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: Platelet-based Biomarkers in Juvenile Idiopathic Arthritis (JIA Biomarkers)

Background & Purpose: Juvenile idiopathic arthritis (JIA) is the most common cause of youth disability in Canada, affecting ~20,000 Canadian children and adolescents. Children and youth with JIA have chronic inflammation in their joints that causes pain, stiffness, swelling and bone degradation. Current treatments for JIA are effective but, because they suppress the immune system, the drugs also increase the risk of severe infections and cancer. Currently, there are no reliable ways to identify those children with JIA that are at risk of severe or progressive disease who might require more aggressive treatment, or conversely, those children with milder disease that could take a lower dose of the drug and/or take it for a shorter period of time.

In this study, doctors from BCCH will invite families to donate samples to help us compare patients with and without JIA to better understand JIA and develop new diagnostic and treatment strategies, that will allow doctors to make better informed decisions in caring for children with JIA.

What's required? Blood samples from non- JIA patient who are undergoing a procedure at BCCH

Who's eligible? Children undergoing elective surgery that are not diagnosed with juvenile idiopathic arthritis (JIA)

Principal Investigator: Dr. Kelly Brown– kbrown@bcchr.ca
Site co-ordinator: Stephanie Hughes – sthughes@bcchr.ca

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.

Principal Investigator: Dr. Stuart Turvey – sturvey@bcchr.ca

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