



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@mppsociety.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@mppsociety.org, Terri Klein at terri@mppsociety.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
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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 positive 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 questionnaire 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 National MPS Society will have a list of patient names linked to their code and also linked to the questionnaire that you will fill out to provide important information about the patient. This questionnaire 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: _____

NEWBORN SCREENING SPECIMEN REQUEST FORM FOR IDENTIFIED SPECIMENS

(Research / Test Development)

1. **Requestor/Title/Institution:**
2. **Phone Number:**
3. **Contact, if different from the Requestor:**
4. **Contact's Phone Number:**
5. **Date of Request:**
6. **Date Required:**
7. **Number of Specimens:**
8. **Participants' Information (attach a copy of the signed consent form for each patient)**

Name	Birth Date	Gender	Hospital of Birth
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9. **Spot Size/Number of Punches per Infant (one full circle must remain on the card; if there is insufficient blood, the sample is considered ineligible for study):**
10. **Preparation (e.g. the arrangement of samples on the plate (Investigators are asked to supply tubes/plates; the arrangement of vacant wells for controls; request for DNA extraction, if applicable. Fee may be assessed depending on amount of preparation and/or number of specimens).**

11. **Study title / Description / Protocol number / Rationale for request:**

12. **Requestor Attestation:**

I affirm that the above information is accurate and the requested specimens will be used for the exempt and/or currently approved IRB study (see #11):

Signature: _____ **Date:** _____

13. Verification by Requestor's IRB if Requestor's Institution is not NYSDOH

Study was last approved by this IRB on (date): _____

7/2020: IRB renewed
through August 2021

I affirm that the requested specimens and their use are consistent with the objectives of study
(<<insert number and title>>).

IRB Program Staff

Name: _____ Title: _____

Signature: _____ Date: _____

14. Verification by NYSDOH IRB:

This specimen request is consistent with the objectives of this study (<<insert number and
title>>):

Study last approved by IRB on (date): _____

IRB Program Staff

Name: _____ Title: _____

Signature: _____ Date: _____

15. Approval of Request for Specimens, Newborn Screening Program:

NBS Program Staff Name: _____ Title: _____

Signature: _____ Date: _____

16. Fulfillment Date:

NBS Program Staff Name: _____ Title: _____

Signature: _____ Date: _____

17. Fee: _____ 18. Specimen linkage secured date: _____

Signature: _____

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:
