



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](mailto: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](mailto:leslie@mppsociety.org), Terri Klein at [terri@mppsociety.org](mailto: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  
919-806-0101  
[leslie@mppsociety.org](mailto:leslie@mppsociety.org)

PO Box 14686  
Durham, NC  
27709-4686  
t: 877.MPS.1001  
p: 919.806.0101  
f: 919.806.2055  
[www.mppsociety.org](http://www.mppsociety.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 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: \_\_\_\_\_

# AUTHORIZATION TO DISCLOSE PROTECTED HEALTH INFORMATION – NEWBORN SCREENING

Michigan Department of Health and Human Services

**Directions:** Type or print all requested information, with the exception of signatures on page 2.

Individual's Name (Beneficiary, Recipient, Patient, Consumer, etc.)			Individual's ID Number (Medicaid, SSN, Other)
Street Address			Individual's Date of Birth
City	State	ZIP Code	Phone

## I AUTHORIZE THE FOLLOWING PERSON/ORGANIZATION TO SHARE MY HEALTH INFORMATION WITH THE MICHIGAN DEPARTMENT OF HEALTH AND HUMAN SERVICES (MDHHS):

Name of Person/Organization			
Street Address			
City, State, ZIP Code		Phone Number	Fax Number
		- -	- -

### HEALTH INFORMATION TO BE SHARED:

List the amount or type of information you would like to share in the section below.

For example, you can say all my health information or list certain types of information you would like to share.

<u>A full circle, newborn dried blood spot from the person named at the top of this form.</u>

### MY HEALTH INFORMATION WILL BE SHARED FOR THE FOLLOWING REASON:

For example, to discuss my health care benefits or at the request of the individual.

<u>The dried blood spot will be used in a research study to help increase the precision of newborn screening and diagnosis of MPS disorders.</u>

### By signing this form, I understand that:

<ul style="list-style-type: none"><li>I do not have to sign this authorization and my refusal to sign this authorization will not affect my ability to obtain treatment, payment for services, enrollment, or eligibility benefits.</li><li><b>Information regarding behavioral and mental health services, substance use disorder treatments, and communicable diseases such as sexually transmitted diseases and human immunodeficiency virus (HIV infection, Acquired Immune Deficiency Syndrome or AIDS related complex) may be shared if I initial here or if I list this type of information above _____.</b></li><li>If I authorize the release of substance use disorder treatment information, the recipient cannot re-disclose this information without my permission unless permitted under federal or state law.</li><li>Other types of information shared under this authorization may be re-disclosed by the person or organization I identified above and may no longer be protected by federal or state law.</li><li>I may change my mind and revoke (take back) this authorization at any time. To revoke this authorization, write to the MDHHS program that maintains your records and include a copy of page 1 of this form.</li><li>Information that has already been shared based on this authorization cannot be taken back.</li><li>I may request a copy of this signed authorization. If I have not previously revoked this authorization, it will expire on (list a date, event or condition):  _____</li></ul> <p>Authorization will expire one year from the signature date if you leave this section blank.</p>
--

Signature of Individual or Legal Representative	Date (Month/Day/Year)
Name of Individual or Legal Representative	
Legal Representative's Relationship to Individual (i.e., parent, guardian, patient advocate, authorized representative, power of attorney. Documentation may be required.)	

**AUTHORIZED PERSON/ORGANIZATION USE ONLY**

This authorization was revoked:

Signature	Date (Month/Day/Year)
-----------	-----------------------

The Michigan Department of Health and Human Services (MDHHS) does not discriminate against any individual or group because of race, religion, age, national origin, color, height, weight, marital status, genetic information, sex, sexual orientation, gender identity or expression, political beliefs or disability.

**AUTHORITY:** This form is acceptable to the Michigan Department of Health and Human Services as compliant with HIPAA privacy regulations, 45CFR Parts 160 and 164 as modified August 14, 2002.

**COMPLETION:** Is voluntary, but required if disclosure is requested

**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:

---