MAYO CLINIC

Name and Clinic Number

Approval Date: October 20, 2020

RESEARCH PARTICIPANT CONSENT AND PRIVACY AUTHORIZATION FORM

Study Title: Clinical and Basic Investigations into Congenital Disorders of Glycosylation

IRB#: 19-005187

Principal Investigator: Eva Morava-Kozicz, MD, PhD and Colleagues

Key Study Information

This section provides a brief summary of the study. It is important for you to understand why the research is being done and what it will involve before you decide. **Please take the time to read the entire consent form carefully and talk to a member of the research team before making your decision.** You should not sign this form if you have any questions that have not been answered.

It's Your Choice	This is a research study. Being in this research study is your choice; you do not have to participate. If you decide to join, you can still stop at any time. You should only participate if you want to do so. You will not lose any services, benefits or rights you would normally have if you choose not to take part.
Research Purpose	The purpose of this research is to study the natural history of congenital disorders of glycosylation (CDG) and its causes and treatments. You have been asked to take part in this research because you have been diagnosed with CDG.
What's Involved	Study participation involves use of your medical record, residual or left over samples that were collected as part of your clinical care and a yearly questionnaire to better understand congenital disorders of glycosylation (CDG).
Key Information	There are no immediate risks to you except the potential that some of your private health information or biological data might be leaked or made public despite our best efforts to keep it private and confidential. You may be reimbursed for travel and lodging expenses up to $700 per year (1 visit per year).

Learn More	If you are interested in learning more about this study, read the rest of this form carefully. The information in this form will help you decide if you want to participate in this research or not. A member of our research team will talk with you about taking part in this study before you sign this form. If you have questions at any time, please ask us.

Making Your Decision

Taking part in research is your decision. Take your time to decide. Feel free to discuss the study with your family, friends, and healthcare provider before you make your decision. Taking part in this study is completely voluntary and you do not have to participate.

If you decide to take part in this research study, you will sign this consent form to show that you want to take part. We will give you either a printed or electronic copy of this form to keep.

If you are signing this consent form for someone else, "you" in the consent form refers to the participant.

For purposes of this form, Mayo Clinic refers to Mayo Clinic in Arizona, Florida and Rochester, Minnesota; Mayo Clinic Health System; and all owned and affiliated clinics, hospitals, and entities.

Contact Information

If you have questions about …	You can contact …
▪ Study tests and procedures ▪ Materials you receive ▪ Research-related appointments ▪ Research-related concern or complaint ▪ Research-related injuries or emergencies ▪ Withdrawing from the research study	**Principal Investigator(s):** Dr. Eva Morava-Kozicz, **Phone:** (507) 266-2967 **Study Team Contact:** Kaitlin Schwartz **Phone:** (507) 293-9114 **Institution Name and Address:** Mayo Clinic 200 First Street SW Rochester, MN 55905
▪ Rights of a research participant	**Mayo Clinic Institutional Review Board (IRB)** **Phone:** (507) 266-4000 **Toll-Free:** (866) 273-4681
▪ Rights of a research participant ▪ Any research-related concern or complaint ▪ Use of your Protected Health Information ▪ Stopping your authorization to use your Protected Health Information ▪ Withdrawing from the research study	**Research Subject Advocate (RSA)** **(The RSA is independent of the Study Team)** **Phone:** (507) 266-9372 **Toll-Free:** (866) 273-4681 **E-mail:** researchsubjectadvocate@mayo.edu
▪ Billing or insurance related to this research study	**Patient Account Services** **Toll-Free:** (844) 217-9591

A description of this clinical trial will be available on http://www.ClinicalTrials.gov, as required by U.S. Law. This Website will not include information that can identify you. At most, the Website will include a summary of the results. You can search this Website at any time.

Why are you being asked to take part in this research study?

You are being asked to take part in this research study because you have been diagnosed with congenital disorders of glycosylation (CDG).

About 150 people will take part in this research study from across all sites participating. .

Why is this research study being done?

The purpose of this research is to study the natural history of congenital disorders of glycosylation and its causes and treatments.

Information you should know

Who is Funding the Study?

The National Institutes of Health (NIH) is funding the study. NIH will pay the institution to cover costs related to running the study.

How long will you be in this research study?

This is an ongoing research study with no determined end date.

What will happen to you while you are in this research study?

If you agree to be in the study, you will be asked to participate in the following:

- We will ask you to follow up with us annually (once per year) over the course of 5 years.
- We will review your medical record for research. We may also ask you to allow us to collect and review medical records from other health care providers related to your CDG. After signing the consent form, we will ask you to sign an Authorization to Release Protected Health Information - Research form authorizing us to request your records from your local provider as part of this study. We will also ask you to fill out surveys and/or questionnaires.
- In the future, you may be contacted by phone or email for follow-up information and questionnaires about your CDG, treatment, or well-being. We may ask you to answer the questionnaires over the phone if you do not plan on returning to Mayo Clinic.
- If any blood, urine, stool, or tissue samples are taken as part of your regular medical care, we may use your leftover samples for research to better understand CDG and assist the laboratory in developing testing to better identify these disorders.

Optional Blood, Stool, and Urine Collection

You are also being asked to take part in optional blood, stool, and urine collections. This will be used to test for biomarkers related to congenital disorders of glycosylation (CDG).

If you choose to participate in optional blood, stool, and urine collections, they will be collected as follows:

- Up to 30 mL (2 tablespoons) of blood will be drawn each year. The amount of blood drawn will depend on your age and weight but no more than 2 tablespoons will be collected for each blood draw. If you are unable to provide blood due to age or other logistical difficulties, we will forego this collection.
- You will be asked to collect a random urine sample each year. If you are unable to provide urine due to age or other logistical difficulties, we will forego this collection.
- You will be asked to collect a random stool (feces) collection each year. If you are unable to provide this sample due to age or other logistical difficulties, we will forego this collection.

You do not have to be in the Optional Blood, Stool, and Urine Collection to participate in the main study.

Please read the following statements and mark your choice:

1. I agree to participate in the Optional Blood, Stool, and Urine Collection:

☐ Yes ☐ No Please initial here: _____Date: _____

What are the possible risks or discomforts from being in this research study?

As with all research, there is a chance that confidentiality could be compromised; however, we take precautions to minimize this risk.

The risks of drawing blood include pain, bruising, lightheadedness, and/or fainting, or rarely, infection at the site of the needle stick.

Are there reasons you might leave this research study early?

You may decide to stop at any time. You should tell the Principal Investigator if you decide to stop and you will be advised whether any additional tests may need to be done for your safety.

In addition, the Principal Investigator or Mayo Clinic may stop you from taking part in this study at any time:

- if it is in your best interest,
- if you don't follow the study procedures,
- if the study is stopped.

If you leave this research study early, or are withdrawn from the study, no more information about you will be collected; however, information already collected about you in the study may continue to be used.

We will tell you about any new information that may affect your willingness to stay in the research study.

What if you are injured from your participation in this research study?

Where to get help:

If you think you have suffered a research-related injury, you should promptly notify the Principal Investigator listed in the Contact Information at the beginning of this form. Mayo Clinic will offer care for research-related injuries, including first aid, emergency treatment and follow-up care as needed.

Who will pay for the treatment of research related injuries:

Care for such research-related injuries will be billed in the ordinary manner, to you or your insurance. Treatment costs for research-related injuries not covered by your insurance will be paid by Mayo Clinic.

What are the possible benefits from being in this research study?

Others with congenital disorders of glycosylation may benefit in the future from what we learn in this research study.

What alternative do you have if you choose not to participate in this research study?

This study is only being done to gather information. You may choose not to take part in this study.

What tests or procedures will you need to pay for if you take part in this research study?

You won't need to pay for tests and procedures which are done just for this research study.

These tests and procedures are:

- *Blood draw*
- *Urine collection*
- *Stool collection*
- *Research only visit (you will be responsible for charges incurred if this occurs during a scheduled clinical visit)*
- *Testing of all samples*

However, you and/or your insurance will need to pay for all other tests and procedures that you would have as part of your clinical care, including co-payments and deductibles.

If the results of tests or procedures performed for research may be useful for your health care, you may be notified. If you decide to follow up, any further medical testing will be considered part of your clinical care, and will not be paid for by the research study. Costs will be billed to you or your insurance.

If you have billing or insurance questions call Patient Account Services at the telephone number provided in the Contact Information section of this form.

Will you be paid for taking part in this research study?

You will not be paid for taking part in this study. However, travel reimbursement up to $700 per visit will be provided for travel to and from the Mayo Clinic for any study visit that you attend. This can cover hotel stays, if required, and mileage. Itemized receipts are required. Your reimbursement for travel is calculated based on the round trip number of miles you travel from your home address to Mayo Clinic and back as determined by a web-based mileage calculator (e.g., MapQuest). This distance (miles) will be documented in your study file. You will receive reimbursement per mile at the current IRS mileage rate.

Will your information or samples be used for future research?

Unless you give your permission below, your information or samples collected for this study will not be used or shared for future research, even if the identifiable information such as your name, Mayo Clinic number or date of birth is removed.

We would like to keep your information and samples for future research. You can still take part in this current study even if you don't want your information or samples used for future research.

Approval Date: October 20, 2020

Researchers at Mayo Clinic who aren't involved with this study may ask to use your information and/or samples for future research. Researchers at other institutions may also ask for a part of your information and/or samples for future studies. Unless you indicate otherwise, the future research may be on any topic. No direct benefits to you are expected from the future research. Your information and/or samples will only be shared consistent with your consent, and with all applicable laws and regulations.

If you approve release of your information and/or samples by checking 'yes' below, Mayo may send the information and/or samples to researchers who request them, but Mayo will not send your name, address, phone number, social security number, or any other identifying information with the information and/or samples. Your information and/or samples may be sent with a code, and only the researchers for this study at Mayo Clinic would be able to link the code to you.

Some future studies may examine your DNA, the genetic information you inherited from your parents (genetic testing). If there are findings which may be useful for your health care, the researchers may contact Mayo Clinic, so Mayo Clinic can give you the option of learning the results. You would be given general information on the potential risks, benefits, and costs of choosing to learn about the findings.

To support future research, de-identified genetic information may be placed in databases accessible by the internet. Some of the information may be available to anyone using the internet, and some will be released only to approved researchers. Combined study information (including genomic summary results) may be published, but the information will not identify you.

Even though information traditionally used to identify you will not be shared, people may develop ways in the future to allow someone to link your genetic information back to you. For this reason, confidentiality cannot be guaranteed. It is also possible that reidentified information could be used in discriminating ways, and there could be additional unknown risks. We will make every effort to protect your confidentiality.

Privacy protections given by the Certificate of Confidentiality for this study do not apply to combined study results, however they do apply to your individual information. (See separate section for information about the Certificate of Confidentiality.)

Please read the following statements and mark your choices:

1. I permit my information and samples to be stored and used in future research of_____ congenital disorders of glycosylation at Mayo Clinic:

☐ Yes ☐ No Please initial here: _____Date: _____

2. I permit my information and samples to be stored and used in future research at Mayo Clinic to learn about, prevent, or treat any other health problems:

☐ Yes ☐ No Please initial here: _____Date: _____

3. I permit Mayo Clinic to give my information and samples to researchers at other institutions:

☐ Yes ☐ No Please initial here: _____Date: _____

You may withdraw your consent for future use of your information and/or samples at any time, by writing to the Principal Investigator at the address provided in the "Contact Information" section of this consent form.

Your information and/or samples would be removed from any repository where they are stored, if possible. Information and/or samples already distributed for research use will not be retrieved.

How will your privacy and the confidentiality of your records be protected?

Mayo Clinic is committed to protecting the confidentiality of information obtained about you in connection with this research study. Data collected will be stored in a research database only accessible by research staff. Any paper records are housed on a secure floor in locked cabinets only accessible by research staff.

During this research, information about your health will be collected. Under Federal law called the Privacy Rule, health information is private. However, there are exceptions to this rule, and you should know who may be able to see, use and share your health information for research and why they may need to do so. Information about you and your health cannot be used in this research study without your written permission. If you sign this form, it will provide that permission (or "authorization") to Mayo Clinic.

Health information may be collected about you from:

- Past, present and future medical records.
- Research procedures, including research office visits, tests, interviews and questionnaires.

Why will this information be used and/or given to others?

- To do the research.
- To report the results.
- To see if the research was done correctly.

If the results of this study are made public, information that identifies you will not be used.

Who may use or share your health information?

- Mayo Clinic research staff involved in this study.

With whom may your health information be shared?

- The Mayo Clinic Institutional Review Board that oversees the research.
- Researchers involved in this study at other institutions.
- Federal and State agencies (such as the Food and Drug Administration, the Department of Health and Human Services, the National Institutes of Health and other United States agencies) or government agencies in other countries that oversee or review research.
- The sponsor(s) of this study and the people or groups it hires to help perform this research.
- A group that oversees the data (study information) and safety of this research.

In addition, individuals involved in study oversight and not employed by Mayo Clinic may be allowed to review your health information included in past, present, and future medical and/or research records. This review may be done on-site at Mayo Clinic or remotely (from an off-site location). These records contain information that directly identifies you. However, the individuals will not be allowed to record, print, or copy (using paper, digital, photographic or other methods), or remove your identifying information from Mayo Clinic.

Is your health information protected after it has been shared with others?

Mayo Clinic asks anyone who receives your health information from us to protect your privacy; however, once your information is shared outside Mayo Clinic, we cannot promise that it will remain private and it may no longer be protected by the Privacy Rule.

Your Rights and Permissions

Participation in this study is completely voluntary. You have the right not to participate at all. Even if you decide to be part of the study now, you may change your mind and stop at any time. You do not have to sign this form, but if you do not, you cannot take part in this research study.

Deciding not to participate or choosing to leave the study will not result in any penalty. Saying 'no' will not harm your relationship with your own doctors or with Mayo Clinic.

Approval Date: October 20, 2020

If you cancel your permission for Mayo Clinic to use or share your health information, your participation in this study will end and no more information about you will be collected; however, information already collected about you in the study may continue to be used.

You can cancel your permission for Mayo Clinic to use or share your health information at any time by sending a letter to the address below:

Mayo Clinic
Office for Human Research Protection
ATTN: Notice of Revocation of Authorization
201 Building 4-60
200 1st Street SW
Rochester, MN 55905

Alternatively, you may cancel your permission by emailing the Mayo Clinic Research Subject Advocate at: researchsubjectadvocate@mayo.edu.

Please be sure to include in your letter or email:

- The name of the Principal Investigator,
- The study IRB number and /or study name, and
- Your contact information.

Your permission for Mayo Clinic to use and share your health information lasts forever, unless you cancel it.

MAYO CLINIC

Name and Clinic Number

Approval Date: October 20, 2020

Enrollment and Permission Signatures

Your signature documents your permission to take part in this research.

	/ /	: AM/PM
Printed Name	Date	Time

Signature

Signature of Parent(s)/Guardian for Child:

I give permission for my child to take part in this research study and agree to allow his/her health information to be used and shared as described above.

	/ /	: AM/PM
Printed Name of Parent or Guardian	Date	Time

Signature of Parent or Guardian

Signature of Legally Authorized Representative for Adult Participant

- I give permission for the participant to take part in this research study and agree to allow his/her health information to be used and shared as described above.

				AM/PM
Signature	Printed Name	Relationship to Participant	Date	Time

MAYO CLINIC

Name and Clinic Number

Approval Date: October 20, 2020

Person Obtaining Consent

- I have explained the research study to the participant.
- I have answered all questions about this research study to the best of my ability.

	/ /	: AM/PM
Printed Name	Date	Time

Signature