

Repository of Tissue Samples and Data from Patients with Fanconi Anemia

What is the purpose of this study?

The purpose of this research study is to help investigators learn more about Fanconi anemia.

Who will be included?

Patients that have Fanconi anemia, a genomic instability syndrome, Seckel disease, Bloom's syndrome, Nijmegen syndrome, and/or a Fanconi anemia-like disorder or you are a parent or sibling of such a patient.

What samples/information do we need?

In this study, bone marrow and/or blood and/or skin tissue and/or oral samples will be collected from you. Data will also be collected through questionnaires.

What is involved?

Bone marrow and tumor cells will be collected from you only when routine samples are being collected for standard diagnostic or treatment related reasons (this includes procedures required as part of another research study on which you are enrolled). Peripheral blood will be obtained when undergoing clinically indicated blood draws whenever possible. Skin biopsies will only be obtained if you are already undergoing a skin biopsy which is clinically indicated for diagnostic purposes, or when the skin biopsy is paired with another clinically indicated procedure, such as a bone marrow aspirate. Buccal cells (cells from the inside of your cheek), saliva samples and oral rinse specimens will be obtained during a routine clinic visit, visit for collection of research samples only, or at a special event.

What will be done with the samples?

Your samples will be labeled with a unique patient number and stored indefinitely in a laboratory at Cincinnati Children's and will be released to researchers as needed. If you decide to allow your samples to be collected and stored, you still have the option to remove the samples at any time. If in the future you ask that your stored samples be discarded, it is important to know that the samples that have already been distributed to researchers prior to your withdrawal request cannot be destroyed.

Collection and Storage of Personal and Medical Information:

Researchers in this study will gather information about your medical history from your chart.

The information from the research study may be published; however, you will not be identified in such publication.

What are the benefits?

If you agree to take part in this research study, there will not be a direct medical benefit for you. The information learned from this research study may benefit other patients with an immune disorder in the future.

Will I get all the facts about the study?

If you are interested in participating in this study, you will meet with a study coordinator who will explain all of the details of the study. The study coordinator will review the consent form and will be sure that all questions are answered. The

consent form describes all the procedures, risks, benefits and who to contact with questions or concerns. Study procedures will not begin until the consent has been signed by the patient (if over 18 years) or the parent/guardian of the patient.

What are the risks?

A detailed list of possible risks will be provided to those patients interested in knowing more about the study.

Who should I contact for more information?

Robin Mueller

Cincinnati Children's Hospital Medical Center

3333 Burnet Ave.

Cincinnati, Ohio 45229-3039

Phone: 513-636-3218

robin.mueller@cchmc.org