Provider and Family Fact Sheets: What Do Healthcare Providers Really Need?

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PROBLEM IDENTIFIED

Upon the notification of an abnormal newborn screening result, the Minnesota Department of Health Newborn Screening Program faxes both a provider and family fact sheet to the child's primary care provider (PCP). However, we are unaware if the child's PCP actually receives or uses the fact sheets. Additionally, when the fact sheets were first created, assumptions were made regarding what information providers and families would find useful.

GOAL

We sought to better understand PCP usage of the fact sheets and the content they found important, with the goal of revising these fact sheets to better serve the needs of our PCPs.

METHOD

We developed an electronic survey to email to PCPs. Providers who had a patient with a positive newborn screening result in the past six months were invited to participate in the survey. These PCPs consisted of MDs, DOs, and NPs. Provider emails were obtained from the Minnesota Board of Medical Practice and Board of Nursing. As a reminder to the survey participants, the survey included examples of our current provider and family fact sheets. We chose to use our cystic fibrosis fact sheet since it accounted for the majority of the results these providers had experienced. The survey was opened on June 10, 2016 and closed on July 15, 2016.

QUICK FACTS

The survey was emailed to **103** primary care providers and **20** completed the survey (**19.4%** response rate). Respondents had been practicing for a median of **16** years.

15 respondents recalled receiving the provider fact sheet and **100%** found it useful overall.

For the family fact sheet, **17** respondents recalled receiving it, **8** actually provided it to the family, **2** did not provide it, and **7** could not recall.

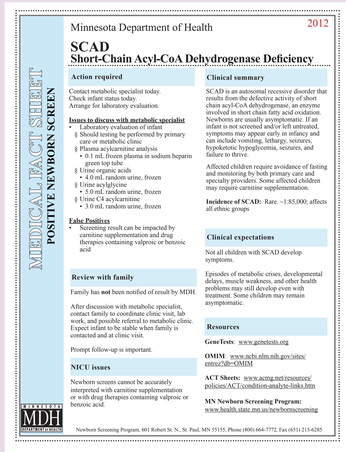
18 respondents preferred to receive the fact sheets by fax (even when offered the option of integration into their EMR system).

When asked if they'd prefer the American College of Medical Genetics ACTion (ACT) Sheets, **15** indicated they were unfamiliar with them.

WHAT DID WE LEARN?

Very Important or Important | **Somewhat or Not Important**

PROVIDER FACT SHEETS


Minnesota Department of Health 2012

MEDICAL FACT SHEET
POSITIVE NEWBORN SCREEN

SCAD
Short-Chain Acyl-CoA Dehydrogenase Deficiency

Action required

Contact metabolic specialist today.
Check infant status today.
Arrange for laboratory evaluation.

Issues to discuss with metabolic specialist

- Laboratory evaluation of infant
  - § Should testing be performed by primary care or metabolic clinic
  - § Plasma acylcarnitine analysis
    - 0.1 mL frozen plasma in sodium heparin green top tube
  - § Urine organic acids
    - 4.0 mL random urine, frozen
  - § Urine acylglycine
    - 5.0 mL random urine, frozen
  - § Urine C4 acylcarnitine
    - 3.0 mL random urine, frozen

False Positives

- Screening result can be impacted by carnitine supplementation and drug therapies containing valproic or benzoic acid

Review with family

Family has **not** been notified of result by MDH.

After discussion with metabolic specialist, contact family to coordinate clinic visit, lab work, and possible referral to metabolic clinic. Expect infant to be stable when family is contacted and at clinic visit.

Prompt follow-up is important.

NICU issues

Newborn screens cannot be accurately interpreted with carnitine supplementation or with drug therapies containing valproic or benzoic acid.

Clinical summary

SCAD is an autosomal recessive disorder that results from the defective activity of short chain acyl-CoA dehydrogenase, an enzyme involved in short chain fatty acid oxidation. Newborns are usually asymptomatic. If an infant is not screened and/or left untreated, symptoms may appear early in infancy and can include vomiting, lethargy, seizures, hypoketotic hypoglycemia, seizures, and failure to thrive.

Affected children require avoidance of fasting and monitoring by both primary care and specialty providers. Some affected children may require carnitine supplementation.

Incidence of SCAD: Rare: ~1:85,000; affects all ethnic groups

Clinical expectations

Not all children with SCAD develop symptoms.

Episodes of metabolic crises, developmental delays, muscle weakness, and other health problems may still develop even with treatment. Some children may remain asymptomatic.

Resources

GeneTests: www.genetests.org

OMIM: www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM

ACT Sheets: www.acmg.net/resources/policies/ACT/condition-analyte-links.htm

MN Newborn Screening Program: www.health.state.mn.us/newbornscreening

MDH
Newborn Screening Program, 601 Robert St. N., St. Paul, MN 55155; Phone (800) 664-7772, Fax (651) 215-6285

OLD

Old: only describes the most likely disorder for the abnormal result

Survey: 100% want what was found on the screen (95% want a differential)

New: provides what was actually found and a list of differential diagnoses

Old: provides actions needed but is vague

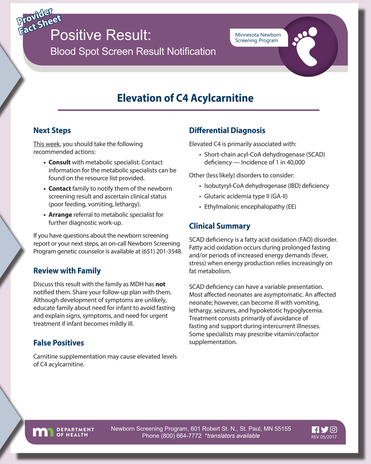
Survey: 95% of PCPs want to know their next steps

New: includes expected timeline, guidance on symptoms, and MDH contact information

Old: gives more provider guidance

Survey: 95% of PCPs want to know what to tell the family

New: actually gives them information to share with the family


Provider Fact Sheet

Positive Result:
Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevation of C4 Acylcarnitine

Next Steps

This week, you should take the following recommended actions:

- **Consult** with metabolic specialist. Contact information for the metabolic specialists can be found on the resource list provided.
- **Contact** family to notify them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy).
- **Arrange** referral to metabolic specialist for further diagnostic work-up.

If you have questions about the newborn screening report or your next steps, an on-call Newborn Screening Program genetic counselor is available at (651) 201-3548.

Review with Family

Discuss this result with the family as MDH has **not** notified them. Share your follow-up plan with them. Although development of symptoms are unlikely, educate family about need for infant to avoid fasting and explain signs, symptoms, and need for urgent treatment if infant becomes mildly ill.

False Positives

Carnitine supplementation may cause elevated levels of C4 acylcarnitine.

Differential Diagnosis

Elevated C4 is primarily associated with:

- Short-chain acyl-CoA dehydrogenase (SCAD) deficiency — Incidence of 1 in 40,000

Other (less likely) disorders to consider:

- Isobutyryl-CoA dehydrogenase (IBD) deficiency
- Glutaric acidemia type II (GA-II)
- Ethylmalonic encephalopathy (EE)

Clinical Summary

SCAD deficiency is a fatty acid oxidation (FAO) disorder. Fatty acid oxidation occurs during prolonged fasting and/or periods of increased energy demands (fever, stress) when energy production relies increasingly on fat metabolism.

SCAD deficiency can have a variable presentation. Most affected neonates are asymptomatic. An affected neonate; however, can become ill with vomiting, lethargy, seizures, and hypoketotic hypoglycemia. Treatment consists primarily of avoidance of fasting and support during intercurrent illnesses. Some specialists may prescribe vitamin/cofactor supplementation.

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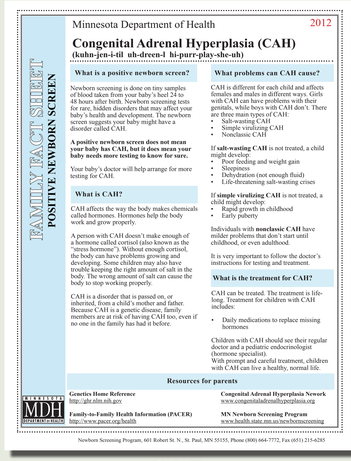
NEW

FAMILY FACT SHEETS


Minnesota Department of Health 2012

FAMILY FACT SHEET
POSITIVE NEWBORN SCREEN

Congenital Adrenal Hyperplasia (CAH)
(kuhn-jen-i-til uh-dreen-l hi-purr-plays-she-uh)

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called CAH.

A positive newborn screen does not mean your baby has CAH, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing for CAH.

What is CAH?

CAH affects the way the body makes chemicals called hormones. Hormones help the body work and grow properly.

A person with CAH doesn't make enough of a hormone called cortisol (also known as the "stress hormone"). Without enough cortisol, the body can have problems growing and developing. Some children may also have trouble keeping the right amount of salt in the body. The wrong amount of salt can cause the body to stop working properly.

CAH is a disorder that is passed on, or inherited, from a child's mother and father. Because CAH is a genetic disease, family members are at risk of having CAH too, even if no one in the family has had it before.

What problems can CAH cause?

CAH is different for each child and affects females and males in different ways. Girls with CAH can have problems with their genitals, while boys with CAH don't. There are three main types of CAH:

- Salt-wasting CAH
- Simple virilizing CAH
- Nonclassic CAH

If **salt-wasting CAH** is not treated, a child might develop:

- Poor feeding and weight gain
- Sleepiness
- Dehydration (not enough fluid)
- Life-threatening salt-wasting crises

If **simple virilizing CAH** is not treated, a child might develop

- Rapid growth in childhood
- Early puberty

Individuals with **nonclassic CAH** have milder problems that don't start until childhood, or even adulthood.

It is very important to follow the doctor's instructions for testing and treatment

What is the treatment for CAH?

CAH can be treated. The treatment is life-long. Treatment for children with CAH includes:

- Daily medications to replace missing hormones

Children with CAH should see their regular doctor and a pediatric endocrinologist (hormone specialist).
With prompt and careful treatment, children with CAH can live a healthy, normal life.

Resources for parents

Genetics Home Reference
http://ghr.nlm.nih.gov

Congenital Adrenal Hyperplasia Newark
www.congenitaladrenalhyperplasia.org

Family-to-Family Health Information (PACER)
http://www.pacer.org/health

MN Newborn Screening Program
www.health.state.mn.us/newbornscreening

MDH
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OLD

Old: only describes the most likely disorder for the abnormal result

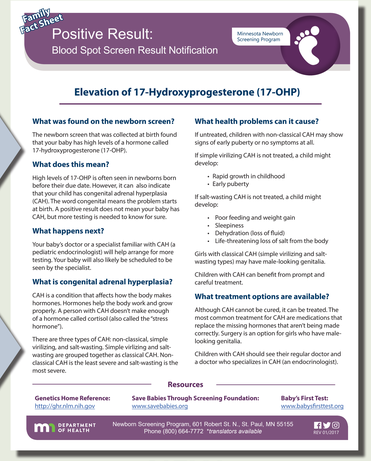
Survey: 94% want families to know what was found on the screen

New: provides actual screening result

Old: minimal discussion about what was found and follow-up plan

Survey: 61% want more information about what to review with the family included

New: reviews the result, what it could mean, and what happens next


Family Fact Sheet

Positive Result:
Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevation of 17-Hydroxyprogesterone (17-OHP)

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of a hormone called 17-hydroxyprogesterone (17-OHP).

What does this mean?

High levels of 17-OHP is often seen in newborns born before their due date. However, it can also indicate that your child has congenital adrenal hyperplasia (CAH). The word congenital means the problem starts at birth. A positive result does not mean your baby has CAH, but more testing is needed to know for sure.

What happens next?

Your baby's doctor or a specialist familiar with CAH (a pediatric endocrinologist) will help arrange for more testing. Your baby will also likely be scheduled to be seen by the specialist.

What is congenital adrenal hyperplasia?

CAH is a condition that affects how the body makes hormones. Hormones help the body work and grow properly. A person with CAH doesn't make enough of a hormone called cortisol (also called the "stress hormone").

There are three types of CAH: non-classical, simple virilizing, and salt-wasting. Simple virilizing and salt-wasting are grouped together as classical CAH. Non-classical CAH is the least severe and salt-wasting is the most severe.

What health problems can it cause?

If untreated, children with non-classical CAH may show signs of early puberty or no symptoms at all.

If simple virilizing CAH is not treated, a child might develop:

- Rapid growth in childhood
- Early puberty

If salt-wasting CAH is not treated, a child might develop:

- Poor feeding and weight gain
- Sleepiness
- Dehydration (loss of fluid)
- Life-threatening loss of salt from the body

Girls with classical CAH (simple virilizing and salt-wasting types) may have male-looking genitalia.

Children with CAH can benefit from prompt and careful treatment.

What treatment options are available?

Although CAH cannot be cured, it can be treated. The most common treatment for CAH are medications that replace the missing hormones that aren't being made correctly. Surgery is an option for girls who have male-looking genitalia.

Children with CAH should see their regular doctor and a doctor who specializes in CAH (an endocrinologist).

Resources

Genetics Home Reference: http://ghr.nlm.nih.gov

Save Babies Through Screening Foundation: www.savebabies.org

Baby's First Test: www.babysfirsttest.org

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NEW

OF NOTE:

No major changes to the following elements: clinical summary, treatment, and expected outcomes as it confirmed their utility.

The majority of surveyed providers felt they did not need a reminder of what newborn screening is in order to help them in their discussion with the family.

OF NOTE:

No major changes to the following elements: symptoms if untreated, treatment options, expected outcomes, and support resources as it confirmed their utility.

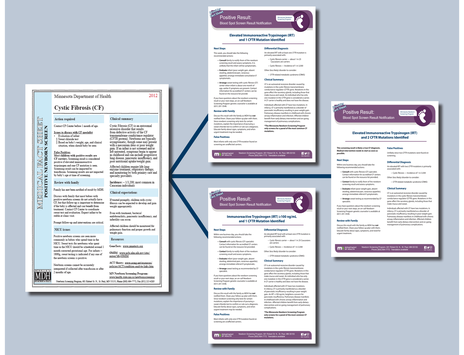
Survey confirmed that providers agree that factors affecting the results (e.g., NICU, environmental, etc.) were not as important to include for families.

BEYOND THE SURVEY

While revising the fact sheets, MDH genetic counselors identified other opportunities for change detailed below and included those in the revisions.

Historically, we did not have a fact sheet for every possible result; only the most common ones. Some of our fact sheets were multi-purposed, for example, a sickle cell disease fact sheet was used anytime S hemoglobin was present in a disease state.

Another example of this is a positive cystic fibrosis result, the same fact sheet was utilized even when the number of mutations and follow-up timeline were not the same. In reality, the follow-up on a one mutation result is very different than a two mutation result. Additionally, the implications of a one mutation result can be very different depending on the immunoreactive trypsinogen level. Thus, we developed fact sheets that better suited the result and the message conveyed to the provider with regards to clinical expectations and how quickly to follow-up on the result.



In addition to this change, we restructured the fact sheets in a more meaningful way. The biggest change was switching the title of the fact sheet from disorder-based to analyte-based. Having an analyte-based fact sheet allows for a discussion of potential differentials and in turn reinforces the fact that this is a screening result and not diagnostic.

Last, while re-writing the content for the family fact sheet, we were mindful to use plain language as much as possible for ease of understanding.

NEXT STEPS

assess what content the families want

ensure provider receipt & family provision

translate into other languages