Clinical Investigation Consent Form

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You are being asked to join a research study, which will take place at The Rockefeller University Hospital. This form tells about the research. You should ask questions of the person who is explaining this form to you. After you feel that you understand the research, if you want to be part of the study, you will be asked to sign the form. You can always ask more questions and can later change your mind about staying in the study.

If you join the research study, you will take part indefinitely unless you request that you be withdrawn. The research study as a whole will last indefinitely.

About 4000 people will take part in the research study.

Title of the research study: **Entrance into the International Fanconi Anemia Registry (IFAR)**

I. <u>What this research study is about, and the reason for doing this research.</u>

The reason for doing this research is to study the nature, diagnosis and treatment of individuals affected with the genetic disease Fanconi anemia, an inherited disorder that leads to bone marrow failure (aplastic anemia). In most cases it is a recessive disorder: if both parents carry a defect (mutation) in the same FA gene, each of their children has a 25% chance of inheriting the defective gene from both parents. When this happens, the child will have FA. Patients may have a variety of birth defects, and may eventually develop acute myelogenous leukemia (AML), head and neck, gynecological, and/or gastrointestinal squamous cell carcinomas. The researchers doing the study will collect information about the medical history, genetics, clinical course, blood test results, treatment, complications and social issues of Fanconi anemia. Information about relatives of Fanconi anemia patients will also be collected. A purpose of this project is to develop a detailed listing, or "registry" of people who may have Fanconi anemia, and their close family members.

The International Fanconi Anemia Registry (IFAR) was established at the Rockefeller University Hospital in May, 1982, to learn if a large number of people with this rare disease, might also show signs of its many various features. This registry has proven to be a very valuable repository for clinical, hematologic and genetic information on FA patients and their families as well as a source of cells for studies.

We are asking you (your child) to take part in this research study because you (your child) may be affected with Fanconi anemia or may have symptoms found in those affected with Fanconi anemia or you may be a relative of person affected with Fanconi anemia (or have Fanconi anemia-like symptoms).

II. What is going to happen in this research study?

Consent Process: Informed consent is a process to help you understand the purpose of the research study, what will happen in the study, possible risks and benefits, and your right to withdraw from the study at any time. All of this information will be explained to you in detail. You should ask any questions you have until you feel that you understand what is asked of you to participate. You may then want to enroll, or you may decide not to join the study. The decision to participate is entirely up to you. Even after the study has started, you may at any time ask more questions, or decide to withdraw from the study.

If you (your child) agree(s) to join the study, you will be asked questions about your medical history, and the history of your family. The information about you (your child) will be entered on a questionnaire form by your (your child's) doctor. The form along with your (your child's) medical records will be submitted to the International Fanconi Anemia Registry (IFAR). In addition to the information collected when you first join the study, your (your child's) doctor will be contacted regularly for updates about your (your child's) medical condition. You may be asked to sign a release form giving us permission to request some of your (your child's) medical records. In addition, the study coordinator at Rockefeller University may contact you to ask for medical updates about you (your child). Lastly, a physical exam may be performed on you/your child to look for any physical features typically associated with Fanconi anemia.

There is also a way to update the study coordinator at your convenience through visiting our website http://lab.rockefeller.edu/smogorzewska/families/ and completing the medical update form under Resources/Links. We will also maintain a Facebook page (Fanconi Anemia Registry or Agata Smogorzewska) to update you on the progress of the studies and to keep in touch with families affected by FA.

The information will be entered into a secure password protected, web-based database, which will hold similar information about many other Fanconi anemia patients and their relatives. You (your child) will be given a code such that your (your child's) identity will never be revealed publicly.

In this part, we explain the meaning of words that we are going to use to describe this study:

> "**Substances drawn from your body**" refer to **liquids** such as blood, urine, or saliva. It can also mean tissues such as skin, cheek cells and **DNA**. **Cell**s make up all parts of your body. DNA is inside all the cells of your body and carries your genetic or inherited information. When we draw blood, take tissue, or take other substances from your body, we are taking a "**sample**."

"**Cell line**" means a group of cells that can live and grow outside of the body. They can also be frozen and can be used for future research.

As part of the study of Fanconi anemia, you are being asked to give a blood sample (less than one ounce), skin sample, saliva or a sample of cheek cells collected with small brushes rubbed on the inside of your mouth or from a mouthwash sample. The sample will be used to obtain cells or chemical components such as DNA to further research. The investigators may want to use your biological sample to make a cell line.

- If testing was performed in our licensed laboratory before the start of 2008, the researcher may tell your doctor the test results that may affect your health. You will need to sign an "Authorization to Release Clinical Results" form if you want the genetic test results for you (your child) released to the physician that provided the sample or to some other health care provider.

- All testing as of 2008 has been performed in our research laboratory. As such, these results need to be confirmed in a clinical "CLIA" laboratory before results could be shared with you or your doctor. We can help your doctor order the necessary testing for confirmation, but it is likely that there will be a cost associated and testing will require a new DNA sample. This confirmational step is optional and you do not have to pursue it, but it is important to realize that without confirming the results, we cannot share this information with you or your doctor.

The samples that you have donated for this research study will be stored in our repository where it can be used by researchers at Rockefeller and elsewhere for future research projects to study the genetic and biochemical changes seen in Fanconi anemia and clinically related diseases.

In this study, you will not receive care for any related medical problems you may have.
In this study, you will not receive care for any other medical problems you may have.

If you are seen at the Rockefeller University Hospital, your medical information and test results will be written in your Hospital chart. The researchers will also keep separate records with information about you and your study tests. If you are not seen at the Rockefeller University Hospital but participate in this study, there will be no Rockefeller University Hospital chart with your medical history. The researchers will keep the records with information about you and your study tests. Sometimes we will need to look at your earlier medical records. We will ask you to sign a form that will let health care providers share your records with us. This could be your doctor, a clinic or another hospital where you have been treated before. All of this information will also be entered into a secure password protected database.

III. What are the risks of taking part in this research study?

There may be some risks and discomforts in taking part in this study. We know that these risks and discomforts may happen during this study:

The possible risks and discomforts associated with a blood draw are slight pain, and possibly a bruise. Occasionally there may be a temporary faintness or an infection at the needle entry site. The risks of a skin biopsy are slight pain and slight bleeding. The skin biopsy will produce a small, permanent scar in the skin; however biopsy sites will be chosen to minimize their appearance. These skin biopsy sites heal in a variety of ways. The final appearance will depend in part on the area of the body biopsied, the reason for the biopsy, and the underlying skin appearance before the biopsy. Scars may continue to change for many years after the sutures are removed. In addition, everyone heals differently, and it is possible that the scars may be red for some time, or become raised, darker or lighter than the surrounding skin. You will most likely have a permanent scar of some kind, and looking at your prior scars may give the best prediction of your long-term healing. An occasional person may develop a superficial, temporary wound infection at the site of the biopsy. The possible risks and discomforts associated with a cheek cell collection are slight irritation at the site of the brushing. The appearance of any of these complications after these procedures is rare.

While the risk is more theoretical at this time, as we do not know how genetic information will be used in the future, there is a potential risk to your privacy and confidentiality through participation in this study. When a large amount of genetic sequencing is done (called whole exome or whole genome sequencing), that information is stored in a secure web-based database. Only certain researchers will have access to this database and the information in the database is not connected to your name, date of birth, or any other information that could identify you. However, your genetic information is like a fingerprint and is unique to you, so it is possible that someone in the future could identify you based on this information. It is important to note that there is a federal law in place to prevent discrimination based on genetic information called Genetic Information Nondiscrimination Act (GINA) (see below).

There may be other risks and discomforts that we do not know about now, but we will tell you any new information discovered which might affect your decision to participate or remain in the study.

IV. What are the benefits of taking part in this research study?

There may be no direct benefit from your participation in this research program. Rather, the benefits that you (your child) might reasonably expect from this research program are indirect. The Registry is expected to improve understanding about diagnosis, genetic basis, course, and treatment of Fanconi anemia, and this could potentially benefit you (your child). In some cases, the study may be able to identify the specific gene that Fanconi anemia is due to in your family. If this happens, the results would need to be confirmed by a clinical laboratory before they could be released to you. To do this clinical testing, your doctor would likely need to take another blood, buccal (sample of cheek cells collected with small brushes rubbed on the inside of your mouth), or skin sample from you (your child). This knowledge may be a direct help to some patients by allowing their doctors to know better whether a bone marrow transplant will

be needed soon. In addition, we have a genetic counselor who is available to speak with you or your child's physician should questions be raised about FA, FA testing, or our research study. However, it is important to know that this study is not intended to provide medical care and as such we can not provide any recommendations about you or your child's medical care.

V. Who will be able to see the information learned about you in this research study?

We will keep your personal information private, and will do our best to keep this information confidential. We will listen to what you say we may do with this information, and we will follow the law. Your information will be made available only to the government agencies that oversee this research and/or its results, and certain University personnel concerned with protecting persons who take part in our research studies.

During the research study, two groups of people will know that the samples came from you. They include the scientists in the Laboratory of Genome Maintenance at Rockefeller University and doctors that work with families affected by FA in both the FA Comprehensive Care Centers and other specialized clinics. No one can have access to your identifiable information without specific permission of our regulatory board called the Institutional Review Board (IRB). In addition, these individuals cannot share your information with anyone else without your permission. Other researchers will have access to the information in the database, but will not be able to identify you, because your information will have a code instead of a name attached to it. These other researchers who study your samples will not know that they came from you and will not be able to figure out that they came from you. If the people doing the study publish the results of this research, they will not publish your name or any identifying information that may be linked to you.

You need to know that if you choose to communicate with the investigators using Facebook we cannot guarantee the confidentiality of the private messages exchanged.

Certificate of Confidentiality

We have obtained a Certificate of Confidentiality from the federal Department of Health and Human Services (DHHS). This Certificate protects your privacy so federal, state, or local courts or police will not be able to force us, through legal demands e.g., court orders, to give out information about you. However, the researchers must give information about you if asked by the Federal Food and Drug Administration or the Federal agency that is supporting this research. Also, the researchers will report to the authorities if they believe that child abuse or neglect has happened, or to prevent serious harm to you or others.

Genetic Information Nondiscrimination Act

A Federal law, called the Genetic Information Nondiscrimination Act (GINA), generally makes it illegal for health insurance companies, group health plans, and most employers to discriminate against you based on your genetic information. Additional information is available upon request.

Sharing of the results

In order to allow researchers to share test results, the National Institutes of Health (NIH) and other central repositories have developed special data (information) banks that collect the results of whole genome studies. The NIH or other data banks will store your genetic information and give it to other researchers to do more studies. We do not think that there will be further risks to your privacy and confidentiality by sharing your whole genome analysis with these databanks; however, we cannot predict how genetic information will be used in the future. The information will be sent with only your code number attached, and your name or other identifiable information will never be given to them. There are many safeguards in place to protect your information while it is stored in repositories and used for research. Research using your whole genome information is important for the study of virtually all diseases and conditions. Therefore, the databank will provide study data for researchers working on any disease, which could include conditions such as HIV/AIDS, cancer, mental illness, and others.

VI. What are the payment arrangements?

There is no cost to you for being in this research study.

If research using your samples helps develop a drug or another product that is sold to the public, the drug company, the University and the researcher may share in some of the profits. For example, a cell line from your samples could be used to make a product for sale. There are no plans to pay you any money resulting from such discoveries. However, by signing this form, you do not give up any rights you may have.

VII. What happens if you don't want to stay in this study or your participation is ended?

You can choose if you want or do not want to be part of this study. If you do not join, there is no penalty and no one will hold this against you. If you decide to join this study, you may change your mind and stop taking part in the study at any time, and this will not be held against you. Information about you up to that time may stay a part of the study.

During this study, the researchers may learn new information that might make you change your mind about whether you want to stay in the study. You will be given that information promptly.

If you decide to join the study now but later want to stop, you should let the researcher know. You can contact one of our study coordinators (Jennifer Kennedy or Erica Sanborn) or the head of the study, Agata Smogorzewska (all contact information on page 8) and tell them that you no longer want to be part of the study.

If you no longer wish to be part of the repository the researchers will anonymize the sample by removing and destroying all identifiers and links to identifiers so that it can not be associated with you, but the researchers will not destroy the sample.

VIII. Consent to the use, storage and sharing of your samples and data for separate research studies

The scientific value of your samples and the information obtained from them is redoubled when we can share them with other scientists at universities and pharmaceutical companies worldwide. The genetic information obtained from your DNA is called genotype. The information about your disease condition and the physiology of your cells is called phenotype. As part of your participation in our study, we may:

- store, use, and share for many years your blood or tissue samples and data including genotype and phenotype data, with other investigators at Rockefeller and elsewhere, possibly worldwide, and including pharmaceutical companies, sample and/or data banks/repositories for separate studies for many years. Your samples will either be stripped of information identifying them as yours or coded (we will hold the key to the code) so that they cannot be identified as having come from you. Other data related to your sample, but that does not identify you may accompany the samples; and

- put a summary of our studies (e.g. number of participants, types of data collected, etc) in a public database. There would be no information in this public database that could identify you; and

- put your coded genotype and phenotype medical data information and data information from more detailed analyses of your coded samples in a NIH controlled-access database/repository. The data information in this database/repository will be available only to qualified researchers from academic institutions and commercial organizations, both domestic and foreign who have received approval from an NIH Data Access Committee.

Any time in the future, you may withdraw your consent to use any samples that have not already been used in research or shared. If you withdraw your consent, the remaining unused samples will be destroyed, unless the samples cannot be identified as having come from you. Data generated using your samples will continue to be used.

IX. Who do you call if a medical problem results from this research study?

If you believe that this study has led to a medical problem, you should call the researcher listed below right away. The researcher will help you get appropriate, available medical care.

Name: Farid Boulad, MD
Phone No.: 212-327-7850 at RU or 212-639-6684 at MSKCC
Fax No: 212-327-8262 at RU or 212-717-3447 at MSKCC
Email: bouladf@mskcc.org

The Rockefeller University does not plan to pay for medical care that you may have as a result of taking part in this study at The Rockefeller University Hospital. However, you do not give up any rights you may have to seek compensation by signing this form.

X. Who do you contact if you have questions about the research study?

Please ask as many questions as you want about this research study and this consent form. If you agree to take part in this study and have questions later on, contact any of the following people:

Agata Smogorzewska, MD, PhD	212-327-7850	asmogorzewska@rockefeller.edu
Jennifer Kennedy, MS, CGC	212-327-8612	jkennedy@rockefeller.edu
Erica Sanborn, MS, CGC	212-327-8613	esanborn@rockefeller.edu

These individuals can also be reached at

The Rockefeller University
1230 York Avenue, Box 182
New York, NY 10065
Fax No.: 212-327-8262

If you have any concerns about your experience while taking part in this research study, you may contact The Rockefeller University Institutional Review Board (IRB) Office at (212) 327-8410, or the Office of Clinical Research at (212) 327-8408.

XI. May we have permission to contact you about future studies?

May we contact you by phone to find out if you are interested in hearing about new research studies? Contact would be made by the Principal Investigator or her staff. If you decide at any time that you no longer want to be contacted, please tell us, and we will stop calling you.

Would you like us to contact you about future research studies?

Yes _____ No _____

If you say "no" to this question, this will not affect your participation in this study.

AGREEMENT TO PARTICIPATE -- SIGNATURES REQUIRED

I have read this consent form, and my questions have been answered.

A copy of this consent form will be given to you. Please keep a copy of the form as it contains important information that you may wish to refer to during the research study and thereafter.

I hereby voluntarily consent to take part in this research study.

Name of the Study Participant (Print) _____

_____ _____

Signature of Study Participant **Date (To Be Filled in by Study Participant)**

ALTERNATE SIGNATURE BLOCK

Adult not legally capable of giving consent

Name of Legal Representative (Print) _____

_____ _____

Signature of Legal Representative **Date (To Be Filled in by Representative)**

Documentation, such as a health care proxy, showing authorization to act as a legal representative, is attached.
[Otherwise, explain why no documentation is available

ALTERNATE SIGNATURE BLOCK

Protocols Involving Children

I hereby voluntarily consent to have my child take part in this research study.

Name of Mother or Guardian (Print) _____

Signature of the Mother or legal guardian	**Date (To Be Filled in by Mother or Guardian)**

Name of Father or Guardian (Print) _____

Signature of the Father or legal guardian	**Date (To Be Filled in by Father or Guardian)**

ALTERNATE SIGNATURE BLOCK

Protocols Involving Pregnant Women

I hereby consent, as the pregnant mother of my child, to take part in this research study.

Name of Pregnant Woman (Print) _____

_____ _____

Signature of Pregnant Woman **Date (To Be Filled in by Pregnant Woman)**

I hereby consent to have the pregnant mother of my child take part in this research study.

Name of Male Parent (Print) _____

_____ _____

Signature of Male Parent of Child **Date (To Be Filled in by Male Parent)**

Signature of the Person Conducting the Informed Consent Discussion

I have explained the research protocol and this consent form to the participant and have answered the participant's questions about this research study and/or the consent process.

Name of Person (Print) _____

_____ _____

Signature of Person Discussing Consent **Date (To Be Filled in by Person Discussing Consent)**

ALTERNATE SIGNATURE BLOCK

Participant requires assistance by a translator

Translation Services Provided by (choose one, by checking one box below):

☐ **Pacific Interpreters**

_____ _____

Language **Translator Identification Number**

Witness to telephone translation:_____
(Print Name)

_____ _____

Signature of witness **Date**

☐ **Other Translator:**

_____ _____

Name of translator **Date**

Witness to oral presentation:_____
(Print Name)

_____ _____

Signature of witness **Date**
