

Lessons Learned from Carrier Screening Sickle Cell Disease

Consumer Perspectives

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National Institutes of Health

**Population-Based Carrier Screening for Single Gene Disorders
Lessons Learned and New Opportunities**

Trait Notification Letter

We have been collaborating with your doctor regarding your baby XX born on XX. S/he was tested for sickle cell disease by the State Department of Health Newborn Screening Program. The test result showed Hemoglobin FAV. His/her confirmatory test which was taken on XX revealed Hemoglobin F 56.2, Hemoglobin A 20.3 and Hemoglobin D-Los Angeles 17.2%

The presence of a FAD-Los Angeles is an indication of Hemoglobin D trait, which is not a disease, and does not affect your baby's mental or physical health. However...

SCDAA – NCEC

Funding and Purpose

To increase the capacity of the HRSA funded SCD newborn screening community-based programs to provide services to families with babies identified with SCD, or as carriers of SCD or other hemoglobinopathies:

Services include:

- model education
- counseling
- follow-up

SCDAA – NCEC

Intended Outcomes

One of five program priorities: Materials Development

Create materials and methods of information delivery that will increase health literacy, particularly about sickle cell disease and genetics.

Through information created for families and for providers, the NCEC will establish a foundation to disseminate standardized information about sickle cell disease.

Sickle Cell Trait Information

Providing Understandable Information to Consumers

Intended audiences:

- Parents of newborns identified as having trait
- Teens and adults with sickle cell trait

Sickle Cell Trait Information

Issues and Challenges

- The challenge of getting agreement on how to adequately translate complicated genetic information prolonged the production process.
- Issues that arose that required decisions that should possibly be treated as policy, in the future.

Getting Input and Feedback

SCDAA National Coordinating and Evaluation Center
2003-2007

1. Focus groups and discussion groups with parents
2. Surveys and individual interviews with providers
3. SCDAA's Medical and Research Advisory Committee

Sickle Cell Trait Information

Themes and Issues

1. Medical terminology
2. Main messages
3. Accuracy
4. Readability

Medical Terminology Issues

What Do We Call Things?

1. What to call “sickle cell trait”
(“trait”, “carrier”, “AS”)
2. How to explain what sickle cell trait actually *is*
(It happens when.... Or it is a...)
3. Whether not, or how, to talk about other hemoglobinopathies
(AC, AD, AE, A beta-plus thalassemia, A beta-zero thalassemia)

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Medical Terminology Issues

How Do We Explain Things?

1. The “S” gene is the one to pay attention to.
2. “other genes that can combine to cause sickle cell disease”
3. “related trait conditions”

Main Messages

What Is Most Important for People to Know?

And in what order?

1. Don't worry. Your baby does not have sickle cell disease
2. What sickle cell trait is
3. What sickle cell disease is (pathophysiology vs symptoms)
4. What hemoglobin is
5. How you get it. Whether or not it's contagious.
6. You can possibly have a child with SCD in the future
7. Having the trait can affect your child's children...
8. In rare cases, sickle cell trait can cause a few problems
9. Why to get tested
10. How to get tested

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Main Messages

What Is Most Important for People to Know?

What about probabilities?

If each parent is AS:

- The chance of having a baby with sickle cell disease is 25% for *each* baby.
- This is the same as 1 in 4 chances.

Accuracy

How do we adequately simplify complicated information?

1. How 2 parents have a child with sickle cell disease
2. Why to get tested
3. How to get tested

Accuracy

Who Can Have a Child with SCD?

“You can possibly have a baby with Sickle Cell Disease in the future if you and your partner both have Sickle Cell Trait (AS).”

What about:

AS and AC?

AS and A beta-plus thalassemia?

Etc...

Accuracy

How Should People Get Tested?

There are several blood tests that are used

The best ones give detailed results. Solubility tests do not.

Avoiding tests that

Only test for the “S” gene

Do not distinguish between the trait and the disease

May give false negatives

Sickle Cell Trait Information

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Literacy Levels of Adults in America

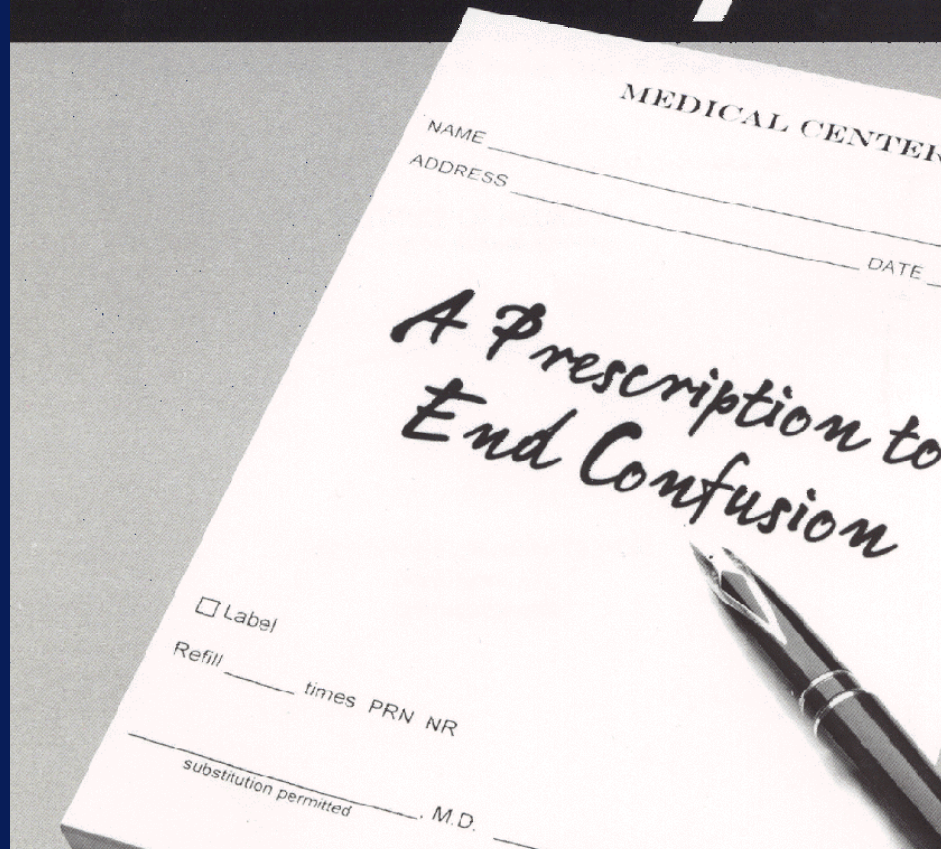
| <u>Reading Ability</u> | <u>NAAL Level</u> | <u>% of Pop.</u> |
|------------------------|-----------------------|----------------------|
| - Below Basic | 1 | 14 |
| - Basic | 2 | 29 |
| - Intermediate | 3 | 44 |
| - Proficient | 4 | 13 |

2003 National Assessment of Adult Literacy (NAAL)
U.S. Department of Education

INSTITUTE OF MEDICINE
OF THE NATIONAL ACADEMIES

Executive Summary

Health Literacy



Before

Date

Ms. «Mothers_First_Name»«Mothers_Last_Name»
«Address»
«City», «State» «Zip»

Re: Baby «Babys_Last_Name»
DOB: «Date_of_Birth»
Unit #: «Med_Rec_No»

Dear Ms. «Mothers_Last_Name»:

All babies born in State of _____ are tested before leaving the hospital for certain genetic or inherited disorders. Sickle Cell Disease/Trait, and other hemoglobin variations are among them. Your child was found to have a **Sickle Cell Trait or Hemoglobin "AS"**. **This is not a disease.** However, it is important for you to be aware of this condition and what it means for your baby.

In order for your baby to have a **Sickle Cell Trait**, it means that baby inherited one normal gene-(Hemoglobin "A"), from one parent and one sickle gene-(Hemoglobin "S") from the other parent. It is important that both you and baby's dad are tested for sickle cell trait, so that you know what your risk is for having a child with Sickle Cell Disease. **Sickle Cell Trait** usually causes no health problems. It is important for you, the parents, to know your hemoglobin status and how it could affect future pregnancies, as well as recognize that when your infant becomes of childbearing age that he/she understands the risk of having children with **Sickle Cell Disease**.

It is vital that you contact our office in the week or two to make an appointment so that we may discuss sickle cell trait and what it means for your baby. **Although rare, Sickle Cell Trait can result in some medical complications.** You will find that this brief session will provide you with the information you need to give your child the appropriate information when the time is right. Please contact us at xxx-xxx-xxxx for an appointment and more information. Inform our receptionist that you have received a letter from us indicating that your baby has **Sickle Cell Trait**. In addition, please inform your child's primary care physician that your child has Sickle Cell Trait.

We look forward to hearing from you.

Sincerely,

Coordinator, Community-Based Sickle Cell Project
Director of Pediatric Hematology/Oncology

After

Date _____

Ms. Mothers_First Name Mothers Last Name

Address _____

City, State Zip _____

Re: Baby Last Name, First Name

DOB: Date_of_Birth _____

Unit #: Med_Rec_No _____

Dear Ms. Mothers Last Name

This letter is to let you know about your baby's blood screening results.

Blood screening results

- All babies born in the state of _____ get screened for sickle cell hemoglobin (pronounced he-mo-glo-bin) right after they are born.
- Your baby's blood screening results shows that your baby has sickle cell trait.

Here are 6 things you should do:

1. **Do not be alarmed. Do not worry.**
 - This is NOT a disease.
 - Some people think that sickle cell trait is a disease. But this is not so.
 - A few people have problems with the trait. But this is rare.
2. **Think about getting each parent tested if you may have another baby.**
 - If your child has sickle cell trait, it means that **at least one** parent carries the sickle cell gene.
 - If **both** parents carry the sickle cell gene, it means that you could have a baby with sickle cell disease in the future. This is something to think about.
3. **Get more information, if you need it.**
 - Read the pamphlet that comes with this letter.
 - Call _____ if you have questions.
 - Come to one of our counseling sessions:
Place: _____ Time: _____
4. **Share this letter with your baby's doctor.**
5. **Keep this letter for your records.**
6. **Keep this information for your child. When he or she is old enough make them aware of their own chance of having children with sickle cell disease.**

We look forward to hearing from you.

Sincerely

Coordinator, Community-Based Sickle Cell Project Director of Pediatric Hematology/Oncology

Printed Materials

1. Text

short in length, simple language, clear directives

2. Layout and typography

Ample white space, well-organized, bulleted points

3. Illustrations

present, useful, clear

4. Conceptual appeal

interesting, practical

Lessons Learned

1. Issues involved in giving people accurate, useful and understandable information about “sickle cell trait” are not straightforward.
2. SCDA/NCEC has an opportunity to sort out these issues and offer guidance on what terminology to use and what messages to convey.
3. Information providers, in turn, have an opportunity to teach consumers new terms and to clarify misconceptions about trait information.