

Informed Consent to Participate in Research

BMT CTN 2402 Hematopoietic Cell Transplant and Gene Therapy for Non-Malignant Blood Disorders Biobank Resource (The 'HOPE Resource')

Your Name: _____

Principal Investigator: _____

[Insert local PI information]

Sponsor: This study is sponsored by the National Institutes of Health, through the
Blood and Marrow Transplant Clinical Trials Network (BMT CTN)

Your study doctor or nurse will review this **Consent Form** with you, including:

- ✓ The purpose of the research
- ✓ Study tests
- ✓ Possible risks and benefits
- ✓ Your rights if you join the study
- ✓ Any questions you may have

1. Study Overview

We invite you or your child (“you”) to take part in this nontreatment research study. The purpose of this study is to support future research including, but not limited to, non-cancer blood disorders by collecting biological (biospecimen) samples.

You are being asked to join because:

- You have one of the following blood disorders:
 - Aplastic Anemia
 - Hemoglobinopathy: such as Sickle Cell Anemia or Thalassemia
 - Other marrow failure disease
- You are receiving one of the following therapies:
 - Hematopoietic stem cell transplant
 - Gene therapy

If you join, you will:

- Be in the study for up to 5 years
- Provide some or all of the following during regular clinic or hospital visits (Table 1) to be sent to a lab and stored for future research:
 - Blood (required)
 - Stool (optional)
 - Skin biopsy (optional)
 - Nail clippings (optional)
 - Hair collection (optional)
 - Bone marrow (optional)

Some possible risks and benefits of joining the study include:

- **Possible Risks:** Discomfort from where blood, hair, nails, or skin are collected.
- **Possible Benefits:** There is no direct benefit to you to join the study, but the knowledge gained from this study may help others.

Choosing **not** to join the study will not negatively impact your treatment.

Key points:

- Being in this research study is your choice.
- If you join the study, you can leave at any time. If you decide to leave the study, it will not affect your care at [name of facility or institution].

- Ask the study staff about anything you do not understand, or if you would like more information. You can ask questions now or at any time.
- Take time to talk about the study with your doctor, study staff, and your family and friends. It is **your** choice to be in the study. If you decide to join, please sign the end of this Consent Form. You will get a copy to keep.

2. Study Purpose

We are doing this study to learn more about how people with serious blood diseases like sickle cell disease, thalassemia, aplastic anemia, or other types of bone marrow failure respond to therapy. We want to understand why some people need special treatments like gene therapy or hematopoietic stem cell transplant, and why some people have problems during and after these treatments while others do not. We also hope to find better ways to treat these diseases and make treatments safer and more helpful for everyone.

3. Study Treatment and Tests

You will be in the study for up to 5 years. Some exams and tests you will be asked to do are part of your regular care, and you will do these even if you do not join the study. Being in this study may require more tests or procedures than your regular care. If the study continues beyond 5 years, you will be asked again to provide reconsent to continue participation.

Your doctor will talk about all study collections with you and answer any questions you may have. All the collections are in Table 1 below. If there are any tests that you do not want or are worried about, please let your doctor know before joining this study.

The following describes the **research procedures** that will be performed if you decide to join this study:

Before Transplant or Gene Therapy:

- Collection of your demographics, eligibility, disease information, and samples

Study Tests and Follow-Up (if you meet the requirements to join the study)

- Collection of the following information after your transplant or gene therapy:
 - Recovery of Blood Counts (when the new, healthy cells (from your donor) start to grow in your body)
 - Infections
 - Medications
 - Reactions
 - Immunizations

- The following samples will be collected, and you are encouraged to discuss each request with your provider prior to collection:
 - Blood
 - Stool
 - In some transplants, the new donor cells do not grow like they should (graft failure) or the new cells from your donor may attack your body (graft versus host disease, GVHD). By collecting stool samples, we hope to learn more about how gut bacteria might affect whether a transplant works or not.
 - Germline (Skin biopsy, nail clippings, hair collection)
 - Germline means cells with genes that you were born with. These samples are helpful for future research aimed at finding new risks for cancers or other blood problems that can occur after treatment, and determining if problems can come from your own body, from the donor or from cells that are used for gene therapy.
 - Bone marrow
 - Your bone marrow is the factory where blood cells are made and where important problems can start after transplant or gene therapy.

Seeing Research Results

You will not have any direct health benefits from providing your specimens for future research. It will probably take a long time for the research done to be used to produce health-related information that we will know how to interpret accurately. For this reason, we will not be able to give you individual results from studies that may be conducted using the specimens. Knowledge from future research studies is likely to yield information that is more widely or generally applicable and not specific to an individual.

Timeline and Participants

Approximately 375 hematopoietic stem cell and gene therapy recipients will be in this study. Study participants will be followed for up to 5 years through March 30, 2031. It is possible the study may be able to continue past March 30, 2031. If that occurs, we will ask you to consent to continue participating past March 30, 2031.

Table 1: Collections During the Study

Sample	Pre-Conditioning Therapy	Infusion Day	Time After Infusion Day								Event Driven ²
			7 Days (+/- 3 days)	14 Days (+/- 3 days)	28 Days (+/- 7 days)	60 Days (+/- 7 days)	100 Days (+/- 7 days)	180 Days (+/- 28 days)	365 Days (+/- 28 days)	Year 2, 3, 4, 5 (+/- 60 days)	
Peripheral Blood	X	X	X	X	X	X	X	X	X	X	X
Stool	X		X	X	X	X	X	X			
Skin Biopsy (Germline)	X										
Hair Plucks (Germline)	X										
Nail Clippings (Germline)	X										
Bone Marrow ¹	BM will be collected at any time any clinical indication										

¹If bone marrow is collected as part of your standard medical care

²Additional samples and data will be collected at the time of certain changes in your health or treatment. This includes, but is not limited to, graft failure, graft-versus-host disease (GVHD), cancer, or other cell infusions. When possible, collection will occur at a scheduled study visit.

RESTRICTED

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4. Risks and Benefits

Possible Benefits

Taking part in this study will not directly benefit your health. Even though the study does not benefit you, the information from this study could help future patients with non-malignant diseases (non-cancerous blood disorder). The information from this study could help doctors learn more about medications used to treat your disease.

Blood Draw Risks

There is a small risk of an infection or fainting from the blood draw. Blood will be taken from a central line if you have one, but if no central line is available, blood will be taken from a venous puncture (needle stick) with all attempts to collect the research sample at the same time as your blood draw for clinical purposes. For children as small as 10 kg, repeated draws at this volume could exceed institutional 'minimal risk' limits over 8 weeks. Explicit limits will be enforced: no more than 35 mL per draw or the maximum blood volume allowable based on age and weight by each center's institutional guidelines (whichever amount is smallest). Research blood will be co-drawn with clinical labs whenever possible to minimize additional venipunctures.

Bone Marrow Collection Risks

As part of your treatment, you may be asked to have a bone marrow biopsy. This is a procedure where a doctor uses a special needle to take a small sample of the soft tissue inside your bone, usually from your hip. If this procedure is being done as part of your routine care, we will ask for a bone marrow sample. This helps researchers learn more about your blood and how it is made.

Like any medical procedure, there are some risks. These may include:

- **Pain or Soreness:** You may feel pain or soreness where the needle went in. This usually goes away in a few days.
- **Bruising or Bleeding:** You might get a bruise or bleed a little at the spot where the sample was taken.
- **Infection:** There is a small chance of getting an infection where the needle went in.
- **Feeling Dizzy or Faint:** Some people feel lightheaded or faint after the procedure.

However, the risks are not increased by taking a small amount of extra cells for research. Doctors and nurses will take steps to keep you safe and make sure you are as comfortable as possible during and after the biopsy.

Skin Biopsy Risks

If you agree to have a skin biopsy, the doctor will remove a very small piece of skin, usually with a special tool, to help researchers learn more about your health.

Most people do well after a skin biopsy, but there are some risks. These may include:

- **Pain or Soreness:** The area may hurt or feel sore for a few days.
- **Bruising or Bleeding:** You might get a small bruise or bleed a little where the skin was

removed.

- Infection: There is a small chance of getting an infection at the biopsy site.
- Scarring: A small scar may form where the skin was taken.

Additional Sample Collection Risks

There are no major risks to collecting your stool samples. There is a small risk of infection from collection of nails, hair and skin.

Medical Information Confidentiality Risk

There is a possible risk of the loss of confidentiality about your medical information. We will do our best to make sure that your personal information is kept private. The chance that this information will be given to someone else is very small.

Unforeseen Risks

Other new risks might appear at any time during the study. These risks might be different from what is listed in this Consent Form. The study team will do everything they can to keep you safe and lower potential negative side effects from collections. If new risks are discovered, they will be shared with you as soon as possible.

For more information about risks and side effects, please speak with your study doctor.

5. Your Rights to Withdraw, Ask Questions, and Seek Other Treatment

Being in this study is your choice. You can choose **not** to be in this study or leave this study at any time. If you choose not to join the study or if you decide to leave this study, it will not affect your regular medical care in any way. If you are considering leaving the study at any time, talk to your study doctor about your health and safety.

You have the right to ask questions about the study at any time. If you have questions about the study, please contact:

[Insert contact details for Principal Investigator or study team]

If you want to talk with someone not directly involved in the study, or have any complaints or questions about your rights as a research participant, you may contact:

NMDP Institutional Review Board (IRB) Administrator at: 800-526-7809

You must tell [insert name of Principal Investigator] if you decide to leave the study.

If you choose not to join, other options are available. Your study doctor will talk with you about your options. If you decide not to join this study, your medical care will not be affected in any way.

6. Privacy, Confidentiality, and Use of Information

Your privacy is very important to us. The study doctors, study sponsor, and other groups with access to your study-related medical information will do everything they can to protect your privacy. The

study doctors can protect your records if there is a court case. However, some of your medical information may be shared if required by law. If this happens, the study doctors will do their best to make sure that any information that goes out to others will **not** identify you.

Your confidentiality is one of our main concerns. We will do our best to make sure that the personal information in your medical record is kept confidential (private). However, we cannot promise total privacy.

To make sure the study is running ethically, some government agencies or other groups may need to access part of your medical records. For this study, those groups include:

- Blood and Marrow Transplant Clinical Trials Network Data and Coordinating Center (BMT CTN DCC), including the Center for International Blood and Marrow Transplant Research (CIBMTR), NMDP and The Emmes Company, who are coordinating the studies of the BMT CTN
- The Food and Drug Administration (FDA) and National Institutes of Health (NIH), which include the National Heart, Lung and Blood Institute (NHLBI) and the National Cancer Institute (NCI)
- Office of Human Research Protections
- Data and Safety Monitoring Board (DSMB), not part of [Institution]
- NMDP Internal Review Board (IRB) responsible for reviewing this study

If information from this study is published or presented at scientific meetings, your name and other personal information will not be used.

Study information may also be used for research in the future. These projects could be related to your disease or similar diseases or development of the study drug.

We might use information from this study to get approval from the government, like the Food and Drug Administration (FDA).

Private information and samples taken during the study will be used for future research. However, the information and samples will not be attached to you or your name in any way. The collected sample(s) will be sent to the BMT CTN Biorepository for storage. The sample(s) will be given a unique bar code that cannot be linked to you by the researchers testing your samples. Results of the research done will not be given to you.

A description of this research study will be available on <http://www.ClinicalTrials.gov/>. 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.

Data about your health, including follow-up after two years will be obtained by the BMT CTN from the CIBMTR, which captures information on all U.S. transplants.

7. Sample Information

- By consenting to this study, the research team will collect blood samples before and after hematopoietic stem cell transplant or gene therapy. At the end of this document, you will

have the opportunity to consent to additional sample types (skin, hair, nail clippings, bone marrow, stool) for this research study. Additional information about your samples is as follows:

- Your samples will not be attached to you or your name in any way. Results of the research, including any genetic studies done with this sample will not be given to you.
- Your samples will be stored in a repository designated by the BMT CTN and may be used for approved research studies by investigators in the broader scientific community.
- If available, after you have received all of your stem cell product, the left-over bag may be washed with a sterile solution and any residual cells collected and stored for future research. Normally, these “left over” cells are discarded with the infusion bag and therefore this procedure will not take away any cells you receive during the transplant. This sample will then be de-identified and used to support studies related to transplant outcomes, cell therapy, and other related research. The sample will be stored in the same secure repository as other biospecimens collected in this study.
- In addition to understanding the types and functions of cells during therapy, your blood and cells have genetic information, called DNA, which contains inherited information, like a blueprint, about the structure of your human body traits such as hair and eye color, but also how your body interacts with environmental exposures such as infections and toxins. DNA also contains changes or mutations that are acquired over a person’s lifetime and may impact how the gene functions. DNA from your stored samples may be studied by scientists to learn if some genes or gene changes predict who will have successful outcomes versus serious complications of transplantation or gene therapy.
- Genome-wide association studies (GWAS) are a way for scientists to identify genes involved in human disease. This method searches the genome across a large number of people for small genetic changes that are more common in people with a particular disease than in people without the disease. Each study can look at hundreds of thousands of genetic changes at the same time. Researchers use data from this type of study to find genes that may raise a person’s risk of developing a certain disease. You and/or your donor samples may be included in GWAS research, but all genetic information identified will not be linked to your identity nor will any results be given to you through the course of the study.
- If your coded samples are used in such a study, the researcher is required to add your test results and sample information into a shared, public research database. This public database is called the NIH Genotype and Phenotype Database and it is managed by the National Center for Biotechnology Information (NCBI). The NCBI will never have any information that would identify you, or link you to your information or research samples, although the results of genetic studies could theoretically include identifying information about you.
- A federal law called the Genetic Information Nondiscrimination Act makes it illegal for health insurance companies, group health plans, and employers of 15 or more persons to discriminate against you based on your genetic information. Health insurance companies and group health plans may not request your genetic information that we get from this research. This means that they must not use your genetic information when making decisions regarding your health insurance. This federal law will not protect you against genetic discrimination by companies that sell life insurance, disability insurance, or long-term care insurance.

- Samples will be stored until they are used up or until it is determined that they are not likely to be used in future research, at which time they will be destroyed.
- If you change your mind and no longer wish to have us store or share your data and biospecimens, you should contact [insert contact info]. We will do our best to honor your request and to retrieve any data and biospecimens and destroy them if this is also your request. However, there may be times we cannot. For example, if we do not have a way to identify your data and biospecimens we will not be able to retrieve them. In addition, if the data and biospecimens have already been used for new research, the information from that research may still be used.

8. Leaving the Study

You can choose to leave the study at any time without any negative impact on your treatment.

You may also be told to leave the study for reasons such as:

- You do not meet the study requirements.
- The study is stopped for any reason.

The study sponsor (National Institutes of Health), or the IRB may also stop the administration of this study at any time for any reason.

If this study is stopped for any reason, you will be told, and your doctor will make any necessary arrangements for continuation of your care.

9. Cost and Reimbursement

You will not be paid for joining this study. You will not be paid or reimbursed for any extra expenses (such as travel or meals) from your participation in this study.

Most of the visits for this study are standard medical care for patients undergoing transplant or gene therapy and will be billed to your health insurance company. You and/or your health insurance company will need to pay for some or all the costs of standard medical treatment in this study.

Some health insurance plans will not pay for costs of care when you take part in a research study. Check with your health plan or insurance company to find out if they will pay.

You or your health insurance company will not be charged for extra tests or research costs for this study.

For questions about your costs, financial responsibilities, and/or health insurance coverage for this study, please contact [Center/ Financial Counselor] at: [Number].

Physical Injury as a Result of Participation

It's important that you tell your doctor, [investigator's name(s)], or study staff if you feel that you have been injured because you provided blood samples, hair plucks, or nail clippings for research. You can tell the doctor in person or call him/her at [telephone number].

You will get all available medical treatment if you are injured as a result of providing blood samples for research.

You or your health plan will be charged for this treatment for injury. The study will not pay for medical treatment.

In case of injury resulting from providing the blood sample for this study, you do not lose any of your legal rights to seek payment by signing this form.

Statement of Consent for the Study

TITLE: BMT CTN 2402: Hematopoietic Cell Transplant and Gene Therapy for Non-Malignant Blood Disorders Biobank Resource

- I have read and understood this Consent Form. The purpose and description of the research study has been explained to me.
- I have had the chance to ask questions and understand the answers I have been given. I understand that I may ask questions at any time during the study.
- I freely agree to be a participant in the study.
- I have had the chance to discuss my participation in this research study with a family member or friend if I choose.
- I understand that...
 - I may not directly benefit from taking part in the study.
 - My name and personal information will not be identified even if information gained during the study is published.
 - I can leave this study at any time and doing so will not affect my current care or prevent me from receiving future treatment.
 - I will be given a copy of this signed consent form.
 - I do not give up any legal rights by signing this form.
 - Information about me and my medical course will be collected for research. This research data will have all identifying information removed so researchers will not know who I am in this study.
 - Blood samples will be drawn at regular study intervals, identifying information removed from the sample, and stored for future research use. All attempts will be made to collect the blood during blood draws done for your usual clinical needs and through an available central line if present.

I understand that drawing additional sample types beyond blood is optional for this study. All identifying information will be removed from the optional samples below and stored for future research use. The following optional sample types, reason for collection, and explanation of how the sample is collected are given below with my decision to agree or disagree to provide the sample by checking the box associated with my preference “YES” or “NO”.

Bone Marrow:

Why: Your bone marrow contains information such as the health of blood forming cells, the early production of gene therapy transformed cells or donor immune cells and may have small genetic changes in blood forming cells that impact outcomes. This information may not be found in blood alone as our blood contains much older or mature cells and early cells can only be studied with a bone marrow sample.

How: If you are undergoing a bone marrow biopsy for clinical purposes, we request permission to take a small amount of additional sample for research purposes and share a portion of remaining tissue left over from usual clinical purposes. Since you are already undergoing the procedure, collection of additional samples from the same site would minimally extend the procedure (usually a few seconds to maximum 1 minute).

- ☐ **YES** I agree to give a bone marrow sample for research
- ☐ **NO** I do not agree to give a bone marrow sample for research

Skin Biopsy/Hair Plucks/Nail Clippings:

Why: Research in rare disorders involves the study of genes and changes in genes that can be different between different people and even between different cell types in our body. Although all the cells in our body usually contain the same genetic information (DNA) when we are born, some cells may develop changes in the genes as we age. For patients with blood disorders, genetic changes can happen in blood cells only and can be difficult for researchers to know if the changes were present at birth or developed at some time after birth. By collecting other cell types, such as skin, hair, and/or nail clippings, researchers can look at the DNA of these cells and compare it to the DNA of blood cells. If the DNA is the same across all cell types, it is likely that any genetic mutations were present at birth. But if blood cells contain a mutation that is not present in other cell types, it is likely that the genetic change happened after you were born and only in blood cells and this would be valuable information for researchers to know to better understand your disease and treatment outcomes.

Skin Biopsy

How: Skin cells will be collected once pre-transplant or pre-gene therapy by performing a small skin biopsy once at any time to be used for research purposes. The skin biopsy is done at the site of another procedure (e.g., central line placement or bone marrow biopsy) whenever possible but can also be done as a separate procedure at any site (e.g. upper inner arm). After washing the skin to sterilize the area, an injection of a local anesthetic is used to numb the area. Then a small cookie-cutter like device is used to remove a small, pencil eraser sized piece of skin. A steristrip or small stitch is used on the surface of the skin to close the small hole and a bandage is applied. This process takes about 10 minutes.

- ☐ **YES** I agree to give a skin sample for research
- ☐ **NO** I do not agree to give a skin sample for research

Hair Plucks (10-15 total):

How: Hairs will be plucked once pre-transplant or pre-gene therapy to include the root by yourself or a research coordinator if you prefer. This process takes about 5 minutes.

- ☐ **YES** I agree to give hair samples for research
- ☐ **NO** I do not agree to give hair samples for research

Nail Clipping:

How: Samples of your nails will be clipped once pre-transplant or pre-gene therapy with standard nail clippers by yourself or a research coordinator if you prefer. This process takes about 5 minutes.

- ☐ **YES** I agree to give nail samples for research
- ☐ **NO** I do not agree to give nail samples for research

Stool:

Why: Your stool (poop) contains material which can impact your overall health and provide information on your health status. For example, the types and amounts of bacteria in your stool may impact the amount of inflammation in your intestines and how you absorb food and medications. Understanding the impact of bacterial and chemical changes detected in the stool may give researchers information on how these differences could impact the outcomes of bone marrow transplant or gene therapy.

How: A small amount of stool that you collect at the hospital, clinic, or home would be used for research purposes. If collected at home, you will be given collection materials and instructions.

- ☐ **YES** I agree to give a stool sample for research
- ☐ **NO** I do not agree to give a stool sample for research

Sign your name on the line below if you agree to be in this study. If you are a legally authorized representative (LAR), sign on behalf of the participant.

Participant Name (or Parent/Guardian/LAR)

Date (MM/DD/YYYY)

Participant Signature (or Parent/Guardian/LAR)

Physician Certification

I certify that I have provided a verbal explanation of the details of the research study, including the procedures and risks. I believe the participant has understood the information provided.

Counseling Physician Name

Date (MM/DD/YYYY)

Counseling Physician Signature

Interpreter Certification (if needed)

I certify that I have provided an accurate interpretation of this consent form. I believe the participant understood the information provided.

Interpreter Name

Date (MM/DD/YYYY)

Interpreter Signature