Continuing the Dialogue

Participants and families will be asked to complete an annual follow-up form in order to update medical and cancer information. People with the syndromes who belong to the Clinical Center Cohort will be asked to visit the Clinical Center annually.

It will be possible for families to change from the Field Cohort to the Clinical Center Cohort. Members of both cohorts will be able to ask questions and stay in communication with the research team.

Anything of clinical significance that would change a patient's current treatment or screening for cancer will be shared with participants. The overall findings of the study will be provided to participants when the analysis is completed.

Other Details

Confidentiality. NCI is required by Federal law not to reveal any information that is collected from study participants to anyone other than to persons directly involved with the study. No information that could allow identification of any specific individuals will be released or published.

Costs. All study-related medical expenses and travel costs to the NIH Clinical Center for participants and immediate family members, or primary caregivers, are paid by NCI.

About the Research Team

Blanche P. Alter, M.D., M.P.H., is the lead investigator for this study. A cancer expert in the NCI Clinical Genetics Branch, she has been caring for and studying patients with bone marrow failure disorders for more than 25 years.

Regarded as one of the leading investigators for these diseases, both in the United States and abroad, she came to the National Institutes of Health in September 2000 with the purpose of developing this study.

Dr. Alter has teamed with a large number of associate investigators in all specialties at the NIH and other medical centers, to provide comprehensive evaluation for people with these complex, multi-system disorders.

For More Information

Phone: **1-800-518-8474** to speak with a member of the research team.

Email: **LisaLeathwood@westat.com**

Web site: **www.marrowfailure.cancer.gov**

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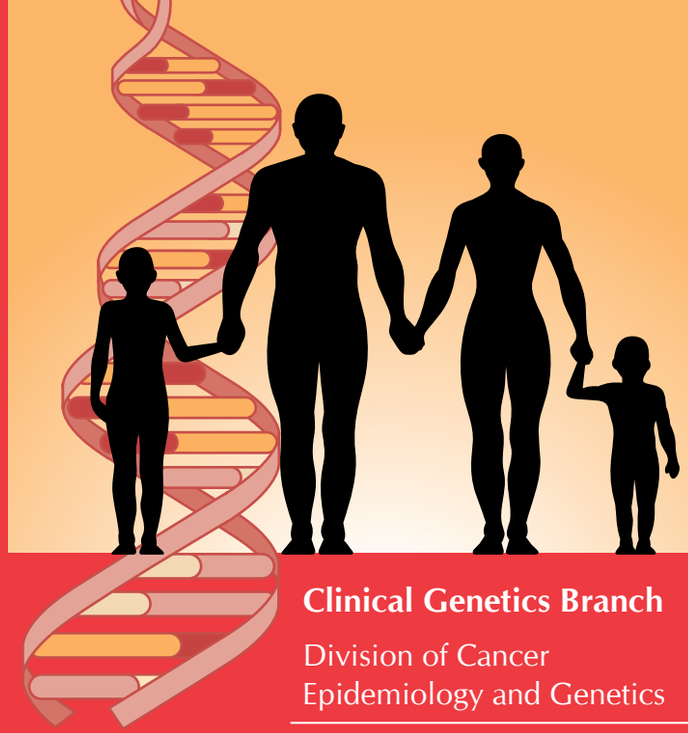

Clinical Genetics Branch

Division of Cancer Epidemiology and Genetics

Inherited Bone Marrow Failure Syndromes

Studying Families with Rare Blood Disorders and Risk of Cancer

National Cancer Institute
National Institutes of Health

Purpose

The National Cancer Institute (NCI) is sponsoring the largest North American study of its kind to focus on people with rare inherited bone marrow failure syndromes (IBMFS) and their immediate family members. This large group, or cohort, is called the NCI IBMFS Cohort. The study's purpose is to:

- Examine the underlying genetic disorders of those diagnosed with an Inherited Bone Marrow Failure Syndrome (IBMFS) and their families
- Analyze how certain factors can affect the course of these syndromes, particularly the connection between these disorders and cancer
- Follow families over a period of years

Who Is Eligible?

People with an IBMFS often have some form of aplastic anemia, in which the bone marrow fails to produce blood. The most common of these inherited syndromes include:

- Fanconi's Anemia
- Diamond-Blackfan Anemia
- Shwachman-Diamond Syndrome
- Dyskeratosis Congenita
- Severe Congenital Neutropenia
- Thrombocytopenia Absent Radii
- Amegakaryocytic Thrombocytopenia
- Pearson's Syndrome
- Bone Marrow Failure Other Than Acquired

Although these are mostly diseases of the blood and bone marrow, people with these disorders also have a high risk of cancer, such as leukemia and solid tumors.

By taking part in this long-term study, patients and their families can learn more about their condition, as well as have the opportunity for appropriate cancer screening.

Adults and children with any of these diagnoses, as well as their close relatives, are invited to join the study. People who are newly diagnosed, and surviving relatives in families where the patient may have passed away are also welcome.

Seeking More Answers

The NCI hopes to answer a number of questions about cancer in people with IBMFS, such as:

Who — Which patients (and which relatives) will develop cancer?

What — What kinds of cancer occur in different patients and in different syndromes?

Where — Where does cancer appear?

When — At what age are patients most likely to develop which kinds of cancer?

Why — Why does cancer develop?

How — How do IBMFS genes and the environment interact?

Participating in Research

Everyone who joins the NCI IBMFS Cohort will initially belong to the Field Cohort. Those who come to the NIH Clinical Center will become part of the Clinical Center Cohort, while those who do not come will remain in the Field Cohort.

A family contact will be asked to provide information about the family's overall medical history. Those with the disorder and immediate relatives will be asked to provide more detailed individual medical information.

For people in the Clinical Center Cohort, the NCI will offer:

- Complete physical examinations, blood and bone marrow studies, imaging, age-appropriate cancer screening and consultation with a team of specialists
- Genetic counseling and an opportunity to learn the results of mutation testing
- Results of clinical tests and cancer screening
- Education on possible ways to reduce cancer risk

The purpose of this study is to identify the relationship among genes, physical examination and laboratory findings, and the risk of getting cancer. Although treatment is not part of the study, options will be discussed with participants.

The NCI will also provide assistance in establishing care with appropriate physicians as needed. Study participants should remain under the care of their primary doctors while participating in the study.