

Wednesday, October 16th, 2013

LSC 3 - Life Sciences Centre

2350 Health Sciences Mall

12-1pm



Dr. Jacob Rozmus

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“The clinical and translational study of rare and previously unknown primary immunodeficiencies ”

Primary immunodeficiency diseases currently consist of more than 150 mostly monogenic disorders that cause various combinations of infection, immune dysregulation, autoimmunity and malignancy. Studying these human phenotypes provides a unique insight into immune function and leads to improved care for these patients. The presentation will focus on several recent cases that have been investigated at BC Children's Hospital and the Child & Family Research Institute that have led to the discovery of previously unknown pathogenic genetic mutations and novel phenotypes, including FOXP3 and MALT1 mutations. This has led to meaningful clinical outcomes and provided biological information about the pathogenesis of inflammation, autoimmunity and malignancy. We hope that this presentation will also serve as a catalyst to form new scientific collaborations to further the study of rare pediatric hematological disorders in Vancouver.

