National MPS Society
Support for Families. Research for a Cure.

July 15, 2020

Dear MPS Family,

The National MPS Society is working with researchers at the University of Washington to improve precision in newborn screening for MPS. The researchers would like to obtain the newborn dried blood spot from your state's newborn screening lab from the affected patient(s) in your family.

At birth, babies have blood samples collected and tested for genetic conditions. These samples are stored as drops of blood on a card. This study will collect a single dried blood spot from the card. Using mass spectrometry, researchers will measure the level of specific biomarkers (glycosaminoglycans) in the blood sample from birth. The goal of the study is to see if the levels of specific biomarkers present at birth can predict the type and severity of MPS. No additional blood work or lab tests are needed to participate; all analysis will be done on samples that already exist.

This information is critical to improve newborn screening and diagnosis of MPS. Several states have started to screen for MPS I and II, and so the study is timely and will be useful for all syndromes. This study has IRB approval through August 2021.

If you would like to participate in this study, please follow the instructions listed below.

Return all forms to leslie@mpssociety.org or mail to
NATIONAL MPS SOCIETY, PO Box 14686, Durham NC 27709.

1. Review all forms in your state-specific packet.
2. Read and sign the **UNIVERSITY OF WASHINGTON CONSENT FORM** if you consent to participating.
3. Complete the **UNIVERSITY OF WASHINGTON QUESTIONNAIRE FOR STUDY** form.
4. Complete the **DRIED BLOOD SPOT REQUEST FORM for your state's newborn screening laboratory.** It has been partially filled out in some cases and you may disregard any dates provided on the form. Some states ask for additional items (such as a copy of the parent's driver's license), so please include these if requested by your state.
5. Return all forms to the National MPS Society by email or mail.

If you have questions about the study, contact Leslie Urdaneta at leslie@mpssociety.org, Terri Klein at terri@mpssociety.org, or call our office at 919-806-0101.

Thank you in advance for your time and assistance.

Sincerely,

Leslie Urdaneta

Leslie Urdaneta, MSW, LCSW
Family Program Director
919-806-0101
leslie@mpssociety.org

PO Box 14686
Durham, NC
27709-4686
t: 877.MPS.1001
p: 919.806.0101
f: 919.806.2055
www.mpssociety.org

UNIVERSITY OF WASHINGTON CONSENT FORM

Glycosaminoglycan Levels in Newborn Dried Blood Spots from MPS Patients

Researcher: Michael H. Gelb, PhD, Professor, Dept. of Chemistry, Univ. of Washington

office phone: (206) 543-7142

If you are a parent providing permission for a child, "you" in this form means your child

Researchers' statement

We are asking you to be in a research study. The purpose of this consent form is to give you the information you will need to help you decide whether or not to be in the study. Please read the form carefully. You may ask questions about the purpose of the research, what we would ask you to do, the possible risks and benefits, your rights as a volunteer, and anything else about the research or this form that is not clear. When all your questions have been answered, you can decide if you want to be in the study or not. This process is called "informed consent." We will give you a copy of this form for your records. You can direct your questions to the staff member of the National MPS Society who sent you this consent form.

PURPOSE OF THE STUDY

Screening of newborn infants for diseases that can be treated very early in life is now done routinely in many countries, including the United States. Screening does not diagnose an infant. Screening identifies those infants at increased risk (those more likely) to be affected based on substances found in their blood shortly after birth. By identifying those infants at increased risk, testing to confirm or rule out the diagnosis can be done. Then, if the child is affected, treatment can be started before major health problems happen.

Diseases can occur when the body is missing an enzyme (a type of protein), and the body cannot break down certain substances correctly. As a result, these substances can be stored throughout the body. One group of disorders that occur due to missing or non-working enzymes are called "lysosomal storage diseases" or "LSDs." (The lysosome is a part of the cell which contains many enzymes. If one is not working, then material is stored in the lysosome and the body).

New treatments are becoming available for LSDs, so it is important to diagnose individuals with these diseases as early as possible. At the University of Washington, new tests are being developed to use for newborn screening for LSDs.

Newborn screening for a subset of LSDs called Mucopolysaccharidoses is carried out by measuring the amount of residual lysosomal enzymatic activity in newborn dried blood spots. When the enzyme is below the cutoff for the screen, additional tests may be performed to determine how likely it is for the newborn to develop one of the Mucopolysaccharidoses syndromes. In our new study we are exploring whether the level of a biomarker called

glycosaminoglycan is elevated in patients that went on to develop a Mucopolysaccharidosis syndrome. We need to evaluate the biomarker level in newborn dried blood spots since that is what will be available in a newborn screening program. We are thus asking you to participate in our study by requesting a stored dried blood spot from your state's newborn screening lab. The lab will send the dried blood spot to Professor Gelb's lab at the Univ of Washington so that his lab can measure the level of biomarkers. We also want to see if there is a correlation between the level of the biomarker and the age of onset of symptoms for the Mucopolysaccharidosis syndrome. This correlation may be useful someday in predicting the severity of the disease in newborns who test postive in newborn screening. We want to focus on the newborn dried blood spot since that is what is most relevant for newborn screening.

_____

STUDY PROCEDURES

If you choose to take part in this study, you would complete your state's form for request of a stored dried blood spot from you the patient or your affected child. We are providing you with a partially completed form from your state's newborn screening lab. You would also complete the patient questionaire form, and send this along with the completed dried blood spot release form and the signed consent form to the staff member of the National MPS Society.

Professor Gelb will never know the identity of the patient, all dried blood spots that his lab will receive will have a code number. The Natioanal MPS Society will have a list of patient names linked to their code and also linked to the questionaire that you will fill out to provide important information about the patient. This questionaire information will be provided to Professor Gelb so that he can interpret the data in his research study. Again, Professor Gelb's research team will never know the identity of the patient.

The only test the Gelb lab will perform on the dried blood spot is the measurement of the glycosaminoglycan biomarker.

RISKS, STRESS, OR DISCOMFORT

You may find that participating in a research study is an invasion of your privacy. We will make every effort to keep all of the information we collect for this study about you safe. More information of confidentiality is described under the CONFIDENTIALITY OF RESEARCH INFORMATION section of this form.

ALTERNATIVES TO TAKING PART IN THIS STUDY

Taking part in this study is voluntary. You do not have to take part if you do not want to.

BENEFITS OF THE STUDY

While you will not directly benefit from this study, we hope that the results of this study will provide important new information that can be used to improve the knowledge gained from newborn screening related to lysosomal storage diseases.

SOURCE OF FUNDING

The study team and/or the University of Washington is receiving financial support from the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK), National Institutes of Health (NIH).

FINANCIAL INTEREST

None.

CONFIDENTIALITY OF RESEARCH INFORMATION

The only individuals who will know the identity of the patients are the administrative staff of the National MPS Society. They will keep these names and code numbers in a secure place.

We have a Certificate of Confidentiality from the National Institutes of Health. This helps us protect your privacy. The Certificate means that we do not have to give out identifying information about you even if we are asked to by a court of law. We will use the Certificate to resist any demands for identifying information.

We can't use the Certificate to withhold your research information if you give your written consent to give it to an insurer, employer, or other person. Also, you or a member of your family can share information about yourself or your part in this research if you wish.

There are some limits to this protection. We will voluntarily provide the information to: • a member of the federal government who needs it in order to audit or evaluate the research;

• individuals at the University of Washington, the funding agency, and other groups involved in the research, if they need the information to make sure the research is being done correctly;
• the federal Food and Drug Administration (FDA), if required by the FDA;

• Local authorities, if we learn of child abuse, elder abuse, or the intent to harm yourself or others.

If we publish the results of the study in scientific journals or present them at scientific meetings, we will not include any information that could identify you.

OTHER INFORMATION

You may refuse to participate in any or all portions of this study.You are also free to withdraw from this study at any time without penalty or loss of benefits to which you are otherwise entitled.

You will not be charged for study-related procedures. You will not be paid for taking part in this study.

If you have any questions about the study, contact the staff person of the National MPS Society. Do not contact Professor Gelb, since he is not to learn the identities of the families.

I consent to the research study described in this consent form.

Printed name:_____

Signature:_____

Date:_____

CONSENT FOR RELEASE OF DRIED BLOOD SPECIMEN FROM GDSP

The undersigned hereby authorizes the release of the Newborn Screening Specimen from the records of the Genetic Disease Screening Program (GDSP).

FOR NEWBORN PATIENT

Name: _____

Gender: ☐ Male ☐ Female Twin: ☐ Yes ☐ No Date of Birth: _____

Hospital Of Birth: _____

Mother's Full Name (including maiden name): _____

Mother's Date of Birth: _____

Patient's Address at Time of Birth: _____

_____

RELEASE TO

Requestor Name: Michael Gelb Phone: 206 543-7142

Requestor Email: gelb@uw.edu

Ship Attention To: Mike Gelb

Facility Name and Address: Dept. of Chemistry, Campus Box 351700, 36 Bagley Hall

Univ. of Washington, Seattle, WA 98195

Facility Phone: 206 543 7142 Facility Fax #: 206 685-8665

REASON FOR REQUEST

Research study of the levels of glycosaminoglycans in MPS patient DBS

This authorization will expire on (Enter Date): August 1, 2021.

You have the right to retain a copy of this consent. You have the right to revoke this consent at any time by writing to: Chief, Genetic Disease Screening Program at 850 Marina Bay Parkway, Richmond, CA 94804, as stated in our privacy notice. Revocation of this consent does not eliminate your responsibilities for payment for services received. The Genetic Disease Screening Program is not responsible for further disclosures of the information by other parties that may result from complying with this consent.

_____ _____

(Parent/Patient/Legal Guardian Signature) (Date)

I understand that any person who requests or obtains any record containing personal information from the California Department of Public Health under false pretenses will be guilty of a misdemeanor and fined up to $5,000 or imprisoned up to one year or both.
Please See Privacy Notification on Reverse

NOTICE OF INFORMATION AND PRIVACY PRACTICES
California Department of Public Health (CDPH)
Genetic Disease Screening Program (GDSP)
The California Newborn Screening Program Note
Effective Date: June 2014

THIS NOTICE DESCRIBES HOW PERSONAL AND MEDICAL INFORMATION ABOUT YOU OR YOUR NEWBORN MAY BE USED AND DISCLOSED AND HOW YOU CAN GET ACCESS TO THIS INFORMATION. PLEASE REVIEW IT CAREFULLY.

Department's Legal Duty
Federal and State laws restrict the use, maintenance, and disclosure of personal and medical information obtained by a State agency and requires certain notices to individuals whose information is maintained.

State laws include the California Information Practices Act (Civil Code 1798 et seq.), Government Code Section 11015.5 and Health and Safety Code Section 124980. The federal law is the Health Insurance Portability and Accountability Act of 1996 (HIPAA), 42 USC 1320d-2(a) (2), and its regulations in Title 45 Code of Federal Regulations Sections 160.100 et seq. In compliance with these laws, you and those providing information are notified of the following.

Department Authority and Purpose for the Newborn Screening Program
The CDPH collects and maintains specimens and information related to newborn screening as permitted in Health and Safety Code Sections 124980, 124977, 124991, 125000, 125001, 125025, and 125030. This information is collected electronically and includes such things as your name, address, medical care given to you and your newborn. Testing is required by law (Health and Safety Code Section 125000) and regulations (17 CCR 6500 through 6510) and if the required information is not provided, serious illness or permanent damage for affected newborns could result.

If you have religious objections to this testing, you may say "no" to the testing in writing and sign a form advising you that your hospital, doctor, and clinic staff are not responsible if your baby develops problems because those disorders were not identified and treated early.

Uses and Disclosure of Health Information
The CDPH uses health information about you or your newborn for screening, to provide health care services, to obtain payment for screening, for administrative purposes, and to evaluate the quality of care that you or your newborn receives. Some of this information is retained for as long as 21 years. The information will not be sold.

The law also allows the Department to use or give out newborn screening specimens and/or general health information about you or your baby, for department-approved studies, such as research related to preventing disease. The material will be provided**, without any personal identifying information.** Researchers can only apply to receive

the information if they have been approved by an institutional review board (IRB) and meet all federal and state privacy law requirements.

The Department is authorized by law to charge approved researchers a fee to recover all the expenses related to the research request (including data linkage, retrieval, data processing, data entry, re-inventory, shipping of blood samples, and related data management).

The Department reserves the right to change the terms of this notice and to make the new notice provisions effective for all protected health information that it maintains. The most current Privacy Notice can be found at the Newborn Screening Program website: www.cdph.ca.gov/programs/nbs. You may request a copy of the current policies or obtain more information about our privacy practices, by calling the numbers listed below or consulting the Program website. You may also request a paper copy of this Notice. This Privacy Notice can also be found at the website: www.ca.gov/programs/pages/Privacyoffice.aspx.

Individual Rights and Access to Information

The Newborn Screening Program must have your written permission to use or give out personal or health information about you for any reason that is not described in this notice. You can revoke your authorization at any time, except if the Newborn Screening Program has already acted because of your permission by contacting the Chief of the Genetic Disease Screening Program at 850 Marina Bay Parkway, F175, Richmond, CA 94804.

You have the right to look at or receive a copy (you will be charged) of your or your newborn's health information and receive a list of instances where we have disclosed health information about you or your newborn for reasons other than payment for screening or related administrative purposes.

You have a right to ask that the Newborn Screening Program contact you only in writing, or at a different address, post office box, or telephone number. Newborn Screening Program will contact you the way you have asked if this is necessary to keep you safe.

You have a right to ask the Newborn Screening Program not to use or share your or your newborn's information and/or specimen in the ways listed in this notice. However, we may not be able to comply with your request.

You have a right to have information in your or your child's records changed if information is missing or you believe the information is incorrect.

Complaints

We will let you know promptly if a breach occurs that may have compromised the privacy or security of your information. If you believe that we have not protected your privacy or have violated any of your rights and wish to file a complaint, please call or

write to the: Privacy Officer, CA Department of Public Health, P.O. Box 997377, MS 0506, Sacramento, CA 95899-7377, (916) 440-7671 or (877) 421-9634 TTY/TDD.

You may also contact the United States Department of Health and Human Services, Attention: Regional Manager, Office for Civil Rights at 90 7th Street, Suite 4-100, San Francisco, CA 94103, telephone (800) 368-1019, or the U.S. Office of Civil Rights at 866-OCR-PRIV (866-627-7748) or 866-788-4989 TTY.

The Department cannot take away your health care benefits or any other protected rights in any way if you choose to file a complaint or use any of the privacy rights in this notice.

Department Contact
The information on this form is maintained by the California Department of Public Health, Genetic Disease Screening Program. Please address correspondence to the Chief of the Genetic Disease Screening Program, 850 Marina Bay Parkway, F175, Mail Stop 8200, Richmond, California, 94804 (510-412-1502).

Electronic Copies of this Notice: To get a copy of this notice in an electronic format call or write to:

Chief, Genetic Disease Screening Program
850 Marina Bay Pkwy, F175, Mail Stop 8200, Richmond, CA 94804
Phone: 510-412-1502 Relay Operator 711/1-800-735-2929

UNIVERSITY OF WASHINGTON QUESTIONNAIRE FOR STUDY:

Glycosaminoglycan Levels in Newborn Dried Blood Spots from MPS Patients

Researcher: Michael H. Gelb, PhD, Professor, Dept. of Chemistry, Univ. of Washington

In order for our research study to lead to interpretable results, we require some specific information about the MPS patient, and would like to ask that you provide the information listed below. Please only complete this form if you are the MPS patient or have legal permission to provide answers for the patient (ie. a parent or legal guardian).

1. Circle the name of the disorder:

MPS-I
MPS-II
MPS-IIIA
MPS-IIIB
MPS-IIIC
MPS-IIID
MPS-IVA
MPS-VI
MPS-VII
MLD
MSD

2. Sex: Male_____, Female_____

3. Current age of the patient: _____yrs, _____months.

4. Approximate age when first symptoms of the disease were noticed : years____, months_____

5. List the first symptoms:

6. List the current symptoms:

7. Approximate age of the patient when the diagnosis by the medical expert was made: years_____, months_____

8. If you have the patient's genotype, please put it below:

_____

9. If you don't know the patient's genotype, please contact your family's physician if you think he/she has the genotype and write it below or indicate that the genotype was never obtained (as far as you known).

_____

_____

Sample label (leave blank, this will be provided by the staff member of the National MPS Society:

_____