Delivering on Canada's Rare Disease Investment

Rare Disease Day Conference

March 28 – 29, 2023

Ottawa Marriott

100 Kent Street, Ottawa

*Virtual option available

Canadian Organization for Rare Disorders

Tuesday, March 28, 2023 **Rare Disease Infrastructure Essential to Optimal Drug Benefit**	
8:30 AM - 9:00 AM	**Registration & Continental Breakfast** Victoria Ballroom (2nd Floor)
9:00 AM - 9:25 AM	**Welcome and Opening Address** Durhane Wong-Rieger, CORD **Keynote Speaker: Canada's Commitment to Rare Disease Patients and Families** • Pamela Aung-Thin, Assistant Deputy Minister, Health Products and Food Branch, Health Canada **Keynote Panel: A Vision for Rare Disease in Canada** • Alex Munter, Children's Hospital of Eastern Ontario (5 min) • Brigitte Nolet, Roche (5 min)
9:25 AM - 9:35 AM	**Challenges to Address Rare Patient Experience in Canada Feedback from patients and families to CORD survey on navigating the healthcare system: what is working, not working, and needs to be changed. What should we do with $1 billion to improve lives of patients and families with rare disease?** • Hyejin Park, Ipsos (10 min)
9:35 AM - 9:45 AM	**Development of Canada's Rare Disease Network: Centres of Expertise** **Setting the Stage** • Daniel MacDonald, Health Canada (5 min) • Ian Stedman, York University (5 min)
9:45 AM - 10:00 AM	**Interactive Discussion with Expert Panel and Participants** Three brief presentations will set the stage for implementing Canada's Rare Disease Network of Centres of Expertise as fundamental to optimal patient care and essential to optimal drug access and value. **Presenters:** • Durhane Wong-Rieger, CORD • Leanne Ward, CHEO • François Bernier, University of Calgary

	• **Centres of Expertise:** How can the Canadian Rare Disease Network assure that every patient and family affected by rare disease gets timely diagnosis, access to specialists, and integrated comprehensive care? How does the CRDN function within a WHO Global Rare Disease Network? Durhane Wong-Rieger, CORD (5 min) • **Specialized Network:** What is an example of a specialized network that serves a specific disease cluster, how it is organized, who are involved, and what services and supports are offered? What additional resources are needed to expand and sustain these specialized networks within a Canadian Rare Disease Network? Leanne Ward, CHEO (5 min) • **Clinical Reference Centres of Expertise:** How can existing children's healthcare centres be organized to serve as the "backbone" of national Rare Disease Network and also to serve as the "hub" for support to local health providers? François Bernier, University of Calgary (5 min) .
10:00 AM - 10:45 AM	**Facilitated discussion: How to Optimize Rare Disease Drug Strategy** **Leads:** • Daniel MacDonald, Health Canada • Durhane Wong-Rieger, CORD 1. The proposed national plan identifies key components, objectives and four "strategic" pillars; are these sufficiently comprehensive; if not, what is missing? 2. How does announced Rare Disease Drug Plan align with CORD's Rare Disease Strategy, which is based on networked Centres of Expertise, Value-based Managed Access to Therapies, and Investment in Research? 3. What are the opportunities and risks inherent in the proposed national Drug Plan to advance diagnosis, care and treatment for patients and families? How can we leverage the opportunities and neutralize or minimize the risks?

	4. What are the success factors to optimize benefits to patients? Who are other key players that need to be at the table to help design the plan and implement the strategy? What other investments and supportive mechanisms are needed, and how do we secure these?
10:45 AM - 11:00 AM	**Refreshment Break**
11:00 AM - 12:00 PM	**Case Studies with Expert Panel** The next are very brief overviews of case examples drawn from "real life" Canadian patient journeys that demonstrate the challenges, as well as facilitators, in the healthcare system. **Expert Panelists:** François Bolduc, University of Alberta; Kym Boycott, CHEO; Pranesh Chakraboty, CHEO; Riyad Elbard, Thalassemia Foundation of Canada; Cathy Evanochko, Tuberous Sclerosis Canada; Kasha Mitton, Defeat Duchenne Canada; Ian Stedman, York University; Kim Steele, Cystic Fibrosis Canada; Sunita Venkateswaran, London Health Sciences Centre; Leanne Ward, CHEO Open discussion with panelists and all audience 1. **Muckle Wells/CAPS:** "Diagnosis became a family affair." *After over six decades of searching, this woman's entire family finally received its rare disease diagnosis when her granddaughter was born with some familiar symptoms. What helped was that a new treatment meant specialists were now alerted to the disease spectrum.* 2. **Fabry Disease:** "Can multi-generation screening unlock the family tree?" *My father died at 29 years when I was four, and his brothers never made it to the age of 30; the women lived longer, to about 50 years. No one knew what killing them. Until at 52 years, I presented with heart problems and my cardiologist said, "Let's do genetic testing." I got a name for the disease, a new treatment, and a new chance for life."* 3. **Aplastic anemia and PNH:** 'Lightning can strike twice."

At 26 years old, I thought I would finally put my struggles behind me when I found a donor match for a life-changing bone marrow transplant. Until just before the procedure, they found I also had paroxysmal nocturnal hemoglobinuria (PNH), an ultra-rare condition that killed off healthy cells, including my new bone marrow cells.

4. **Myasthenia gravis:** "It's all in your head."
   I was in my late 20s when I started to have difficulty speaking. Every doctor said it was in my head. After my first child was born, things got worse but still no one believed me. What saved me was a sinus infection and my throat closed up, so I ended up in the ER, where a doctor gave me an injection and a diagnosis.
5. **Hereditary angioedema:** "Rare when remote is twice as hard."
   Because I live in rural Manitoba, I had to travel long distances to meet with allergists and immunologists. My symptoms — with swelling that would subside for a while and then come back worse than ever — didn't match with anything I had seen from years working in my local ER. Even with a diagnosis, the anxiety of an attack is always in the back of my mind.
6. **Cholangiocarcinoma:** "Delayed diagnosis made a bad cancer worse."
   I was happy and healthy. Then, within two weeks, I found out I had stage four bile duct cancer, and I was being scheduled for radiation and chemo. There are no good cancers but this is a particularly bad one.
7. **Spinal Muscular Atrophy:** Will a one-month delay make a lifetime difference?"
   It was nearly a month before our son would be diagnosed with the most severe form of SMA, a rare genetic disease that weakens and wastes muscles. If he had been born in Ontario, he would have been tested at birth with a heel prick and had immediate access to treatment before symptoms set in.

Expert Panel Discussion

Expert panels will discuss case examples of patient journey challenges to diagnosis and access to specialists to identify what is working in the current system, what are the challenges experienced, and what changes could be addressed in a Rare Disease Network model.

- What are the barriers and facilitators to timely diagnosis?

	• What are the challenges and opportunities in access to specialists? • How is access to comprehensive integrated multidisciplinary and supportive care for rare diseases working, or not?
12:00 PM - 1:00 PM	**Lunch**
1:00 PM - 1:15 PM	**Value-based Healthcare and Real-World Evidence: TOWWERS Showcase Bridging Centres of Expertise and Managed Access to Therapies** - Julie Frappier, Data 4 Actions
1:00 PM - 1:30 PM	**Alternative Pathways for Access to Rare Disease Drugs:** Brief Presentations and Facilitated Expert Panel Discussion • **Managed Access/Value-Based Drug Access Schemes: Global Perspectives:** What are different models of Managed Access Schemes and where and how are these being used in various countries to provide access to rare disease therapies? What are the benefits and risks of each approach? What are requirements for effective implementation, including risk-sharing financial arrangements, real-world evidence, and outcomes assessment. Fred Little, Pfizer Canada • **Managed Access Schemes—Canadian Experience:** What has been the Canadian experience with Managed Access Schemes? Where and how have they worked and what have been challenges to making them work? Tara Cowling, Medlior • **HTA and Managed Access:** How can health technology assessment contribute to the development of managed access plans? When and how should Canada's HTA agencies participate in MAS for rare disease drugs? Sylvie Bouchard, INESSS • **Provincial Perspectives on Value-Based Rare Drug Access:** How can a value-based approach to a rare drug access provide timely access to those who could benefit while also assuring affordability and sustainability in public drug plans? How can strategies for risk-sharing between pharma and drug plans accelerate access for patients and mitigate risk to the payers? Mitch Moneo, BC Ministry of Health **Moderators:** • Alexandra Chambers, Novartis • Bill Dempster, 3Sixty Public Affairs

	Panelists: • Fred Little, Pfizer Canada • Allison Wills, 20Sense • Tara Cowling, Medlior • Sylvie Bouchard, INESSS • Mitch Moneo, BC Ministry of Health • Gaby Bourbara, Alexion Canada • Rosalie Wyonch, CD Howe Institute • Julie Frappier, Data 4 Actions • Peter Brenders, BeiGene
2:30 PM - 2:45 PM	**Afternoon Break**
2:45 PM - 3:45 PM	**Case Studies with Expert Panels** **Addressing Challenges to Access to Treatment through Managed Access Programs** **Cases: Access to Therapy** 1. **SMA Type3:** Access beyond clinical trials *The treatment is here, it has been approved, but it's not available to me because of my age and my SMA type*. 2. **Retinal Blindness:** When there are no more cones, cannot treat *Raising awareness through advocacy is the way we get our message heard and bring about real change.* 3. **Fibrodysplasia Ossificans Progressive**: Protect against future injury *It's so tough as a teenager, when you're finally ready to be more independent and to start doing more on your own, to instead find yourself going backwards.* 4. **XLH:** Clinical trials: an adventure far from home *Once we had the diagnosis, that's when the real journey started — the around-the-clock phosphate supplements, the surgeries, the exercises, and the flights back and forth to the states to be in a clinical trial.* 5. **ALS:** Diagnosis delayed; drug denied What is it worth for one more month to hug your kid? One more week? One more day? Most Canadians with ALS are being denied access because of very restrictive criteria.

	6. **aTTP:** Rescue and prevent future crises *You never know when a life-threatening episode will occur, so even in remission, the disease remains an ongoing challenge.* 7. **LC-HAD/LC-FAOD:** Risk of bias … statistical uncertainty *I've been trying to avoid every little sickness for my whole life. The one thing I haven't been able to control, though, is the vision loss. They said no to the only effective treatment.* 8. **VHL:** "The tumours are like salt and pepper all over the cord." *With each spinal cord surgery, the scar tissue becomes more extensive…. I'm terrified they will grow and require more surgery. I can't keep doing this …* **Panel A:** Brad Alyward, CORD; Sylvie Bouchard, INESSS; Claudia Caminit, Answering TTP; Wayne Critchley, Global Public Affairs; Chad Mitchell, Associate Deputy Minister, Alberta; Beth Vanstone, CF Get Loud (former) **Panel B:** Alexandra Chambers, Novartis; Carrie Connell, Canadian FOP Network; Tara Cowling, Medlior; Jida El Hajjar, Loeys-Dietz Syndrome Foundation; Bob McLay, Sobi; Stephen Parrott, Canadian VHL Alliance **Panel C:** Martine Elias, Myeloma Canada; Cheryl Greenberg, Children's Hospital Research Institute of Manitoba; Fred Horne, former Health Minister Alberta; Fred Little, Pfizer Canada; Mitch Moneo, BC Ministry of Health; Christine Mossa, Ipsen; Susi Vander Wyk, CureSMA Canada; Rosalie Wyonch, C.D. Howe Institute **Expert Panel Discussion** Expert panels will discuss case examples of patient experiences of challenges to timely access to appropriate treatments, including innovative medicines, to identify what is working in the current system, what are the challenges experienced, and what changes could be addressed with Alternative Pathways, including Managed Access Programs • What are best practices in other jurisdictions that should be considered for Canada? • Clinical trials for rare/ultra-rare disease drugs are often small, short, and single-arm. Outcomes data may rely on surrogate

	measures, including biomarkers, and short-term patient-reported outcome measures (PROMs) that are not easily captured by conventional, validated, quantitative Quality of Life (QoL) scales. Because of the small patient population, cost may be high on a per-patient basis but have limited budget impact. How should regulators, value assessors, and payers take all of these factors into consideration when deciding on approval and access?
3:45 PM - 4:00 PM	**Day 1 Wrap-up**
5:00 PM - 7:00 PM	**Networking Reception** – Summit (29th Floor) CORD is delighted to host an event for all conference participants to network with CORD Board of Directors, speakers, industry and fellow rare disease advocates.

Wednesday, March 29, 2023 **Rare Disease Infrastructure Essential to Optimal Drug Benefit**	
8:30 AM - 9:00 AM	**Continental Breakfast** Victoria Ballroom (2nd Floor)
9:00 AM - 9:15 AM	**Day 1 Recap** Durhane Wong-Rieger, CORD
9:15 AM - 10:45 AM	**Canadian Rare Disease Research in Drug Development, Trials, and Outcomes**
	Interactive Discussion with Expert Panel and Participants Creating Canada's Rare Disease Research Network Canada boasts world-class researchers engaging in leading-edge innovative research programs focused on rare disease and precision drug development, including cell and gene therapy. We are also experiencing renewed and expanded investment by pharmaceutical companies in Canadian-based new drug development, clinical trials, and patient support programs for rare diseases. Finally, public and private investments are creating new initiatives and opportunities to partner with Canadian rare disease patients and patient organizations to ensure Canadians benefit from new knowledge and new technologies. Experts will discuss research initiatives and what these mean to rare disease patients and families and beyond. They will also discuss the benefits of creating a Canadian Rare Disease Research Network and the requisite requirements. **Presenters, Panelists and Group Leads:** Étienne Richer, CIHR, Institute of Genetics; Carrie McElroy, Sanofi Canada/RAREi; Thierry Lacaze, MICYRN; Risini Weeratna, National Research Canada; Stéphanie Michaud, BioCanRx; Karen Dewar, Genome Canada; Susan Marlin, Clinical Trials Ontario, Brian Ballios, Toronto Western Hospital; François Bolduc, University of Alberta; Conor Douglas, York University **Presentations** (10 min each)

	• **Social Pharmaceutical Innovation:** Made-in and for Canada initiative for research and development of sustainable, accessible, and affordable pharmaceutical innovations to benefit patients and families, in Canada and beyond - Conor Douglas, York University • **Canadian Institutes of Health Research:** What are current funded research projects that are focused on or have application to rare disease? What are future potential opportunities for research funding? What programs exist for training and supporting patients to engage in CIHR-funded initiatives? Étienne Richer, CIHR, Institute of Genetics • **National Research Canada:** What are initiatives led by, funded by, or supported by NRC that have benefit and relevance to rare disease patients in Canada, and beyond? What are specific aspects of the Canadian scientific, health and social ecosystems that make Canadian an effective and competitive environment for rare disease, precision, and targeted research and development? Risini Weeratna, National Research Canada • **Genome Canada:** How has Genome Canada been active in the funding and support of research initiatives that benefit and/or support rare disease researchers, clinicians, and policy makers with direct and relevant benefits for patients and families? Karen Dewar, Genome Canada • **MICYRN:** How does MICYRN function to attract, coordinate, stimulate, and support rare disease research and product development? What is the vision of MICYRN in the proposed Rare Disease Network and Rare Disease Drug Strategy? Thierry Lacaze, MICYRN • **Clinical Trials Ontario:** What are the initiatives and capacities at CTO that directly and indirectly support researchers and clinicians in the recruitment and implementation of clinical trials and related activities, with direct engagement of and benefit to rare disease patients and families? Susan Marlin, Clinical Trials Ontario • **UHN Clinical/Research Project on Adult Retinal Disease:** Unique pilot project to bridge gap for adult patients with retinal disease. Brian Ballios, Toronto Western Hospital

	• **AI and Rare Disease Research:** Applications of Artificial Intelligence (AI) to understanding and advancing rare disease research with focus on Fragile X and autism. François Bolduc, University of Alberta • **RAREi (Canadian Forum for Rare Disease Innovators):** informal network of research-based bio pharmaceutical innovators committed to monitoring, responding and shaping policy issues in the Canadian rare disease environment. Carrie McElroy, Sanofi
10:45 AM - 11:00 AM	**Refreshment Break**
11:00 AM - 12:00 PM	**Expert Panel Discussion: Steps Toward Creating Canada's Rare Disease Research Network** In what ways might a Canadian Rare Disease Research Network stimulate, support, and deliver on the following issues: • R&D on Innovative Therapies • Patient Outcomes Research • Understanding Rare Diseases and Patient Histories
12:00 PM - 1:00 PM	**Lunch**
1:00 PM - 3:00 PM	**Critical Success Factors**? Improve research on rare diseases
1:00 PM - 1:40 PM	**Patient Data Platforms** - Brad Milson, IQVIA To support an effective Rare Disease Drug Strategy based on the three pillars of Centres of Expertise, Managed Therapeutic Access Schemes, and Research, an integrated data infrastructure is essential This would have the following objectives: (1) Integrate data from multiple stakeholders; (2) Expedite diagnosis and treatment; (3) Improve research on rare diseases; (4) Enable cost-sharing with multiple players; The infrastructure collects and links data flows from multiple sources and users sustainably via a standardized data structure and content, with the following core components: (1) Multifunction patient portal; (2) Electronic health records; (3) Patient registries, especially for clinical trials; (4) real-world data; (5) Patient profiles of different patient groups;

	(6) Standardize structure and language to facilitate a data sharing across domains and efficient data management and analysis.
1:40 PM - 2:20 PM	**Economics of Returning Value for Investment** - Lindy Forte, Eversana Rare diseases are often severe, chronic, progressively debilitating and life-threatening conditions with psychological and physical effects that seriously compromise quality of life. It is important to the understand the economic, social and quality of life impacts on individuals, families, and society, including medical and non-medical costs as well as loss of productivity, informal support, and systematic inefficiencies in diagnosis, care and treatment, with a goal of establishing a potential return on investment for a properly designed and implemented integrated comprehensive rare disease system.
2:20 PM - 3:00 PM	**Genomic Sequencing: Equitable and Accessible for Clinical Diagnosis** - Magda Price, CHEO An update on the Genome Canada-funded AllforOne Health Data Ecosystem will be shared. This national data sharing initiative for rare disease is proposed to support high quality clinical genomic testing as well as make some of this data available for research. The AllforOne Clinical Network will enable comprehensive variant sharing between Canadian diagnostic laboratories for improvement of clinical genome-wide sequencing. AllforOne Connect will be a centralized registry to connect individuals with a rare disease to research studies they may be eligible for. This initiative will ultimately improve the health and wellness of Canadians with rare disease, be one an innovative contribution to rare disease research in Canada, and lay the foundation for precision health in Canada.
3:00 PM - 3:30 PM	**Next Steps**

Conference Sponsors

The Canadian Organization for Rare Disorders acknowledges the contribution of all our Corporate Partners to improving the lives of patients and families with rare disorders.

We are especially grateful to the following that have supported this conference.

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