Attachment 7
Introductory Letter and Instructions to Parents

Dear Parent,

Newborn screening is an important health test for all babies. A few drops of blood from the baby's heel are placed on a special card and sent for testing. The tests can find serious conditions, such as phenylketonuria (PKU), that are not apparent at birth. If found early, simple treatments can save lives or prevent serious health problems.

On occasion, a disease goes undetected or is later found by a clinical examination. Such missed cases may be the result of a program error or some other factor. For example, a variant of the disorder may not have the chemical marker used for screening. Or, a disaster, such as the September 11 situation, may cause screening to take a longer time than usual.

This letter asks for your help with a research study that will attempt to identify late diagnosed cases that should have been identified by newborn screening. The study will classify the cause of the delay in diagnosis or treatment in order to improve screening across the country. Only conditions mandated at the time of the birth will be included. A listing of these disorders is included with this letter.

Delayed diagnosis or delayed treatment (a missed case) is defined as a sick (or dead) child not identified by newborn screening or who was treated after a long delay following birth even if the newborn screening test identified the disorder. This study is interested in the entire newborn screening system from heel stick through diagnosis and medical management. While not all delays result in poor outcomes, it is important to identify reasons for delay so that the system can be improved.

Facts about missed cases will be collected from laboratory directors, follow-up coordinators, metabolic clinics, and parent groups. A Case Report Form is included with this letter. Please help by reporting one or more cases of delayed diagnosis or treatment (missed cases) occurring from 1984 and 2004. Completing the form should take about 10 to 15 minutes. Please do not submit information that you know has already been submitted by someone else.

Because information about missed cases is sensitive and is not without its liabilities, we are taking several steps to protect your privacy and the privacy of any patients. The facts you give us will not be linked to you. Your name or other facts that might point to you or the child will not appear in our records or any presentations of publications. We ask only for basic case facts: the disease; the month/year of birth; the reason the diagnosis or treatment was delayed (if known); state of birth; and any health and legal outcomes. Please do not send your name, names of children or their families, child's date of birth or the names of towns or cities where the cases occurred. Any facts given will remain private. Data will be kept in locked files, and only staff from this study will be allowed access.

You are free to join or not to join this study. There is no penalty if you choose not to join. You may quit at any time with no penalty. This letter functions as informed

consent. We will take the return of a data collection form as your consent to be a part of this study.

If you have any questions about the study, please contact Dr. L. Omar Henderson at (770) 488-7972 If you have questions about your rights as a participant in this research study or believe you have been injured by this study, please contact the office of CDC's Deputy Associate Director for Science at (800) 584-8814. Please leave a brief message including your name and phone number. Say that you are calling about CDC protocol # 3618. Your call will be returned as soon as possible.

We hope you will help in improving newborn screening systems by sharing any information you may have on late diagnosed or treated cases. Please use a different Case Report Form for each case (you may write us a letter if you cannot print the form). Return the form(s) to:

L. Omar Henderson, Ph.D.
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Centers for Disease Control and Prevention
4770 Buford Highway, N.E., Mailstop F-43
Atlanta, GA 30341-3724

by [insert date]. If you have any questions, please do not hesitate to contact us.

Thank you for your help!

Sincerely,

L. Omar Henderson, Ph.D.; CDC
Lisa Kalman, Ph.D.; CDC
Harry Hannon, Ph.D.; CDC
Scott Grosse, Ph.D.; CDC
Brad Therrell, Ph.D.; NNSGRC
Kenneth Pass, Ph.D.

L. Omar Henderson, Ph.D.

Centers for Disease Control and Prevention
4770 Buford Highway, N.E., Mailstop F-43
Atlanta, GA 30341-3724
(770) 488-7972 (phone)
(770) 488-4255 (fax)

Instructions

For this study, the children of interest were diagnosed with a screened metabolic disorder identified outside of the newborn screening system (i.e. clinically) or identified through newborn blood spot screening but with a delay in diagnosis or initiation of treatment. These children must have been born in the United States, Puerto Rico, or the Virgin Islands and have a clinical diagnosis of classical phenylketonuria, primary congenital hypothyroidism, galactosemia (any type if law is not restrictive), maple syrup urine disease, homocystinuria, biotinidase deficiency, classical congenital adrenal hyperplasia (saltwasting or simple virilizing), or sickle cell disease if the states where they were born screened for these disorders at the time of the child's birth.

- Please fill out the following Case Report Form to help us get this information.
- If you are telling us about more than one case, please copy the form before you fill it out and use a separate form for each case. You may send us a letter if you are unable to print the form.
- Please return the form(s) to the following address:

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Centers for Disease Control and Prevention
4770 Buford Highway, N.E., Mailstop F-43
Atlanta, GA 30341-3724
(770) 488-7972 (phone)
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Thanks for your help!