RÉSEAU CANADIEN DES MALADIES RARES

CANADIAN RARE DISEASE NETWORK

Canadian Rare Disease Innovation Showcase

AGENDA

The Westin Calgary Downtown (320 4 Ave SW) & Virtual (Zoom)

Monday, April 28, 2025

4:00 – 8:15 PM MST

Program

Location: Westin Downtown Calgary in the meeting space Britannia & Belaire

Time (MST)	Session Details
4:00 – 4:30 PM	**Registration & Welcome (In-Person Only)** Kickstart the event with light refreshments and connect with fellow attendees.
4:30 – 4:45 PM	**Opening Remarks** **Speakers:** Svenja Espenhahn (CRDN) & Francois Bernier (University of Calgary)
4:45 – 5:00 PM	**A Parent's Perspective on Caring for a Child with an Ultra-Rare Disease** **Speaker:** Ashley Davis
5:00 – 5:45 PM	**Diagnostic Advances – Genome and Beyond** The journey to a rare disease diagnosis is often long and complex. This session will explore how innovations in genomics, multi-omics, AI-driven tools, and robust patient registries are revolutionizing the diagnostic landscape. **Speakers**: Alexandre White-Brown (Children's Hospital of Eastern Ontario), Benedikt Hallgrimsson (University of Calgary), Lawrence Korngut (University of Calgary)
5:45 – 6:15 PM	**Networking Break with Appetizers** Enjoy appetizers and connect with attendees.

6:15 – 7:00 PM	**From Discovery to Treatment: Advancing Rare Disease Therapies** This session will dive into advancements in therapy development and precision medicine. Learn how innovative clinical trials and approaches are bringing life-changing therapies from the lab to patients. **Speakers:** Peter Kannu (University of Alberta), Eliza Philips (Alberta Children's Hospital), Eli Kinney-Lang (University of Calgary)
7:00 – 7:45 PM	**Solutions for Challenges in Rare Disease Care and Support** A rare disease diagnosis affects every aspect of life—from accessing specialized care to navigating school, work, and mental health challenges. This session will highlight real-world solutions that make a tangible difference for patients and families. Speakers will share lived experience and initiatives focused on improving care and quality of life—such as innovative family support programs, peer-led research, and tools to support smoother transitions across the lifespan. **Speakers:** Michelle Batthish (McMaster Children's Hospital), Deborah Marshall (University of Calgary) and Jessica Head (Family Engagement in Research), Rachel Martens (University of Calgary) and Genevieve Currie (Mount Royal University)
7:45 – 8:05 PM	**Reflection Panel: The Future of Rare Disease Innovation** A reflective and forward-looking discussion on strengthening partnerships and improving patient-centered care and innovations. **Panelists:** Genevieve Currie (Mount Royal University), Ashish Marwaha (Alberta Children's Hospital, University of Calgary), and Ian Stedman (York University)
8:05 – 8:15 PM	**Closing Remarks** **Speaker**: Francois Bernier (University of Calgary)
8:15 – 9:00 PM	**Networking Session (In-Person Only)** Continue conversations and build connections.

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We would like to extend our heartfelt gratitude to our sponsors for their generous support and commitment to making this event possible

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Help us build on our momentum by raising awareness and sharing knowledge to improve rare disease research and care.

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#RareLivesSharedStrength #Canada4Rare

SCAN ME

MEET OUR SPEAKERS AND PANELISTS

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Meet Our Moderator Jordan Witzel

Jordan Witzel

Jordan Witzel is a former weather broadcaster known for his engaging on-air presence and creative approach to delivering forecasts. After transitioning from radio to television, he became a household name through his work with Global News Winnipeg and Calgary, where he gained recognition for his humorous and memorable weather segments. Beyond broadcasting, Witzel has worked as a weather consultant, advising private companies and film productions on weather conditions in Western Canada. His experience includes involvement in major film productions such as Interstellar, Ghostbusters: Afterlife, and The Revenant. Motivated by a desire to make a more direct impact, Witzel is now pursuing a career in medicine at the University of Calgary, aiming to combine his communication skills and compassion to support patient care. His transition from media to medicine reflects his deep commitment to community service and helping others on a more personal level.

Meet Our Parent Perspective Ashley Davis

Ashley Davis

Ashley Davis' career aspirations began as a focus in pediatric medicine. After a few exhausting semesters at U of A, she took a break as most 20 something's do. This break led her to the hospitality industry; it quickly became evident she had a knack and a deep passion for restaurants and food service. During the following decade, Ashley would open 30+ restaurants across western canada; then to focus her efforts into produce procurement from the western united states into the western provinces.

Exciting career aside, Ashley achieved the most exciting thing of all in 2021, parenthood. Shortly after their son Jake was born, Ashley and her Husband Mat learned of Jake's ultra rare diagnosis, of ERCC-1 deficiency - an inability to repair DNA. Ashley's life path has come full circle now leaving behind beloved food service and once again focusing on pediatrics; specifically rare diseases and advocacy for her son, as well as the Alberta Children's Hospital.

Meet the Speakers-
Diagnostic Advances- Genome and Beyond

Alexandre White-Brown

Alexandre White-Brown is a clinical Genetic Counsellor with the Prenatal Diagnosis Team at the Children's Hospital of Eastern Ontario. He also holds the position of Research Associate at the CHEO Research Institute, focusing on the use of novel technologies and electronic medical records for the identification and diagnosis of rare genetic diseases. Alexandre additionally volunteers on the Medical Advisory Board of the DLG4-SHINE Foundation, helping to build a non-profit organization to advocate for individuals and families with DLG4-Related Synaptopathy.

Benedikt Hallgrímsson

Dr. Benedikt Hallgrímsson is an internationally recognized biological anthropologist and evolutionary biologist whose pioneering research integrates developmental genetics, bioinformatics, 3D imaging, and morphometrics to study phenotypic variation. Dr. Hallgrímsson leads a research program that explores the mechanisms driving anatomical variation, structural birth defects such as craniofacial anomalies, and the connections between evolution and development.

Lawrence Korngut

Dr. Lawrence Korngut is a neuromuscular neurologist, researcher, and entrepreneur dedicated to improving patient outcomes through clinical research, innovation, and data-driven healthcare solutions. As a Professor at the University of Calgary's Cumming School of Medicine and a member of the Hotchkiss Brain Institute (HBI), he specializes in diagnosing and treating neuromuscular conditions, performing electromyographic (EMG) studies, and advancing real-world health data collection to drive better care for patients with diseases like ALS.

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Meet the Speakers-
From Discovery to Treatment: Advancing Rare Disease Therapies

Peter Kannu

Dr. Peter Kannu is a Paediatrician and Clinical Geneticist specializing in diseases affecting the skin and bones. His research focuses on understanding biologic pathways that influence skeletal growth and maintenance, with the goal of improving human skeletal health. At the University of Alberta, he leads efforts to identify novel disease-causing genes in inherited bone and joint disorders. His work includes studying osteofibrous dysplasia, primordial dwarfism, and scoliosis genomics.

Eliza Ani Phillips

Dr. Eliza Ani Phillips is a dedicated medical geneticist at Alberta Children's Hospital in Calgary, Alberta. She completed her medical degree at the University of Calgary in 2017, followed by a residency in Medical Genetics and Genomics. Dr. Phillips was drawn to medicine by its unique ability to provide care and support during some of the most difficult moments in a patient's life. Her passion for genetics stems from the challenge and fulfillment of diagnosing rare diseases—an area where answers can bring immense relief and clarity to patients and families.

Eli Kinney-Lang

Dr. Eli Kinney-Lang is a biomedical engineer dedicated to advancing pediatric brain-computer interface (BCI) research. As the technical lead for BCI4Kids within the Department of Biomedical Engineering at the University of Calgary, he collaborates with researchers, clinicians, industry partners, and families to develop innovative BCI solutions that enhance communication and mobility for children with disabilities.

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Meet the Speakers-
Solutions for Challenges in Rare Disease Care and Support

Michelle Batthish

Dr. Michelle Batthish is a pediatric rheumatologist at McMaster Children's Hospital and Associate Professor of Pediatrics at McMaster University. She is also the physician lead of Ambulatory Care at McMaster Children's Hospital and Vice-President, Pediatrics at the Ontario Rheumatology Association. Her clinical and research interests are in healthcare transition in patients with chronic rheumatic disease and quality improvement in pediatric rheumatology.

Deborah Marshall

Dr. Deborah Marshall is a Professor and Svare Chair in Health Economics, Value and Impact in Community Health Sciences at the University of Calgary. She is also the Scientific Director for Research and Partnership Evaluation and Impact at Alberta Children's Hospital Research Institute, and One Child Every Child, Value, Impact & Knowledge Mobilization Co-Facilitator. She leads the health economics, socioeconomic benefits, patient preferences and patient engagement activities for several national and international research programmes in precision health and patient- oriented research.

Jessica Head

Jessica Head is a mother of three, rare disease advocate, and current trainee in the Patient and Community Engagement Research (PaCER) program at the University of Calgary. Her journey into research and advocacy began with her daughter Ava's diagnosis of Kabuki syndrome, a rare genetic disorder that reshaped her understanding of healthcare, community, and the transformative power of lived experience. With a professional background as a chef and holistic nutritionist, Jess brings a unique perspective to patient-oriented research. In addition to her work with PaCER, Jess recently joined Rare Kids-CAN as a parent partner.

Meet the Speakers-
Solutions for Challenges in Rare Disease Care and Support

Rachel Martens

Rachel Martens has been a part of the disability community in varying capacities for 20+ years. She is the bereaved parent to a young man born with medically complex disabilities who passed away in 2020. She is also someone with lived experience of disability herself as an ambulatory wheelchair user with a neurological condition. Rachel currently serves as a Knowledge Broker with the Azrieli Accelerator and a Research Engagement Strategist with CanChild, where she supports family-researcher partnerships and knowledge mobilization.

Genevieve Currie

Genevieve Currie is a parent of a child with a rare medically complex disease. She is a parent advocate, registered nurse, and researcher in rare disease, medical complexity and disability. She is an Associate Professor in the School of Nursing and Midwifery at Mount Royal University, Calgary, Alberta. She sits on several national committees around rare disease and medical complexity including as Co-Chair of the Children with Medical Complexity Network with Children's Healthcare Canada. She also works with CanChild Center for Disability Research to deliver Family Engagement in Research courses for caregivers, researchers and clinicians.

Meet the Reflection Panelists

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Ashish Marwaha

Ashish (Ash) Marwaha is an early career clinician scientist and clinical medical geneticist focusing on complex immune dysregulation disorders. He has used a collaborative approach to build an integrated research care pathway that aims to diagnose the cause of rare diseases and lead to improved targeted treatment. His research expertise includes whole genome sequencing, RNA sequencing, polygenic risk score analysis, epigenomics and machine learning integration of multi-omic data. He ultimately wants to use his extensive clinical trial experience to implement precision therapy for this patient population.

Ian Stedman

Ian Stedman is an Associate Professor in the School of Public Policy and Administration at York University and is also CRDN's "Care, Support and Empowerment" Pillar Lead. Diagnosed with Muckle Wells Syndrome in 2012, Ian is a rare disease patient, parent, and advocate. Having formerly served as a board member of the Canadian Organization for Rare Disorders (CORD), Ian now serves as the board chair for the Canadian Autoinflammatory Network / Réseau Auto-inflammatoire Canadien, and also as a legal member of the research ethics board at SickKids Hospital in Toronto.