



BC CHILDREN'S HOSPITAL
BioBank



CURRENT RESEARCH AT BC CHILDREN'S HOSPITAL

A summary of relevant BC Children's Hospital research studies which currently require biological specimens is provided below. **If you agree to take part in the BioBank, a portion of your biological sample(s) may be utilized for some or all of these studies**, depending on the suitability and specific study requirements. In addition, remaining samples will be stored in the BioBank for future research.

If you are interested in obtaining more detail about any of the following studies, please contact the investigator, as indicated.

STUDY: Characterizing IRF4 deficiency, a novel human primary immunodeficiency

Background & Purpose: Primary immunodeficiency diseases (PIDs) are a group of genetic disorders in which parts of the human immune system are missing, dysfunctional, or poorly regulated. We have identified multiple pediatric patients diagnosed with combined immunodeficiency who have an identical genetic variant in the *IRF4* gene, which has not been previously seen in humans.

The IRF4 protein controls genes that enable the maturation of protective immune cells, which is important for the development of a strong immune system capable of protecting against pathogens like bacteria, viruses, and parasites. The patients we have identified therefore suffer various infections as a consequence of their IRF4 impairment.

The goals of this study include investigating the specific consequences of IRF4 impairment on T cell function in the pediatric patients compared to healthy children, which will be accomplished by analyzing the immune cell populations present in the blood. This study will help us better understand the role IRF4 plays in the immune system, and how that knowledge can be used to inform the care of the patients in this study, as well as potentially discovering new therapeutic targets to treat other immune-mediated diseases.

What's required? Blood samples from patients with non-inflammatory conditions who are undergoing a procedure at BCCH.

Who's eligible? Children undergoing elective surgery that are not diagnosed with an inflammatory condition.

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STUDY: Evaluation of Automated Plasma Cytokine Analysis for Cytokine Release Syndrome

Background & Purpose: Cytokines are small proteins produced by our cells. Cytokine Release Syndrome (CRS) happens when the immune system responds more aggressively than it should, and releases high amount of cytokines in blood. CRS is a common side effect in patients who have received CAR-T therapy (a treatment for acute leukemia). Because CRS symptoms can be similar to other serious conditions, such as sepsis (a blood infection) and HLH (hemophagocytic lymphohistocytosis, a rare condition with severe inflammation), it can be hard for doctors to know which of these conditions a patient has. It would be helpful for doctors to determine this because each condition should be treated differently.

In this study, cytokines will be compared from patients who have undergone CAR-T treatment, patients with bacterial sepsis, and patients with HLH. The study will help to learn about cytokine patterns in each condition, and explore if cytokine measurement would help to distinguish between them. Blood samples from study participants might also be used to assess other immune responses.

Who's eligible? Patients at BC Children's Hospital with any of the following conditions:

- B-Cell Acute Leukemia (B-ALL), scheduled to be treated with CAR-T therapy
- Suspected Bacterial Sepsis
- Suspected HLH (hemophagocytic lymphohistocytosis)

What's required? A blood sample (1 tsp) taken at the same time as usual blood tests as follows:

CAR-T patients	<ul style="list-style-type: none">• Before you receive CAR-T treatment• 1 to 2 days after your CAR-T treatment• 3 to 5 days after your CAR-T treatment <p><i>If you develop a fever (over 38°C) blood will also be collected:</i></p> <ul style="list-style-type: none">• Within 48 hours of fever onset (<i>Additional sample is only required if this does not coincide with the prior time points</i>)• 1 weeks after fever onset
Patients suspected of having sepsis or HLH	Within 48 hours of your fever assessment

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