Alberta Health Services

Alberta Children's Hospital

Please join us Tuesday, 11 June 2024

8:00 am – 9:00 am

Presentation on:

Sphingosine phosphate lyase insufficiency syndrome (SPLIS): a rare pyridoxine-responsive inborn error of sphingolipid metabolism affecting the Hutterite community

Dr. Julie Saba is a pediatric oncologist at the University of California San Francisco where she is Professor of Pediatrics and holds the John and Edna Beck Chair of Cancer Research. Dr. Saba attended The Johns Hopkins University, University of Maryland School of Medicine, and completed her pediatrics residency, hematology/oncology fellowship and PhD in Cell Biology at Duke University Medical Center. During her graduate studies, Dr. Saba identified the first sphingosine phosphate lyase gene using a genetic screen in budding yeast. She later found the mouse and human genes encoding this enzyme, called SGPL1, which is critical for metabolism of sphingolipids. In 2017, she and her colleagues discovered SPLIS—a rare inherited disorder of sphingolipid metabolism that results in steroid-resistant nephrotic syndrome, adrenal insufficiency, peripheral neuropathy and immunodeficiency. SPLIS is caused by inactivating recessive mutations in SGPL1. Dr. Saba is now focused on elucidating the pathologic basis of SPLIS and developing targeted therapies to treat SPLIS and related conditions. Her research is funded by the National Institutes of Health, the California Institute of Regenerative Medicine, UCSF's Catalyst and REAC Programs, and the Swim Across America Foundation for Cancer Research. She is the recipient of the first Eli Lilly Grand Challenge in Genetic Medicine and a Harrington Scholar/Innovator.

Alberta Children's Hospital
Conference Room 4
4th Floor

Zoom:
https://albertahealthservices.zoom.us/j/67435266808?pwd=g9RUUecPami1ljob2AnvhKoBBcyx1c.1
Meeting ID: 674 3526 6808
Passcode: 183533