabolize the milk sugar galactose results in GALT and occurs in about 1 in 30,000 newborns. The classical form detected by newborn screening can lead to cataracts, liver cirrhosis, mental retardation, and/or death. Treatment eliminates galactose from the diet, usually by substituting soy for milk products.

WHAT PARENTS WANT TO KNOW ABOUT NEWBORN SCREENING

From Their Baby's Health Care Professional

Homocystinuria (HCU) HCU is caused by an enzyme deficiency that blocks the metabolism of an amino acid that can lead to mental retardation, osteoporosis, and other problems if left undetected and untreated. The incidence is approximately 1 in 350,000 U.S. newborns. Treatment may involve dietary restrictions and supplemental medicines.

Maple Syrup Urine Disease (MSUD) MSUD is a defect in the way that the body metabolizes certain amino acids and is present in about 1 in 200,000 U.S. newborns. Early detection and treatment with dietary restrictions can prevent death and severe mental retardation. There is an increased risk in Mennonites.

Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency The most common disorder in the way the body metabolizes fatty acids is called MCAD deficiency. Undetected, it can cause sudden death. Treatment is simple and includes ensuring regular food intake. The incidence from newborn screening is not yet known, but is thought to be approximately 1 in 15,000 newborns.

Other Fatty Acid Oxidation (FAO) Disorders Besides MCAD deficiency, other FAO disorders may be detected through newborn screening. They are usually described in categories based on the length of the fatty acid involved. Undetected and untreated they can cause seizures, coma, and even death. The incidences of various FAO disorders are not known since it is only recently that early detection through newborn screening has occurred.

Phenylketonuria (PKU) An enzyme defect that prevents metabolism of phenylalanine, an amino acid essential to brain development, is known as PKU. It occurs in approximately 1 in every 10,000 U.S. newborns. Undetected and untreated with a special diet, PKU leads to irreversible mental retardation. Persons of European descent are at increased risk.

Sickle Cell Disease (SCD) Sickle cell anemia (Hb-SS-Disease) is the most common SCD and causes clogged blood vessels resulting in severe pain and other severe health problems. Other common SCDs include Hb-S-C-Disease and various thalassemias. Newborn screening detects about 1 in 2,500 newborns with SCD annually. Persons of African or Mediterranean descent are at an increased risk.

Tyrosinemia (TYR) People with tyrosinemia have problems breaking down an amino acid called tyrosine, which is one of the building blocks of protein. If not treated, the condition causes severe liver disease and other serious health problems. Treatment consists of medication and a diet low in tyrosine. The estimated incidence is 1 case in every 100,000 live births.

Organic Acid (OA) Disorders Organic acidemias are a group of metabolic disorders that lead to build up of organic acids in the blood and urine and may be detected in newborn screening through analysis of acylcarnitine profiles. Restricting protein in the diet and supplementation with vitamins and/or carnitine can diminish symptoms. Because newborn screening for these disorders is relatively new, the degree of occurrence in newborns is not yet known.

Urea Cycle Disorders (UCD) A UCD is a genetic disorder caused by a deficiency of one of the enzymes responsible for removing ammonia from the blood stream. Some UCDs may be detected as a part of newborn screening. They are characterized by seizures, poor muscle tone, respiratory distress, and coma, and result in death if left undetected and untreated. Because newborn screening for these disorders is relatively new, the degree of occurrence in newborns is not yet known.

What Parents Want to Know

Help for Brief Discussions:

8 THINGS PARENTS WANT TO KNOW ABOUT NEWBORN SCREENING

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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

KEEPING CENTRAL TEXAS CHILDREN WELL
First Annual Pediatric Conference

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

KEEPING CENTRAL TEXAS CHILDREN WELL

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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

