DR. ALISON ELLIOTT
BC CHILDREN'S HOSPITAL RESEARCH INSTITUTE
13:30-14:00
TITLE: GENETIC COUNSELLING CONSIDERATIONS IN RARE DISEASE

Dr. Alison M. Elliott is a PhD geneticist and board-certified genetic counsellor, with expertise in rare disease, genetic and genomic counselling research. She obtained her BSc Honours at Queen's University, her MS in Genetic Counselling at the University of Cincinnati, and her PhD from the University of Manitoba. She is a tenured Associate Professor in the Department of Medical Genetics at the University of British Columbia and an Investigator at BC Children's and Women's Hospital Research Institutes. She was the Project Lead for the Genome Canada Large-Scale Applied Research Project grant "GenCOUNSEL – Optimization of Genetic counselling with implementation of genome-wide sequencing" and is currently leading a CIHR grant evaluating workforce and genetic counselling in Canada. In 2018 Alison was awarded the Canadian Association of Genetic Counsellors (CAGC) Professional Practice, Innovation and Advocacy Leadership national award. She was awarded a Killam teaching award from the Faculty of Medicine in 2025 and the Overall Excellence Distinguished Achievement Award from the Faculty of Medicine in 2025. This award recognizes outstanding contributions in the areas of research, education and service.

DR. ANNA LEHMAN
BC CHILDREN'S HOSPITAL RESEARCH INSTITUTE/VGH
14:00-14:30
TITLE: LOST IN TRANSLATION: FIFTEEN YEARS OF GENOME DECODING

Dr. Anna Lehman is a medical geneticist trained at the University of British Columbia and currently an Associate Professor at UBC. She has practiced in the areas of medical genetics and inherited metabolic diseases at BC Children's Hospital and Vancouver General Hospital. She is currently director of the Metabolic Diseases Clinic at VGH. Her research has used genomic technologies to find the causes of rare diseases. Working together in international collaborations, her research team has found the causes for more than 30 genetic disorders.

TRAINEE PRESENTATIONS:

ANA ACOSTA - BCCHRI 12:00-12:30
TITLE: IDENTIFICATION OF COMPOUND HETEROZYGOUS CYP11A1 VARIANTS VIA REANALYSIS OF CLINICAL SEQUENCING DATA

MATTISON STOJCIC - BCCHRI 12:30-13:00
TITLE: IRF4 T95R COMBINED IMMUNODEFICIENCY: GENOTYPE-SPECIFIC CONSIDERATIONS IN INBORN ERRORS OF IMMUNITY

SIMRAN SAMRA - BCCHRI 13:00-13:30
TITLE: HUMAN GERMLINE BIALLELIC LOSS-OF-FUNCTION OSMR VARIANTS CAUSE SEVERE ALLERGIC DISEASE

RareKids CAN
PEDIATRIC RARE DISEASE CLINICAL TRIALS AND TREATMENT NETWORK

BC Children's Hospital Research Institute

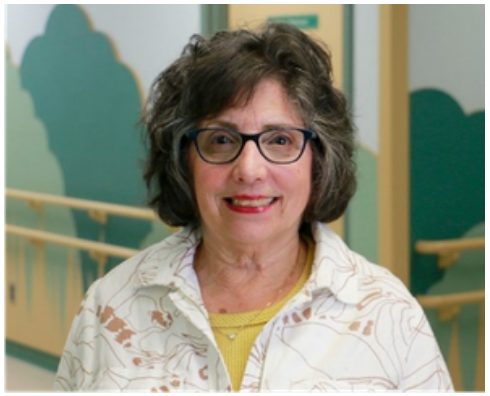
27 FEB 2026

IN HONOUR OF RARE DISEASE DAY

Rare Disease Research Symposium 2026

PROGRAM AT A GLANCE:

9:00 Refreshments available
9:30 Opening remarks
9:40 Kelly Sandhu
10:00 Dr. Cheryl Greenberg
10:30 Dr. Danielle Baribeau
11:00 Dr. Jeff Bone
12:00 Ana Acosta
12:30 Mattison Stojcic
13:00 Simran Samra
13:30 Dr. Alison Elliott
14:00 Dr. Ana Lehman
14:30 Closing remarks

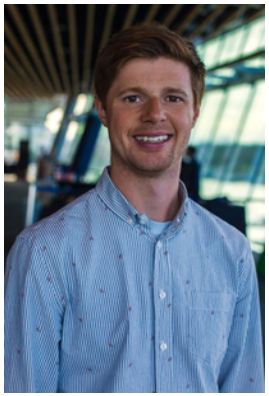


DR. CHERYL GREENBERG
CHILDREN'S HOSPITAL RESEARCH INSTITUTE OF MANITOBA
10:00-10:30
TITLE: RARE GENETIC DISORDERS: THE NEXT FRONTIER

Dr. Cheryl Rockman-Greenberg completed her medical studies as well as postgraduate specialty training in Paediatrics and Medical & Biochemical Genetics at McGill University in Montréal. She subsequently moved to Winnipeg in 1978 where she is proud to have lived ever since. Throughout her professional career at the University of Manitoba in the Departments of Pediatrics & Child Health and Biochemistry & Medical Genetics and in the Program of Genetics and Metabolism, Shared Health, she has focussed on caring for patients and their families with hereditary metabolic disorders, focused on a "bedside to bench and back to bedside approach" to further clinical discovery and new ways to treat patients.

DR, DANIELLE BARIBEAU
HOLLAND-BLOORVIEW KIDS REHABILITATION HOSPITAL
10:30-11:00
TITLE: N-OF-1 TRIALS IN ULTRA-RARE GENETIC CONDITIONS: METHODS, OPPORTUNITIES, AND EXAMPLES OF IMPACT

Dr. Danielle Baribeau is a child and adolescent psychiatrist and clinician-scientist at Holland Bloorview Kids Rehabilitation Hospital in Toronto. Her clinical interests include autism, rare genetic neurodevelopmental disorders, and psychopharmacology. Her research program is focused on clinically translating genetic advances into improved health care for children with neurodevelopmental disabilities, with a focus on innovative trial designs, ultra-rare genetic conditions, and administrative health data. She is also the lead of the Ontario Provincial Genetics Program Expert Group for genetic testing in neurodevelopmental disorders.



DR. JEFF BONE
BC CHILDREN'S HOSPITAL RESEARCH INSTITUTE
11:00-12:00
TITLE: STUDY DESIGNS FOR RARE DISEASES: BAYESIAN METHODS, EXTERNAL CONTROLS, AND TARGET TRIAL EMULATION

Jeffrey Bone is the Biostatistical Lead at BC Children's Hospital Research Institute where he manages a team of biostatisticians supporting clinical and epidemiological research, and an Adjunct Professor in the Department of Statistics at the University of British Columbia. He has worked across a wide range of areas within women's and children's health with a methodological focus on the design of randomized controlled trials and causal inference approaches to observational data. He has a strong focus on translation and presentation of statistical methods to non-technical audiences.