Ackert Hall, Room 120
Wednesday, April 9, 2025
4:00 P.M.

Coffee and Cookies
Chalmers Hall, Room 168
3:45 P.M.

Biochemistry & Molecular Biophysics Seminar

From Transcription Factors to Glucose Immunometabolism: Leveraging Monogenic Diseases to Learn Human Immunology

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In the early 70s, David Vetter was born in Texas without circulating T and NK cells. He was immediately placed in a sterile environment, where he remained for his entire life since his immune system was not equipped to deal with the outside world. His disease became one of the best-known rare diseases by the general public, and the media coined it the "Bubble boy disease". Five decades later, David's disease is one type of a broader category of diseases called Inborn Errors of Immunity (IEI). IEIs are monogenic, caused by mutations in a single gene in each individual, and these mutations alter the development or function of the immune system. IEIs are clinically heterogeneous: they may confer susceptibility to severe infection, autoimmunity, autoinflammation, allergy, atopy, or cancer, depending on the gene mutated in each patient. Identifying the disease causal mutation for each patient is critical since this knowledge provides a basis for personalized therapies that can be life-saving. Unfortunately, despite IEI affecting an estimated 1 out of every 500 individuals, the genetic etiology in most patients remains unknown. From the basic science perspective, IEI can be instrumental in understanding human immune mechanisms. Studying these patients, often completely deficient for a specific immune-modulating protein, provides a unique opportunity to learn about critical mechanisms of human immunology without relying on animal models. Unfortunately, in a sizeable fraction of patients, the molecular consequences of their mutations are unknown. In this seminar, I will present a new IEI caused by mutations in a transcription factor and talk about how a founder mutation in a metabolic enzyme holds the key to understanding the glucose requirements of the human immune response.