ASHBi SEMINAR

Inborn errors of immunity in a Finnish cohort – From CVID to a novel combined immunodeficiency caused by *SIT1* deficiency

Lecturer: Juha Grönholm, MD, Ph.D.
Principle Investigator,
Translational Immunology Research Program, University of Helsinki

Date Friday, 21 March 2025

Time 11:00 – 12:00 [JST]

Venue Conference Room Onsite Only*
B1F, Faculty of Medicine Bldg. B

Register here

Abstract

Dr. Juha Grönholm is a pediatrician and immunology researcher. He received his PhD from the University of Tampere, Finland, in 2010, focusing on the regulatory mechanisms of the JAK/STAT signaling cascade. During his postdoctoral fellowship at the National Institutes of Health, USA, he contributed to identifying a novel inborn error of immunity (IEI) caused by BACH2 haploinsufficiency (Nat. Immunol. 2017). Currently, Dr. Grönholm leads a research group at the Translational Immunology Research Program (TRIMM) at the University of Helsinki, Finland, while working as a pediatric hemato-oncology fellow at the New Children's Hospital, HUS Helsinki University Hospital. His research explores the molecular mechanisms of IEIs and the transcriptional regulation of antibody class-switch recombination in human B cells. In his lecture, Dr. Grönholm will discuss IEIs enriched in the Finnish population and present his latest findings on a novel combined immunodeficiency caused by Signaling Threshold Regulating Transmembrane Protein 1 (SIT1) deficiency. SIT1 encodes a transmembrane adaptor protein that negatively regulates T cell receptor signaling. SIT1 deficiency results in T cell hyperactivation and paradoxically impaired CD8 T cell cytotoxicity, providing new insights into immune dysregulation.

Organizer : Graduate School of Medicine
Institute for the Advanced Study of Human Biology (WPI-ASHBi), Kyoto University

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