

Screening Protocol for Genetic Diseases of Lymphocyte Homeostasis and Programmed Cell Death

What is the purpose of this study?

The purpose of this protocol is to find the biochemical and genetic causes of inherited immune diseases affecting lymphocytes. A second purpose is to develop new knowledge which will help research doctors find better ways to prevent, diagnosis, and treat these inherited immune diseases.

Who will be included?

You are being asked to take part in this research study because you have or have had constantly enlarged lymph nodes or spleen, along with autoimmune disease, immunodeficiency, lymphoma, or other immune problems affecting certain white blood cells called lymphocytes.

What samples/information do we need?

You will provide a blood specimen for this study. If you have any type of tissue removed during your regular clinical care, we may ask that any leftover samples be sent to Cincinnati Children's Hospital Medical Center for research testing. Types of tissues include lung, liver, teeth, or any other type of biopsy.

Your medical information will also be collected.

What is involved?

The blood specimen for this study will be drawn through a needle that will be placed in your arm or hand (venipuncture). Your personal doctor will arrange for blood drawing and shipment to the National Institutes of Health (NIH).

What will be done with the samples?

The blood will be tested by the research doctors at the NIH. The research doctors will use the blood to test for white blood cell (lymphocyte) growth and death. They will also test how your lymphocytes function (how well the cells work), and the biochemistry of the cells (chemical reactions in the cells). Researchers will also use the blood to test the genetic (DNA) structure of your cells. For some studies, the genetic material (DNA) will be extracted from your blood sample and tested to look for the genes that might cause immune diseases.

Collection and Storage of Personal and Medical Information:

In the course of this research we will obtain blood samples, some of which may be frozen away for future studies. In addition, certain information will be stored in your medical record. In the future, it is possible that your blood samples or your medical records will be used for other research purposes that are not specifically outlined in this consent form. If this is done, your specimens will be de-identified before they are shipped to the NIH.

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?

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