



Canadian Organization  
for Rare Disorders

# Rare Disease Day 2024 Summit

Celebrate with  
CORD on Rare  
Disease Day!



**RARE DISEASE DAY®**

**February 28 — 29, 2024**

**Ottawa Marriott Hotel, 100 Kent Street, Ottawa, Ontario**

(\*Virtual option will be available for those who wish to participate remotely)

# Rare Disease Day 2024 Summit

## February 28 — 29, 2024

Ottawa Marriott Hotel, 100 Kent Street, Ottawa, ON

### Join CORD in Celebrating Canada's Most Impactful Rare Disease Day Ever!

- A date so rare it only occurs "1 in 1,741" days, which is altogether fitting for diseases so rare they occur in fewer than "1 in 2,000" persons.
- Rare Disease Day 2024 is a year of "1<sup>st</sup> 's". It marks the 1<sup>st</sup> Anniversary of the announcement of Canada's 1<sup>st</sup> National Rare Disease Drug Strategy with allocation of \$1.5 billion over three years.
- As importantly, we celebrate the launch of Canada's 1<sup>st</sup> Ever Rare Disease Network, uniting patients, professionals, policy makers, payers, public, and pharmaceutical developers under a shared vision and coordinated action.
- This is the 1<sup>st</sup> year that patients could have access to an unprecedented number of rare disease drugs, approved by Health Canada and recommended by the HTA bodies. Among these are drugs with some life-saving "1<sup>st</sup> 's"! They are the 1<sup>st</sup> effective treatment for a disease, yield significantly better outcomes than current therapies, result in lower risks or fewer adverse effects, are based on a novel mechanism of action, can be precisely targeted to a patient profile, and/or have long-lasting effects, tantamount to 1<sup>st</sup> cure.
- Hot flash! Canada was the 1<sup>st</sup> country worldwide to approve two of these therapies, a very rare happening.
- Join CORD and our partners in the rare disease community not only to celebrate these "firsts" but importantly to collaborate on putting these into actions that will: generate better outcomes for patients and families. That is, actions that will reduce hospitalizations, return people to work and school, reduce disability, improve mental health, and empower people to do more of whatever it is they wish to do.

After several years' hiatus, CORD is bringing back our Rare Hero Awards. This very special Rare Disease Day we will be honouring very special individuals who have contributed outstanding service to the rare disease community over many years.

Join CORD as we honour these very special heroes. Recognition at Conference and presentation at Reception.

### **Rare Hero Awards 2024**

**Healthcare Leadership Award:**

**Susanne (Susa) Benseler**, Director, Alberta Children's Hospital Research Institute

**Public Sector Leadership Award:**

**Suzanne McGurn**, President and CEO: CADTH

**Patient Group Leadership Award:**

**Regroupement québécois des maladies orphelines (RQMO)**

**Healthcare Media Excellence:**

**André Picard**, Health Columnist, Globe and Mail

**Patient Rights Champion:**

**André Marin**, Former Ontario Ombudsman, Law Professional

### **NOT Ready to Celebrate**

Today, nearly one year later after the federal government announced \$1.5 billion to implement Canada's Rare Disease Drug Strategy, we have not heard of a single patient who has received a single drug through the Drug Strategy. Instead, hundreds of patients on provincial drug plans who have been denied access to lifesaving and life-altering drugs for months and years are still waiting. Some of these are priority drugs, the first effective treatment for a serious rare condition; others are breakthrough therapies offering significant improvements over existing therapies. Already approved by Health Canada, recommended by CADTH, prescribed by their doctors, and available to patients in other countries but still not in Canada.

## **The Right to be Impatient.**

*In 2005, Ontario Ombudsman André Marin issued a clarion call on behalf of a rare disease community that was still struggling to make its voice heard. The issue was the Ontario government's delay in implementing newborn screening. Ombudsman Marin articulated and legitimized our patient "right to be impatient."*

*He unflinchingly denounced the "...consequence of politicians and bureaucrats failing to sense the urgency and ... to remedy the situation peremptorily..."*

Fast forward to 2023, days after Rare Disease Day. The federal government announced \$1.5 billion Rare Disease Drug Strategy to make good on a 2019 promise to "...increase access to promising and effective drugs for rare diseases..." starting with a "small set that would be cost-shared and covered in a consistent way across Canada." About \$1.4 billion would be available for drug funding through federal-provincial bilateral agreements.

CORD and the entire rare disease community cheered and proposed an initial list of drugs, most of which are the first and only effective therapy for a specific rare condition. These drugs save or extend patient lives. They reduce disability, the risk of catastrophic harm or the risk of life-threatening surgery.

Approved and recommended, many of these drugs are now stalled in bureaucratic morass: in price negotiations with the panCanadian Drug Alliance, in budget impact assessments with provincial Health Ministries, or with Drug Plan Managers who have no more monies to allocate.

*Back in 2005, Ombudsman Marin wrote, "...there are ongoing operating and maintenance expenses as well as the treatment costs that successful screening requires. Still, there is every reason to believe that these costs will be more than offset by the savings on treatment and support that would otherwise be required for those who are needlessly harmed by not testing and acting."*

That same "return on investment" argument can be made for the therapies that CORD is recommending as the first drugs for immediate access through the Rare Disease Drug Strategy.

*But it was Ombudsman Marin's next sentiment which has proven even more prescient in challenging our current lack of action, "But any long-term financial benefits that will be realized pale by comparison to the human dimension. ...the*

*Government's plan and its newfound commitment to make newborn testing as good as it can be will save lives and prevent or reduce suffering." Today, for "newborn screening", read "access to therapy." Patients have the Right to be Impatient.*

### **How to Make the Most of \$1.5 billion? Invest it, One Drug at a time.**

We cannot wait for negotiations to deliver a master plan to spend \$1.4 billion in bilateral agreements. We can and must act to make urgently needed drugs immediately available. CORD has proposed an implementation plan by which rare disease therapies that are approved and recommended can be started immediately with a defined cohort of patients along with a plan for on-going monitoring, data collection, and assessment of benefit and potential harm.

Who are the patients in urgent need of approved therapies?

- Patients with HoFH, a rare form of very high cholesterol that manageable with statins and transfusion, leading to cardiovascular disease and premature death.
- Children with NF1, large tumours growing next to the nerves that cannot be surgically removed.
- Patients with FOP whose muscles and tendons turn to bone creating a second skeleton and restricting movement and breathing.
- Patients with oHCN whose thickened heart muscles restrict blood flow leading to heart failure, stroke, arrhythmias, and sudden cardiac death.
- Adults with VHL with proliferating noncancerous tumours in kidney, brain, or pancreas that cannot be surgically removed.
- Adults with SMA whose symptoms are later onset and thereby excluded from drug therapy, resulting in muscle weakness, fatigue, limited mobility, and difficulty breathing.
- Adults with late-onset Pompe's Disease denied access to treatment.
- Patients with Myasthenia Gravis waiting while pricing negotiations are taking place.

As a start to fulfilling the 2023 vision, CORD is recommending immediate implementation of pathways for access to these therapies. These are not

experiment or novel approaches but are based on effective managed access schemes being used in Canada and elsewhere. They will serve as learning models and “proof of concepts” for new and emerging rare disease drugs.

By the end of 2024, we will have implemented or have in development all the components of a patient-centred, evidence-based, cost effective and sustainable Rare Disease Drug Program.

These include:

- Canadian Rare Disease Network, bringing together all healthcare providers, researchers, and patient groups across Canada to coordinate on diagnosis, care and treatment pathways, and data management.
- Newborn screening program with national principles, guidelines, and best practices as well as recommendations and provisions for enrollment in patient registries, data sharing, referral, and access to care and treatment.
- Clinical trial network
- Patient registries and platforms for real world data collection
- Care management pathways
- Rare disease patient organizations and community support

Healthcare professionals, researchers, and patient organizations are all doing their part. We need the federal and provincial governments to do their jobs: provide patients with immediate access to new and emerging treatments.

***Ombudsman Marin’s summary opinion in 2005 is apropos and worth repeating:***

*First, while we all want fiscal responsibility, decisions based on what I have called “line-item accounting” have to be avoided. Government responsibilities to citizens are owed at large, not in isolation by divisions of government. ... The right thing to do ... is to discharge those responsibilities faithfully and with commitment, while trying to achieve reorganization. It is never appropriate to abdicate those responsibilities while waiting for reorganization to occur. True in 2005. True in 2024.*

# Rare Disease Day 2024 Summit

## AGENDA (Draft)

### Wednesday February 28, 2024

#### 8:30 am – 9:00 am

Registration and Continental Breakfast

Ottawa Marriott Hotel – Victoria North Ballroom (2<sup>nd</sup> Floor)

#### 9:00 am – 9:15 am

##### Welcome and Setting the Context

- Cathy Evanochko, CORD (Land Acknowledgment)
- Durhane Wong-Rieger, CORD
- Alex Munter, The Children's Hospital of Eastern Ontario

#### 9:15 am – 11:00 am

##### Session 1: **NEW Canadian Drug System: Working for Rare**

###### **A. UPDATE: Implementation on the National Strategy for Drugs for Rare Diseases** (75 min)

Health Canada and delivery partners at the Canadian Agency for Drugs and Technologies in Health (CADTH), Canadian Institute for Health Information (CIHI), and Canadian Institutes of Health Research (CIHR) will present an update on the progress of the various elements of the National Strategy for Drugs for Rare Diseases since its launch in March 2023. The session will be moderated by Health Canada and include presentations followed by time for an audience question and answer session.

Moderator: Daniel MacDonald, Health Canada

- Heather Logan, CADTH
- Tracy Johnson, CIHI
- Étienne Richer, CIHR-Institute of Genetics

###### **B. Impacts on Rare Disease Drugs** (30 min)

- **Proposed** PMPRB Regulatory Changes and Implications for Rare Diseases

Wayne Critchley, Global Public Affairs

- **NEW Quebec Rules: Implications for Rare (INESSS)**

Sylvie Bouchard, INESSS

- **NEW** and Emerging Therapies

Bob McLay, Sobi

**11:00 am – 11:15 am**

**Break**

**11:15 am – 12:30 pm**

**Session 2: How to Make the Most of \$1.5 billion? Invest it, One Drug at a time.**

### **A. Canadian Pathway to Rare Disease Drug Access**

Moderator: Bill Dempster, 3Sixty Public Affairs

Multistakeholder Expert Panel 1:

Sherry O'Quinn, Morse Consulting; Lindy Forte, Eversana; Alexandra Chambers, Bayer; Joan Paulin, PHA Canada; Bonnie Kam, Janssen; Christian Dong, Pfizer; Farah Jivraj, Biogen; Rute Fernandes, Takeda; Declan Hamil, IMC

### **1. Bumpy Road to Access: Navigating the speed bumps, potholes, and roadblocks (Presentation to Discussion)**

- A Review of Time to Access for Rare Disease Medicines: Are we Making Progress?
- Lessons from Abroad: Options for Upgrades to Canada's Drug Access Pathway: UK, France, Germany, Italy, Australia
- What would NICE do? Progress from STA to HST to even more flexible HST
- Are we There Yet: Shorter, Faster, Barrier-Free Routes to Save Lives, Reduce Harm
- How could \$1.5 billion be invested to accelerate time to patient access?

### **2. Addressing Challenges of Innovative Rare Disease Therapies**



### **(Presentation to Discussion)**

- How do Canadian regulatory approvals and HTA recommendations compare to those of other countries, especially Europe and USA?
- How does being rare creates challenges for drug development and access.
- Why traditional development and regulatory pathways are not (well) suited to (innovative) rare disease therapies.
- HTA Revisited: Why innovative rare disease drugs fail the ICER test and what should be done.
- How does delayed and denied access discourage clinical trials and new drug submissions in Canada.
- How could \$1.5 billion be invested to improve access to innovative therapies?

**12:30 pm – 1:30 pm**

**Lunch**

**1:30 pm – 2:45 pm**

#### **B. Negotiating Access with \$1.4 Billion Stake (75 mins)**

Moderator: Durhane Wong-Rieger, CORD

Multistakeholder Expert Panel 2: Grant Perry, BeiGene; Leanne Ward, CHEO; Jida El Hajar, ALS Action Canada; Sara Ethier, CORD; Ed Dybka, Ipsen; David Page, CHS; Shari Van Vugt, Canadian XLH Network; Carrie McElroy, Sanofi; Imran Ali, BMS; Rebecca Auer, Ottawa Hospital

#### **1. Rare and Ready ... On Road to Go**

##### **HC Approved, HTA Recommended; pCPA LOI @ Drug Plans**

- Brukinsa (zanubrutinib) for Waldenström macroglobulinemia
- Abrioza (sodium phenylbutrate) for ALS

##### **HC Approved and HTA Recommended; @ pCPA negotiations**

- Sohonos (palovarotene) for fibrodysplasia ossificans progressiva (FOP)

- Evkeeza (evinacumab) for homozygous familial hypercholesterolemia (HoFH)
- Koselugo (selumetinib) for neurofibromatosis Type 1 (NF1)
- Welireg (belzutifan) for Von Hippel-Lindau (VHL) syndrome
- Beqvez (Fidanacogene Elaparvovec) gene therapy for hemophilia B
- Camzyos (mavacamten) for obstructive hypertrophic cardiomyopathy (oHCM)
- Optune device for glioblastoma multiforme (ndGBM)

### **HC Approved @ HTA Agencies**

- Vyvgart (efgartigimod alfa) for myasthenia gravis

### **HC Approved, HTA (CADTH) NOT Recommended**

- Pemazyre (pemagitinib) for bile duct cancer (INESSS recommended)
- Leqvio (inclisiran) for hypercholesterolemia and ASCVD
- Evrysdi (risdiplam) for SMA Adults
- Nexviazyme (avalglucosidase alfa) for Adult Onset Pompe Disease

## **2. Discussion of Drug Reviews:**

- For each drug, what is time for each stage of review from submission to current access status?
- For each condition, what are current treatments and unmet need?
- For each new drug, what is evidence from clinical trials and other sources about addressing unmet need? What are limitations and potential risks based on evidence? What is budget impact?
- What additional information needs to be collected through clinical trials or real-world data platforms to assure effective and safe use of funded therapies?
- What are options for risk-sharing and cost-sharing between

government funders and manufacturers? What are roles for rare disease centres, healthcare professionals, researchers, patient groups, and patients to optimize (safe, effective, cost-effective, sustainable) use?

- What is reasonable pathway forward to make drug immediately available to appropriate patients while mitigating risks and collecting additional data for long-term demonstration and evaluation?

### **3. Pathways to Patient Access**

- How have timelines for rare disease drug improved (or not) from regulatory submission to patient access?
- Based on these cases, what are factors that seemingly increase or reduce review time; what are facilitators and barriers that limit or expand criteria for access?
- Under the (emerging) status quo, what are recommendations for improving access (who gets access and how quickly)?
- What are reasonable pathways to make drug immediately available to appropriate patients while mitigating risks and collecting additional data for long-term demonstration and evaluation? How could we better use existing mechanisms like Quebec's *"patient d'exception"*, Ontario' Exceptional Access Program, or BC Exceptional Drugs for Rare Diseases program, to provide more timely access to rare disease drugs while undergoing negotiations
- How can the \$1.4 billion from the Rare Disease Drug Strategy over next two years be invested to optimize access, and what are recommendations for the next three years?

**2:45 pm – 3:00 pm**

**Break**

**3:00 pm – 3:45 pm**

### **C. New Frontiers in Canadian Rare Disease Research**

Moderator: Ian Stedman, York University

Research Investors Panel: Sapna Mahajan, Genome Canada; Christopher McMaster, CIHR; Andrew Taylor; Health Canada; Risini Weeratna, National Research Council

The Canadian government is making significant investment in research, including rare disease research, through coordination and collaboration among partners, including patients. This panel will discuss projects that are aligned to and enhance the Rare Disease Drug Strategy to benefit patients.

**3:45 pm – 4:00 pm**

**Day 1 Summary and Day 2 Actions** Durhane Wong-Rieger

**4:00 pm – 4:45 pm**

**D. Celebrating CORD Rare Heroes 2024**

**A. Introduction of Honorees:** Master of Ceremonies: Madi Vanstone

**B. Honoree Keynote Addresses: A Decade of Progress in Rare Disease in Canada and What Next**

**Healthcare Leadership Award:**

Susanne (Susa) Benseler, Director, Alberta Children's Hospital Research Institute

**Public Sector Leadership Award:**

Suzanne McGurn, President and CEO: CADTH

**Patient Group Leadership Award:**

Regroupement québécois des maladies orphelines (Gail Ouellette, RQMO)

**Healthcare Media Excellence:**

André Picard, Health Columnist, Globe and Mail

**Patient Rights Champion:**

André Marin, Former Ontario Ombudsman, Law Professional

**5:00 pm – 7:30 pm**

**Networking Cocktail Reception (29<sup>th</sup> Floor, Summit Room)**

All conference participants are invited to attend.

**Thursday, February 29, 2024**

## **Awareness and Call to Action**

**7:30 am – 9:00 am**

**Rare Disease Day Breakfast Reception** with Members of Parliament, Senators and staff. This event is by *invitation only* due to restricted space.

**9:30 am – 10:00 am**

**Ottawa City Hall Flag Raising Ceremony:** Organized by RAREsies Rule/Ollier's Disease Canada, join members of the rare disease community, CORD, CHEO, Care4Rare and others to raise awareness and celebrate Rare Disease Day at Ottawa City Hall (110 Laurier Ave W)

**9:30 am – 10:15 am**

**Rare Disease Day Press Conference:** Breakfast will be served in the Victoria North Ballroom and the press conference will be live streamed for conference attendees.

**10:15 am – 11:00 am**

### **Taking Message to Parliament Hill:**

Join CORD as we march together to raise awareness for Rare Disease Day. All are welcome to attend. Meet in the lobby of the Ottawa Marriott at 10 am where we will march from the hotel to Parliament.

**12:00 pm – 1:00 pm**

**Lunch** (Victoria North Ballroom)

**1:00 pm – 2:00 pm**

### **Launch of Canadian Rare Disease Network**

#### **1. Coordinating Rare Disease Care across Canada**

Svenja Espenhahn, University of Calgary; François Bernier, University of Calgary; Kym Boycott, CHEO; Craig Campbell, LHSC; James Dowling, HSC; Angela Genge, The Neuro; Lawrence Korngut, University of

Calgary; Thierry Lacaze, MICYRN; Deborah Marshall, University of Calgary; Jacques Michaud, CHU Sainte-Justine; Gail Ouellette, RQMO; Ian Stedman, York University; Leanne Ward, CHEO; Durhane Wong-Rieger

- Networking: Rare Disease Experts, Rare Disease Centres, Best Practices and Guidelines, Patient Organizations

**2:00 pm – 3:00 pm**

## **2. Creating Rare Disease Expanded, Accelerated, Sustainable Access**

Moderator: Durhane Wong-Rieger, CORD

Multistakeholder Expert Panel 3: Pranesh Chakraborty, CHEO; Beth Potter, University of Ottawa; Maureen Smith, INFORM RARE; Homira Osman, Muscular Dystrophy Canada; Ilayda Ulgenap, ALS Society of Canada; Alice Williams, Wilson Disease Assoc; Cory Cowan, Alexion; David Shum, Roche; Carla Chabot, Bridgepoint; Joe Farago, IMC; Ajay Singh, McKesson

- What are core building blocks for effective Managed Access Drug Plans for rare disease drugs: timely accurate diagnosis, access to optimal medicines and therapies, monitoring and real-world data collection, follow-up and reassessment?
- What is needed to provide timely diagnosis: newborn screening, genetic (and genomic) testing, comprehensive assessment, counselling, referral and follow up?
- How can a Canadian Rare Disease Network help provide all patients and families with specialist care, optimal drug treatment, multidisciplinary care (rehabilitation, mental health, educational and occupational, and financial support)?
- In what ways should the federal-provincial governments invest \$1.4 billion to create a healthcare infrastructure to optimize timely, sustainable access to new drugs for rare disease patients?

**3:00 pm – 3:30 pm**

**Day 2 Summary Discussion and Next Steps**

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